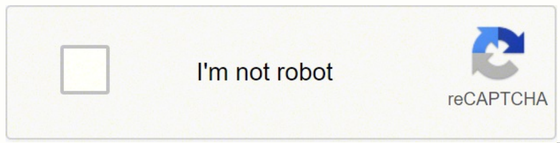

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Additional findings include hepatomegaly, wide QRS complex, and elevated blood creatine kinase levels. Retinopathy of Prematurity (ROP) VIII. It will be important for the radiologist to assess the posterior urethra during the VCUG to rule out a posterior urethral valve. Vigabatrin is frequently effective for patients with infantile spasms associated with tuberous sclerosis and is often used in combination with ACTH. Further testing is directed on the basis of clinical suspicions. Lloyd Brown To Karen, Olivia, and Penelope, for providing me with life's true education, day after day. All patients with aHUS have a higher risk of progression to ESRD than patients with STEC-HUS. Nasopharyngeal tumors typically present with epistaxis, airway obstruction, and chronic sinusitis. Acquired adrenal insufficiency 1. Type of vaccine: inactivated c. Nonclassic CAH (i.e., late-onset with mild cortisol deficiency and no mineralocorticoid involvement). Up to 60% of all infants have episodes of spitting up or vomiting not related to overfeeding or GI disease. Arteriographic embolization may be used for serious bleeding from vascular malformations. On examination, you note that the boy has brown rings at the edge of his corneas bilaterally. A complex febrile seizure lasts more than 15 minutes, has focal features, or recurs within 24 hours. Systemic or topical acyclovir may speed recovery if administered within 1–2 days of onset. Myelomeningocele is a clinical diagnosis based on physical examination at birth. Healthy children who fall behind schedule should be caught up before their fifth birthday. Electromyography shows small, polyphasic muscle potentials but normal nerve conduction velocities. Urea cycle disorders (UCDs). Administration of trimethoprim/sulfamethoxazole (TMP/SMX) for probable infection with Shigella B. Cushing disease. Treatment includes fluid resuscitation, parenteral antibiotics, and analgesia. BAER is typically performed to confirm abnormalities found on OAE. Obtain serum acetaminophen level 2–4 hours after ingestion. Carbon monoxide (see section VIII.D.6) i. Elevated antistreptolysin O (ASO) or anti-DNase B (ADB) titer may indicate a recent streptococcal infection. The timing of symptoms may provide key diagnostic information. 371 IV. Characteristic facies include synophrys (single eyebrow resulting from both eyebrows growing into one another), long eyelashes, small upturned nose, and microcephaly. Cross-references to the text appear in square brackets. Abnormal Tearing A. It typically manifests as seborrheic dermatitis of the diaper area and scalp (it mimics cradle cap) and is most common in intertriginous areas. The oculocephalic maneuver is also termed doll's eyes, and an abnormal response (when the head is turned, the eyes follow the head and continue to look straight ahead rather than drifting to midline) suggests a damaged vestibular system. Inheritance is X-linked and occurs in 1 in 5000–10,000 male births. Intravenous deferoxamine should then be continued. In addition, activated charcoal interferes with visualization during endoscopy and therefore should not be used for caustic ingestions. Red reflex assessment, in which a direct ophthalmoscope is directed at the patient's eyes from a distance of 2–3 feet (Bruckner test), is the single best screening examination for infants and children. It is an important cause of lower GI bleeding in infants and children. Germ Cell Tumors (Germinomas) Germ cell tumors are rare malignancies derived from the cellular precursors of sperm and eggs. All patients are at risk for developing hypertrophic cardiomyopathy during their lifetime. Because head size generally reflects brain size, microcephaly is always associated with a small brain. However, conjunctival cultures and Gram stain should be obtained in severe cases. Any growth on urine collected by suprapubic aspiration b. Nutrition II. Before puberty, incidence is higher in males; after puberty, incidence is higher in females. Infants with galactosemia are vulnerable to Escherichia coli sepsis, and if the condition is not diagnosed, they may die in early infancy. Initial labs usually include a blood gas showing respiratory alkalosis due to hyperventilation. Education and reassurance of parents are vital. Table 10-3 Organic Causes of Constipation Hirschsprung disease Neuroenteric dysfunction secondary to ischemia, trauma, spinal cord abnormality Medications (e.g., narcotics, sedatives) Low-fiber feeding regimens Anatomic abnormalities (e.g., stricture, adhesion, anteriorly displaced anus) Systemic disease (e.g., dehydration, celiac disease, hypothyroidism, cystic fibrosis, diabetes mellitus) Infant botulism Lead toxicity Anorexia nervosa 383 VIII. Determining the targeted MPH is important in evaluating all patients with short stature (Figure 6-1). Newborn Skin Diseases III. Hypercholesterolemia d. Immunosuppressive agents are useful in inducing and maintaining long-term remission. Fever, anorexia, nausea, and vomiting are common. Fractures usually heal without resulting deformity. While a small amount of protein is normally present in the urine, proteinuria greater than 100 mg/m2/day or an elevated urine total protein/creatinine ratio are considered pathologic. Pathologic short stature, in which height fall more than 3 SDs below the mean with abnormal growth velocity (i.e., growth velocity less than 2 inches [or 5 cm] per year), may be categorized as proportionate or disproportionate. Contraindications to immunization include the following: a. The incidence of this tumor peaks during the rapid growth spurt of adolescence. Removal of environmental allergens b. Mature teratomas often contain skin, hair, or teeth, whereas immature teratomas contain fetal- or embryonal-type structures. Wang, MD, FAAP, Associate Director, Internal Medicine–Pediatrics Residency Training Program, Cedars-Sinai Medical Center, Los Angeles, California, Assistant Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Frederick D. Transmission to affected infants is through their affected mothers in more than 90% of cases. Anticipatory guidance includes a discussion of safety issues and upcoming developmental issues. Immunophenotyping reveals that the leukemic cells have pre–B-cell phenotype and are common acute lymphocytic leukemia antigen (CALLA) negative. Other aspects of her health screening are normal, including her head circumference, language and ambulation skills, as well as her hemoglobin and lead levels. Rates of disease are highest in infants < 1 year of age, and in teens and young adults ages 16–23 years. Complications of systemic disease include meningitis, osteomyelitis, and lymphadenitis. UGI bleeding 1. 469 2. The 6-month-old infant has congenital myotonic dystrophy, an autosomal dominant disorder presenting in infancy with hypotonia, facial weakness, and feeding problems. Inheritance of VUR is most likely to be autosomal recessive. Clinical features of cholestasis 1. Repletion phase a. Delays in seeking medical attention, implausible histories, and histories that change or are inconsistent among caregivers are suspicious for abuse. Watery eye with increased tear lake (meniscus of tears upon the lower eyelid margin that spill over onto the cheek and eyelid) b. This condition is caused by a transient activation of the HPGA, resulting in transient ovarian follicular stimulation and a release of low levels of estrogen. Really good book. Topical erythromycin ointment may also be applied. Abnormalities of Maturity III. SGA: Infants with a birth weight below the 10th percentile for their gestational age are SGA. Lumbar gibbus deformity (structural, sharp-angled kyphosis resulting in a prominent "hump" on the back) in infancy that resolves once the patient starts walking. Infantile botulism is bulbar weakness and paralysis that develops in infants during the first year of life secondary to ingestion of Clostridium botulinum spores and absorption of botulinum toxin. 240 million people are chronically infected worldwide, with up to 30 million new cases every year. Causes of neonatal conjunctivitis may include infections or chemical irritation. Infection was transmitted by a maternal genital infection. Start a low-dose inhaled corticosteroid. Unwitnessed injury should not be assumed to be nursemaid's elbow, and radiographs should be obtained. Urinalysis typically reveals 3+ to 4+ protein, sometimes microscopic hematuria, and a very elevated urinary TP/CR. Trisomy 18 C. Hip A. Prognosis varies with the extent and location of disease. Progressive coarsening of facial features, large tongue, umbilical hernia, hearing loss, and cardiomyopathy 3. Chondrolysis (degeneration of articular cartilage of the hip) 3. X-linked For each patient, select the likely mode of inheritance of the disease. Erb palsy is an upper brachial plexus injury involving the C5 and C6 nerve roots. Conservative management a. This infant should be treated empirically with oral antibiotics on an outpatient basis and reevaluated within 24 hours. Loss of posture, urinary incontinence, and a postictal state do not occur. Dialysis is initiated or transplantation is considered when the glomerular filtration rate is 5–10 WBCs/HPF) and a positive nitrite or leukocyte esterase on dipstick. Management includes factor IX replacement. Within hours, the pain usually localizes to the right lower quadrant. (This syndrome is termed "shaken impact syndrome" if the child is thrown after the shaking.) Retinal hemorrhages, subdural hematomas, metaphyseal fractures, and significant brain injury are characteristic. Because this patient has grade 2 VUR, she should be placed on prophylactic antibiotics. His maintenance water requirement is 1000 mL/m2/day of body surface area. Type 4 adult onset d. Maternal systemic lupus erythematosus (SLE) would not result in hematuria in the infant; however, maternal SLE is associated with infant heart block. Diarrhea XII. The disease most commonly affects children between 4 and 8 years of age. The condition has a wide spectrum of severity. Specific Parasitic Infections XVI. Reticulocyte count may be used to differentiate among the disorders [see Figure 13-1]. Chlamydia trachomatis pneumonia generally occurs in a young infant, 1–3 months of age, and would be very unlikely in a 5month-old infant. DTaP is recommended at 2, 4, and 6 months of age with boosters at 12– 18 months and 4–6 years of age. Electromyography would be expected to show normal nerve conduction. No part of this book may be reproduced or transmitted in any form or by any means, including as photocopies or scanned-in or other electronic copies, or utilized by any information storage and retrieval system without written permission from the copyright owner, except for brief quotations embodied in critical articles and reviews. Recurrence of dislocation approaches 90% in the adolescent population. Precipitating factors. Three common types are infantile spasms, absence epilepsy of childhood, and benign rolandic epilepsy. Noncommunicating (obstructive) hydrocephalus is due to an obstruction of CSF flow. Decremental response to low-frequency (3–10 Hz) repetitive nerve stimulation 3. Allergic colitis from sensitization to protein antigens in cow's milk, soy milk, or breast milk may develop in neonates and infants. Lead a. The prognosis for children with Guillain–Barré syndrome is excellent, and complete recovery is likely. within the carotid artery, usually from plaques. Evaluation of the airway and intubation by experienced personnel in a controlled setting are necessary. Klinefelter syndrome is characterized by tall stature, gynecomastia, and a small penis and testes; Down syndrome, by characteristic facial features, endocardial cushion defects, duodenal atresia, mental retardation, single palmar creases, and a wide space between the first and second toes; Prader–Willi syndrome, by infantile hypotonia, hypogonadism, short stature, and obesity and hyperphagia later in childhood; and Williams syndrome, by a loquacious very friendly personality, supravalvular aortic stenosis, and hypercalcemia. 90. There are no tongue fasciculations. Especially important in areas where misinformation regarding the vaccine has led to decreased vaccination rates and unfortunate outbreaks of the following: 1. A magnetic resonance imaging scan of the spine is normal. Use should be carefully monitored owing to the risk of renal disease in SLE. pylori should be 388 treated with appropriate antibiotic therapy. Propionic aciduria (PA) and methylmalonic aciduria (MMA) typically present after the first few days of life with vomiting, poor feeding, hypotonia, and other neurologic symptoms, which will progress if left untreated. Platelet count c. The answer is D [VI.A, VI.B, VI.E, and VI.G.5]. Spread of infection may occur, either by contiguous spread infecting a joint or distant seeding of other organs. 21 II. Surgery may involve closed or open reduction of the hip. Headache b. Some patients may present with adrenal crisis (hypotension, hypoglycemia, severe lethargy), and this is a medical emergency! b. Large anterior and posterior fontanelles and split sutures b. Trauma IV. It is important to rule out acute life-threatening causes, such as sepsis, meningitis, or an acute metabolic disorder. The answers are A and B, respectively [Tables 9-4 and 9-5 and IV.A.7]. The review questions are followed by answers and detailed explanations for both the correct and incorrect choices. Management is supportive in the acute phase with specific therapies for chronic disease. Direct hepatocellular damage or damage to the biliary system results in derangements in the liver's synthetic ability, excretory function, or detoxification. Ewing sarcoma is the second most common malignant bone tumor. Intraoperative cholangiogram with laparotomy, or percutaneous cholangiogram, to examine the biliary tree confirms the diagnosis. 219 V. Microscopic urinalysis may also provide helpful clues. There is no evidence of a link between vaccines and autism. Table 1-1 Rules of Thumb for Expected Increase in Weight Age Birth–2 weeks (average birth weight is 3.5 kg) 2 weeks–3 months 3–6 months 6–12 months 1–2 years 2 years–puberty Expected Weight Increase Lose up to 10% of birth weight, usually in the first week of life Regain birth weight by 2 weeks of age 30 g/day 20 g/day Double birth weight by 4–6 months of age 10 g/day Triple birth weight by 12 months of age 250 g/month or 3 kg/year Quadruple birth weight by 2 years of age 2 kg/year 30 g = 1 oz body weight. Most newly diagnosed patients are started on MDI and can transition to an insulin pump, if they chose, after several months. Causes include drugs (e.g., sulfonamides, anticonvulsants, chloramphenicol), infections (e.g., human immunodeficiency virus [HIV], Epstein– Barr virus [EBV], cytomegalovirus [CMV], hepatitis), chemicals, and radiation. Elevated platelet count of 840,000 platelets/μL D. The external genitalia do not virilize because there is a lack of testosterone and of DHT. Nyong'o, MD, Associate Principal Investigator, Palo Alto Medical Foundation Research Institute, Associate Clinical Professor, Department of Ophthalmology, UCSF School of Medicine, San Francisco, California Kathy L. Pathogenesis a. Neither shoe orthotics nor physical therapy is useful in scoliosis. Within a few weeks after initial diagnosis, many patients exhibit a temporary progressive reduction in their daily insulin requirements. The answer is D [VI.B and VI.C]. Unlike in microcephaly, the size of the head in a patient with macrocephaly does not necessarily reflect brain size. Death occurs as a result of respiratory insufficiency or pneumonia. Patients will present with increased TSH, low T4, and positive antithyroid peroxidase antibodies. Cardiology. This presentation is consistent with hand–foot–mouth disease, a contagious viral infection caused by coxsackievirus (an enterovirus). Surgical repair is indicated if diplopia persists 2–4 weeks after injury or if enophthalmos is significant. Cholestatic diseases of infancy 1. The seizure discharge is caused by an imbalance between excitatory and inhibitory input within the brain, or abnormalities in the membrane properties of individual neurons. In children with FTT, weight is typically affected before linear growth, which is usually affected before head circumference growth. The severe form is caused by expansion of the number of CTG repeats in the DMPK gene. Absence of any signs of a neurodegenerative disorder e. Random plasma cortisol levels are usually not helpful (although a cortisol level > 20 μg/dL in the presence of stress excludes adrenal insufficiency). Kallman syndrome. Intravenous (IV) doxycycline B. FIGURE 6.1 Determination of midparental height (MPH). He has fever, headache, and a typical rash and has just returned from an endemic area. Measles is a severe illness with complications that include pneumonia associated with significant mortality. This diagnosis should be considered in any infant with macrocephaly, basal ganglia changes on magnetic resonance imaging (MRI), and a movement disorder exacerbated by intercurrent illness. Diagnosis is on the basis of physical examination. Alcohol intoxication can cause pupils to be slow to react but pupils would not be nonreactive. Single lesions may spontaneously resolve after curettage. Pathologic fracture at the site of the infection may occur as a result of healing and remodeling of bone, bone necrosis, or a sequestrum. Hepatic involvement is more common than neuropsychiatric symptoms during childhood. Start a short course of systemic corticosteroids. Clinical features are highly variable. Short-acting inhaled β_2-agonist as needed for symptom relief plus daily medium-dose inhaled corticosteroid B. Congestive heart failure is treated with diuretics, dietary salt restriction, digoxin, and bed rest. Subluxation may recur. Hyperchloremia D. Vitamin D supplementation is not necessary as long as the mother continues supplementing with formula. Skin and Soft Tissue Infections X. Food Allergy V. Physical examination is normal. It is the most common brachial plexus injury and most often involves the right arm as the result of shoulder dystocia, macrosomia, or forceps delivery. Truncal ataxia with deterioration of gait is characteristic. Gaucher disease type I is caused by β-glucocerebrosidase deficiency and is the most common lysosomal storage disease. Electromyography would be expected to demonstrate decreased nerve conduction velocity or conduction block. Approximately 20% of all children in the United States and 80% of children in developing nations have anemia at some time during childhood. Infectious enterocolitis from bacterial pathogens such as Salmonella, Shigella, Campylobacter, Yersinia, and E. Most patients, when they have completed their growth, will be within ±2 standard deviations, or 4 inches, of the MPH. This child has benign intoeing due to tibial torsion. Normal physiology a. Laboratory findings include prolonged aPTT and PT. Congenital muscular torticollis is very common and is usually the result of uterine constraint or birth trauma, either of which causes contracture of the ipsilateral sternocleidomastoid muscle. Abdominal pain is initially diffuse but eventually worsens and localizes to the right upper quadrant. Definitions. Which of the following should be part of her management? A 4-year-old girl who recently returned from Southeast Asia presents with a history of watery diarrhea, vomiting, and decreased urine output. Serum α-fetoprotein is elevated in yolk sac tumors. RPR = rapid plasma reagin; VDRL = Venereal Disease Research Laboratories; ANA = antinuclear antibody. Patients with 3β-hydroxysteroid dehydrogenase deficiency have increased levels of DHEA and 17-hydroxypregnenolone. Myasthenia gravis C. Table 12-3 Differential Diagnosis of Seizure-like Events Breath-holding spells (in infants) Gastroesophageal reflux disease (Sandifer syndrome) Syncope 470 Migraine Vertigo Movement disorder (e.g., tics, chorea) Sleep disturbances (e.g., night terrors, somnambulism) Transient ischemic attack Rage attacks Psychogenic nonepileptic events (pseudoseizures) 471 VI. Tay–Sachs disease is a neurodegenerative disorder caused by hexosaminidase A deficiency. Pseudomonas aeruginosa infection of a bone in the foot can occur if a child has a puncture wound passing through his or her sneaker into the foot. A 5-year-old boy is brought to your office by his parents, who noticed that when their son urinated earlier in the day, his urine appeared red. The answer is C [, section V.D.2]. High reticulocyte count reflects high bone marrow production of RBCs as seen in hemolytic anemias, recent acute hemorrhage, or any other condition associated with shortened RBC life span. It remains a primary cause of cardiac death in the developing world in patients younger than 50 years. Amblyopia is subnormal visual acuity caused by abnormal visual development early in life. Outcome is excellent. Diagnosis is based on clinical features, laboratory studies and genetic testing. This is a group of chronic, inflammatory GI disorders that includes ulcerative colitis (UC) and Crohn Disease (CD), characterized by exacerbations and remissions. Biochemical management includes monitoring and management of serum electrolytes, BUN, creatinine, calcium, and alkaline phosphatase. Dysuria, urinary frequency, and fever are absent, and he is well appearing on examination. A special indication exists for infants 6–12 months of age traveling to countries where MMR is endemic. progressively neurodegenerative disease caused by hexosaminidase A deficiency. Tourette syndrome is defined as having two or more motor tics and at least one vocal tic (do not have to occur at the same time), for at least 1-year duration and beginning before 18 years of age. Secondary AIHA is associated with an underlying disease process, such as lymphoma, systemic lupus erythematosus (SLE), or immunodeficiency. Response rate to current treatments is high. Organic constipation. 3. Microcephaly may be associated with cerebral palsy or seizures. Patients are at increased risk of developing Escherichia coli sepsis. Type I is the most common and the mildest form of OI. G—genital abnormalities, including genital hypoplasia 6. The patient is typically very thin from loss of muscle and body fat. Systemic lupus erythematosus may be diagnosed when four of eleven diagnostic criteria are fulfilled, including photosensitivity and a malar rash. A clinical response with weight gain and resolution of symptoms when gluten is removed from the diet also helps confirm the diagnosis. Any child is at risk for abuse. Protein intake should be restricted to the recommended dietary allowance (RDA) of protein for age. Surgery is required for very severe curves or to stop curves that progress in spite of bracing. Decreased serum ceruloplasmin and increased serum copper b. The liver is the major site of production of most coagulation factors. In infants and preverbal children, the bilateral red reflex test [see section I.B.3] is the best screening test. The booster should given at least 6 months later but ideally no later than 2 years of age. Hepatosplenomegaly, short stature, and corneal clouding 2. Incidence is 1 in 30,000 live births. IEM should be considered in patients with the following clinical presentations. Rewarming of body core with warm saline gastric lavage, bladder washings, or peritoneal lavage should be performed if needed. Major features a. Hereditary hemorrhagic telangiectasia is an autosomal dominant disorder characterized by locally dilated and tortuous veins and capillaries of the skin and mucous membranes. A. Transfusions as needed for severe anemia and thrombocytopenia. Antibiotics are not helpful. Mild disease, seen in 60% of cases, presents with rectal bleeding, diarrhea, and abdominal pain. Therapy includes supportive care and treatment of the underlying cause. Rib hump is visible when the child performs forward bending test. Lead poisoning (see Chapter 1, section IV.J) and chronic diseases, such as malignancy, infections, and kidney disease (termed anemia of inflammation), may present with a 494 microcytic, hypochromic anemia. Neuroimaging may be warranted as well. MELAS = mitochondrial encephalopathy, lactic acidosis, and strokelike episodes; MERRF = myoclonic epilepsy and ragged-red fibers. Patients require ophthalmologic screening owing to the side-effect of potential vision damage with its use. Ryan Coller To Kai, the rest of my family, and my dear friends, for their unwavering support, encouragement, and love, and to my mother, who was excited and very proud of this project and others at her 90th birthday celebration, with much love from Lee. Workup reveals a sweat chloride level of 90 mmol/L, which is consistent with cystic fibrosis. Diagnosis is made by abdominal imaging with CT or MRI scan and finding elevation of the serum tumor marker α-fetoprotein. Easy bruising. For simple febrile seizures, anticonvulsant treatment is usually not initiated until a patient has multiple febrile seizures. Clinical features include pallor, jaundice, and splenomegaly. A 14-year-old Japanese American boy has 3+ protein and 4+ blood with red blood cell casts on a routine screening urinalysis conducted as part of a health maintenance evaluation. Complications of severe UC 1. Hypokalemia is not specifically associated with RTA; however, type IV RTA is associated with hyperkalemia. The prognosis is poor; he will likely have a chronic relapsing course, with a high chance of end-stage renal disease. I have been so privileged to learn from outstanding teachers and mentors at UCLA School of Medicine, Cedars-Sinai Medical Center, and Stanford School of Medicine. transient myeloproliferative disorder, and subsequently for acute myelogenous leukemia (question 108). Key point: Cortisol excess states may be confused with obesity. Racemic epinephrine should be administered immediately. Symptoms of chronic osteomyelitis are usually insidious, so the diagnosis is often delayed and complications are common. Routine chemistries (electrolytes) may demonstrate metabolic acidosis, which may be caused by acquired renal tubular acidosis (in severe cases the heavy proteinuria of nephrotic syndrome may cause RTA). Osteogenesis imperfecta (OI) a. Addison disease is adrenal insufficiency resulting from autoimmune destruction of the adrenal cortex by lymphocytic infiltration. Although unconjugated hyperbilirubinemia predominates early, conjugated hyperbilirubinemia eventually develops as hepatocellular function increases to meet demand. The mechanism of action of folic acid is uncertain. Risk factors for an abusive caregiver include the following: 1. Management is directed toward identifying and treating the underlying cause. I would not have discovered my love for teaching without his mentorship and encouragement. The answer is B [, section V.D.4]. The answer is D [V.K.7]. Disorders of Lymphocytes (Figure 15-2) XI. Family history a. Diagnosis of SS disease is now usually made at birth through state newborn screening programs. Toxicity is typically on the basis of the amount ingested per kilogram of body weight. Further history reveals a 7-day history of fevers and sore throat. However, oral rehydration therapy should not be used for patients with severe lifethreatening dehydration, paralytic ileus, or gastrointestinal obstruction. Chromosomal disorders (e.g., Down syndrome, Turner syndrome) c. Elliptical RBCs are found on blood smear in older children. coli hemorrhagic colitis with certain antibiotics may increase the likelihood that the patient will eventually develop HUS, there is some preliminary evidence that fosfomycin may be protective and could be considered during outbreaks of E. Salinger, MD, FAAP, Palo Alto Medical Foundation, Palo Alto, California, Clinical Instructor, Stanford University School of Medicine, Stanford, California Srinath Sanda, MD, Assistant Adjunct Professor of Pediatrics, Division of Endocrinology, Department of Pediatrics, UCSF School of Medicine, San Francisco, California Margaret Sanford, MD, Associate Pediatric Hematologist-Oncologist, Cedars-Sinai Medical Center, Los Angeles, California, Clinical Assistant Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Harry W. Causes of fat malabsorption a. Access to GnRH agonists (GnRHa) is limited in the United States; therefore, short-acting GnRHa such as leuprolide acetate 208 are used in stimulation tests. Carbohydrates 1. The patient's signs and symptoms are suggestive of Wilson disease, which is caused by a defect in copper excretion leading to copper deposition in the brain, eyes, and liver. The type of intravenous fluids administered depends on the serum level of sodium in the blood. α-Thalassemia and β-thalassemia syndromes a. Older children may have intermittent, crampy abdominal pain and vomiting. Meningitis V. NSAIDs are useful for control of joint pain and swelling, but if given before definitive diagnosis, they may obscure the diagnosis by halting the development of migratory arthritis. Menkes disease is a progressive neurodegenerative X-linked recessive disorder caused by a defect of copper transport. The laboratory and clinical findings of coagulation disorders are summarized in Table 13-6. In severe disease, fresh-frozen plasma (FFP) may be needed. Intussusception D. The patient's history and findings, especially sharp pain that worsens when lying down, in the face of a pulsus paradoxicus, are classic for pericarditis with a pericardial effusion. Myelomeningocele. I am so grateful for my incredible partners on this project, Dr. Lloyd Brown and Dr. Ryan Coller, both phenomenal educators and role models who have taught me so much over the years. Many infectious agents have been associated with Guillain– Barré syndrome, but the most common infectious agent is Campylobacter jejuni, which causes a prodromal gastroenteritis. General Concepts II. Renal imaging to look for structural kidney abnormalities 7. Genetic factors a. Intussusception 1. Premature closure of the coronal suture results in a shortened skull (termed brachycephaly). Although nonaccidental trauma must always be considered, there is no other bruising of concern on examination, and the clinical presentation is most consistent with the diagnosis of Henoch–Schönlein purpura. Child Abuse A. Infants acquire the disorder through autosomal dominant inheritance, most commonly from an affected mother. This infant has a febrile urinary tract infection (UTI) consistent with pyelonephritis. 379 VI. SB occulta. Hydrocephalus. Electromyography (EMG) and nerve conduction studies may be considered if no recovery has occurred within 6 months. NAC is given orally as a 140 mg/kg loading dose and is followed with 70 mg/kg every 4 hours for 17 doses. No amount of alcohol is considered safe, but the risk for FAS is higher with binge drinking or chronic alcohol consumption during pregnancy. There are three categories of NS: 1. Annual screening with TSH/T4 and TTG IgA is recommended. Disorders of Sexual Differentiation (DSD or Formerly Known as Ambiguous Genitalia) IV. 20 mL/kg boluses of intravenous (IV) solutions with a high enough oncotic load (e.g., normal saline or lactated Ringer solution) are commonly used. Atresias 1. Complications include spondylolisthesis, which is discussed below. Perinatal history is remarkable for a term gestation complicated by gestational diabetes mellitus. After birth, chest radiographs reveal little or no gas in the abdomen, absence of the diaphragmatic dome, mediastinal shift to the contralateral side (usually to the right), and bowel loops in the thorax (usually on the left side). Treatment includes transfusions. Diarrhea may also be present, and severe proteinuria and hypertension are more consistently found. Long-term complications 1. Infections (e.g., sepsis, hepatitis, viral infections) 2. Any pathologic condition that unilaterally blurs the retinal image, such as a congenital cataract B. Onset usually occurs during early infancy for organic causes of constipation, and after toilet training for functional constipation [see section VII.D]. Bruises on fleshy or protected areas, such as the face, neck, back, chest, abdomen, buttocks, and genitalia, are often consistent with inflicted injury. Complete blood count (CBC), erythrocyte sedimentation rate (ESR), thyroxine (T4), thyroid-stimulating hormone (TSH), tissue transglutaminase IgA (TTG IgA), total serum IgA, serum electrolytes including calcium and phosphorus, and serum creatinine and bicarbonate levels should be obtained. A direct Coombs test is positive (detects coating of antibodies on the surface of RBCs or complement). After birth, copious oropharyngeal secretions with feeding difficulty, with increased risk of choking and aspiration c. During a health maintenance visit, the parents of a 9-month-old infant note that their infant is unable to sit alone or roll over and startles very easily to sound. Four-year-old boy with microcephaly, hypertelorism, mental retardation, and a deletion on the short arm of chromosome 5 3. Because of the availability of DNA testing, EMG and muscle biopsy are no longer indicated. Muscular dystrophy rarely presents during infancy. You can also use it for revision of other exams. 712 IX. Rehydration with half normal saline (½ NS) with 20 mEq of KCl 21. In young children, the headache is often bifrontal. She has decreased urine output and appears very pale. Sphingolipidoses. Hurler syndrome, a mucopolysaccharidosis in which glycosaminoglycans deposit in various tissues causing a progressive clinical picture, is characterized by corneal clouding, changes to the bone termed dysostosis multiplex, organomegaly, and progressively coarsened facies, including frontal bossing, widened nasal bridge, and thickening of the nasopharyngeal tissues. Gastritis or ulcers may occur as a result of severe stress of illness, surgery or burns, or from medications. Child Abuse VII. Neural tube defect is a broad term that includes all forms of failure of neural tube closure, from anencephaly to sacral meningocele. Half of the cases are inherited, and the remainder are spontaneous mutations. The ABCs are the initial management priority. Evaluation. Newborns may swallow maternal blood during delivery or while nursing from a bleeding nipple. Coller, MD, MPH, FAAP, Chief, Division of Pediatric Hospital Medicine, Assistant Professor of Pediatrics, University of Wisconsin School of Medicine and Public Health, Madison, Wisconsin Lee Todd Miller, MD, FAAP, Associate Dean for Student Affairs, Professor of Pediatrics, Director, Global Health Education Programs, David Geffen School of Medicine at UCLA, Los Angeles, California Dorsey Bass, MD, FAAP, Co-Director, Stanford Children's Inflammatory Bowel Disease Center, Associate Professor of Pediatrics, Stanford University, Stanford, California Robert M. Head circumference is initially spared in FTT. A urinalysis demonstrates 25–50 white blood cells per high-power field. See Table 17-2. 649 2. Amniotocele (dacryocele) 1. It typically involves the large joints of the lower extremity and can wax and wane based on GI disease activity. Neonatal metabolic (state) screening 1. Medications may be very helpful. Galactosemia is an autosomal recessive disorder caused by galactose-1-phosphate uridyltransferase (GALT) deficiency. Painful blisters typically develop on palms, on soles, and in the mouth. Intestinal atresia is the most common cause of obstruction in the neonatal period. Autosomal dominant disorder caused by mutations in certain genes involved in the RAS/MAPK pathway. An oculocephalic maneuver is negative. 365 3. Surgery is indicated if there is more than 50% involvement of the femoral head, or if there is subluxation of the femoral head out of the acetabulum. Miscellaneous Infections Review Test Answers and Explanations Chapter 8 Cardiology I. The clinician should take a detailed history about possible causes of failure to thrive, focusing in particular on caloric intake. Liver transplantation may be necessary in cases of severe liver failure. For all vaccines, moderate to severe illness (with or without fever). As a result of this obstruction, the pathways of the ventricular system do not communicate correctly, and there is a buildup of CSF proximal to the blockage. WBC count at diagnosis 99%) in SLE and is an excellent screening test. In addition, the infant appears very sleepy, and his mucous membranes are dry. A positive family history is common. Consider rickets in patients with frontal bossing, bowed legs, low serum phosphorus level, and high serum alkaline phosphatase [see section X.C]. 7. IVIG may also be effective. The answer is D [IV.B.7]. Treatment includes identifying and stopping the causative agent, transfusions as needed, bone marrow transplant, and immunosuppressive therapy. Definitions a. However, toxic-appearing children, neonates, and patients who have significant dehydration should be hospitalized and administered intravenous antibiotics initially. Patients are also at an increased risk for infection with encapsulated organisms, such as Streptococcus pneumoniae, and therefore may present with spontaneous bacterial peritonitis, pneumonia, or overwhelming sepsis. Active immunization involves induction of long-term immunity through exposure to live attenuated or killed (inactivated) infectious agents. As with upper GI bleeding, assessment of hemodynamic status, monitoring for ongoing bleeding, and stabilization are essential. Unlike anti-dsDNA, anti-Sm antibody levels cannot be used as a measure of disease activity. The male-to-female ratio is 2–3:1. Amblyopia 1. Idiopathic (e.g., neonatal hepatitis) 6. Trisomy 21 is the cause of Down syndrome and is associated with hypotonia, prominent epicanthal folds, upslanting palpebral fissures, and single palmar creases. Diagnosis after birth a. Perkins, James Heaysung Lee 20 I. Prader–Willi syndrome is associated with hypogonadism, almond-shaped eyes, short stature, and hyperphagia with obesity during childhood. Inheritance Patterns II. A 7-year-old boy has just returned from a month long visit with his grandparents in the Midwest. Testes are unaffected in Down syndrome and in Williams syndrome. This syndrome is due to defective 3βhydroxysteroid-Δ7 reductase, the last step in cholesterol biosynthesis. Pharyngoconjunctival fever is characterized by an upper respiratory infection that includes pharyngitis, fever, and bilateral conjunctivitis. Strong hereditary component (stronger for type 2 than for type 1) 2. Note that caput succedaneum (scalp edema) and cephalohematoma (subperiosteal hemorrhage of the newborn cranium after a traumatic delivery) 22 may interfere with accurate head circumference measurement in newborns. Plasma lipid clearance is reduced because of reduced activity of lipoprotein lipase in adipose tissue. Preventive treatment options include propranolol, amitriptyline, topiramate, valproic acid and cyproheptadine. Increased risk for foramen magnum stenosis and compression of the craniocervical junction. Kamil, MD, Pediatric Nephrologist at Cedars-Sinai Medical Center and Harbor-UCLA Medical Center, Health Sciences Clinical Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California James Heaysung Lee, MD, FAAP, Associate Program Director, Pediatrics, Assistant Clinical Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Deborah Lehman, MD, FAAP, UCLA, Los Angeles, California Professor of Pediatrics, Assistant Dean for Student Affairs, David Geffen School of Medicine at Jason T. DIC refers to a group of laboratory and clinical features indicative of both accelerated fibrinogenesis and fibrinolysis. The angle at the intersection of these lines is the Cobb angle. NS that results from systemic diseases, including SLE and HSP D. Diagnosis is made by resolution of acute symptoms within a few days after complete withdrawal of the suspected antigen. Older infants and toddlers, who may wander into unfenced pools or tip over into water containers (e.g., toilets and buckets) 2. Major advantages include immunogenicity and efficacy in preventing meningitis, pneumonia, bacteremia, and otitis media from the most common pneumococcal strains in children younger than 2 years. Wilson disease should always be considered in a patient with personality changes, ataxia, and seizures. Fever, anorexia, and vomiting are common. West syndrome is a triad of infantile spasms, hypsarrhythmia, and developmental delay or regression. Nonmetastatic hepatoblastoma carries a cure rate of about 75%, whereas hepatocellular carcinoma carries a cure rate of about 40%. Lysosomal Storage Diseases A. Clinical problems of SGA infants are listed in Table 4-4. Common preceding infections include varicella, coxsackievirus, Epstein–Barr virus (EBV), and mycoplasma. In the prepubescent state, sex steroids (testosterone and estradiol) are suppressed owing to an inactive hypothalamic–pituitary–gonadal axis (HPGA). Fetal hemoglobin (Hgb F) is a major constituent of Hgb during fetal and early postnatal life. 643 III. Table 18-1 presents information about the differential diagnosis of an abnormal red reflex. I. Hydroxychloroquine is useful for patients with mild disease and is often used as a maintenance therapy. SIDS is the most common cause of death in children younger than 1 year. Immunizations IV. The answer is B [IV.C]. Infant jaundice is defined as elevated bilirubin after the neonate period and within the first year of life. As symptoms progress, weight loss, vomiting, and dehydration occur. The great majority of patients (70–80%) with acute rheumatic fever have elevated antistreptolysin-O titers. Obstruction of the cystic duct causes increased intraluminal pressure and distension, increased secretion of enzymes and prostaglandins, and progressive inflammation. Tuberous sclerosis E. McCulley, MD, Division of Neonatology, Assistant Professor of Pediatrics, University of Wisconsin School of Medicine and Public Health, Madison, Wisconsin Omondi L. Dysautonomia, arrhythmia, orthostatic hypotension, and urinary retention may occur. Detailed history and complete physical examination. Management includes supportive care for symptoms and enzyme replacement therapy. Bone scan 621 or MRI can detect osteomyelitis a few days after the onset of symptoms. DNA testing for the abnormal gene is diagnostic in >90% of cases. 384 IX. Note that Tdap rather than DTaP is given to children 7–10 years of age if they did not complete their series of DTaP by their seventh birthday. The diagnosis of systemic-onset juvenile idiopathic arthritis should be considered in a child presenting with fever of unknown origin, an evanescent salmon-colored rash, arthritis, organomegaly, and polyserositis. Her risk of developing coronary artery aneurysms if left untreated is approximately 20%. Chronic diseases can cause pubertal delay (e.g., inflammatory bowel disease, anorexia nervosa, renal failure, and heart failure). Twenty-four–hour urinary protein collection (normal is 3.5), hypoalbuminemia, hypercholesterolemia, and edema. For patients not in adrenal crisis, replacement with oral hydrocortisone (~8 mg/m2 per day) and mineralocorticoids is sufficient. Constipation is very common during childhood. Systemic-onset JIA A 3-year-old girl is referred to you for evaluation of fever. AKI is managed as described in section X.E. c. Coller Lee T. Patients with achondroplasia are short from birth with shortening of the proximal long bones (rhizomelic short stature), those with Russell–Silver syndrome have short stature with skeletal asymmetry, and those with Marfan syndrome are tall, not short. It is rare in Asians and African Americans. Approach to the Comatose Patient V. Breastfed infants defecate more frequently during the first months of life; however, this decreases rapidly, so that there is no difference in stool pattern between breastfed and formula-fed infants by 4 months of age. Advantages: may be administered as early as at 6 months of age. 10. Attacks of severe unilateral facial/orbital pain b. This group of disorders involves inflammation of entheses (tendinous insertions into bones), joints, or axial skeleton and are characterized by the absence of RF, ANA, or other disease-specific serologic markers. Timing of vaccination: HIB vaccine is recommended at ages 2, 4, and 6 months with a booster at ages 12–15 months. Patients with 11β-OH deficiency have increased levels of 11-deoxycortisol and 11deoxycorticosterone. Hereditary Renal Diseases IX. Respiratory Distress Syndrome (RDS) A. Chiari type II malformation is characterized by downward displacement of the cerebellum and medulla through the foramen magnum, blocking CSF flow. The subluxated vertebra can impinge on nerve roots. Patients with renal failure may require dialysis, fluid and electrolyte management, and ultimately renal transplant. Normal periodic peristaltic waves of contraction of the esophagus transport ingested food into the stomach. About one-third of patients have only one episode, one-third have infrequent relapses, and one-third will have frequent relapses. Women who carry the gene are less likely to develop ESRD, but it may occur. Patients have a painful limp with pain in the groin, hip, or knee. 31 c. Autosomal recessive polycystic kidney disease (ARPKD) or infantile polycystic kidney disease 1. These headaches are extremely rare during childhood. 412 413 FIGURE 11-2 Differential diagnosis of hematuria. Shaken baby syndrome D. Diagnosis. High risk for lens dislocation. 541 Ewing Sarcoma Flat bones and diaphysis of tubular bones; occasionally extraosseous Most common sites (in order): axial skeleton (especially pelvis), humerus, femur Pain, swelling, and soft tissue mass Fever, malaise, and weight loss Leukocytosis and elevated ESR Periosteal reaction with "onion skin" appearance Destructive changes Occurs in 25% at presentation Lungs (50%), bone (25%), and bone marrow (25%) VIII. The disease is more common in boys and has a male-to-female ratio of 4:1. In severe classic forms of the disease, death usually occurs by later adolescence. Congenital infection with mumps has not been described. Children who are treated, even with arthritis or neurologic manifestations, have an excellent prognosis. The causes of central cyanosis are broad and include impairments in ventilation (e.g., central nervous system and airway abnormalities), lung pathology (e.g., imbalance of ventilation and perfusion, diffusion abnormalities), right-to-left cardiac shunts (e.g., the "5 T's" of cyanotic congenital heart disease: tetralogy of Fallot, transposition of the great vessels, truncus arteriosus, tricuspid atresia, and total anomalous pulmonary venous connection—see also Chapter 8, section IV), hematologic disorders (e.g., polycythemia), and metabolic abnormalities (e.g., hypoglycemia, hypocalcemia, hypothyroidism, and hypothermia). Laboratory evaluation and management(Table 5-4) depends on presenting features and clinical status of patient (acute episode or stable between episodes). Complications. Bleeding and complaints of "growing pains" may be presenting complaints. The answer is A [, section V.A.3]. Definitions 1. Frog-leg posture when in the supine position, with generalized hypotonia, weakness, and areflexia 4. The rash on the trunk is nonpruritic and is likely evanescent. Serial transcranial Doppler ultrasound or magnetic resonance angiography is recommended beginning at 2 years of age to identify patients at increased risk for stroke. Soft Tissue Tumors VII. Thirty percent of patients have bilateral involvement, and patients with hypothyroidism are especially at risk for bilateral disease. You will be required to select one answer 603 for each statement in the following set. The diagnosis of meningococcemia is also unlikely because this patient is nontoxic, alert, and afebrile. These results should be available rapidly, and levothyroxine can be initiated when hypothyroidism is confirmed. The intestines normally return to the abdomen through the umbilical cord 372 during the 10th week of gestation and undergo counterclockwise rotation about the axis of the superior mesenteric artery. The majority of infections 394 in children and adolescents are caused by Hepatitis A infection (HAV) and Hepatitis B infection (HBV). The underlying cause of his mental retardation has never been identified. Reproductive Health Issues in Males Review Test Answers and Explanations Chapter 4 Neonatology I. Hemolytic uremic syndrome (HUS) is suspected. His voice is muffled. Ro (SSA) and La (SSB) antibodies are less strongly associated with SLE (more specific for Sjögren syndrome). Iron a. Definition a. Acute Kidney Injury A. As the disorder progresses, patients develop blindness, seizures, and autonomic dysfunction. Two-thirds of cases in children present before 5 years of age. Malabsorption III. Systemic signs and symptoms depend on the cause and severity of the renal insult but often include lethargy, nausea, vomiting, respiratory distress, hypertension, and sometimes seizures. FTT was previously categorized as either nonorganic (due to psychosocial factors) or organic (due to underlying organ system pathology). The earlier the diagnosis, the less likely surgical intervention will be necessary. Persistent, inconsolable crying lasting ≥3 hours occurring within 48 hours after prior vaccination c. Head and neck exam. Symptoms include diarrhea, bloating, and abdominal pain. You are called to the newborn nursery to evaluate a 1-day-old female infant with unusual physical findings. This is termed a negative oculocephalic maneuver or negative doll's eyes. Other examples of defects of amino acid metabolism are described in this section. Consider multiple agents in adolescents. Fetal alcohol syndrome (FAS) a. 499 FIGURE 13.1 Classification and differential diagnosis of anemia. ARPKD is uncommon, occurring in approximately 1 in 10,000–40,000 live births. The boy is very active, easily distracted, and unable to perform skills at the same level as others in his class, and the teacher expresses concern and wonders whether the boy could have a severe learning problem. Congenital glaucoma [see section VI], which is characterized by tearing, an enlarged globe, and corneal edema b. Bag-and-mask ventilation should not be used because this may distend the bowel and increase compression of the lung. Two-thirds of patients present between the ages of 4 and 6 weeks with jaundice, dark urine, and pale or acholic stools. From exposure to those who are genuinely better than me, I have received invaluable education, encouragement, and perpetual inspiration. Tanner staging. Development of chronic and debilitating arthritis is likely. Specific causes of PPP in males that result in testicular enlargement 1. Which of the following laboratory findings would be most consistent with suspected renal tubular acidosis? Although the erythrocyte sedimentation rate is often elevated, it is nonspecific because it is also elevated in many other inflammatory conditions, such as infectious disorders, some malignancies, and other rheumatologic conditions. Medical Management d. Long-term survival is >95%. Oral rehydration therapy may be effective even if this child has a secretory diarrhea. Alagille syndrome a. Red eye in older infants and children 1. Age is an important factor in determining the cause of lower GI bleeding (Table 10-5). Intravenous deferoxamine, an iron-binding ligand, should be given if any of the following condition exists: a. This leads to pulmonary hypoplasia, which may be incompatible with life. Bone and Joint Infections (see Chapter 17, section III.B) XI. Etiology of hypergonadotropic hypogonadism a. It occurs with excessive external rotation, abduction, and extension of the shoulder, as it may occur in gymnastics or wrestling. Endocrinology. Which of the following statements regarding UTI in this infant is correct? Rheumatic fever is now rare in the United States and Western Europe but was a worldwide problem during the 1960s. This process affects the skin and vessels of the heart, kidneys, lungs, and GI tract. Proportionate short stature is defined as short stature with a normal U/L ratio [see section I.C.2.c]. Other laboratory abnormalities may include leukocytosis, hyperglycemia, hypocalcemia, elevated transaminases, and coagulopathy. Sudden onset of pain, which is difficult to localize b. Thus, HPV vaccine is one of the two cancer-preventive vaccinations, the other being Hepatitis B vaccine. Classification based on reticulocyte count is also helpful. Well-Child Care—General Concepts A. Overview. Review of systems should focus on associated symptoms including fever, vomiting, diarrhea, constipation, anorexia, dysuria, or worsening pain with eating. Children with corneal involvement should be referred to an ophthalmologist. Brown In health and education fields, we are constantly striving to be idealized versions of ourselves— better than we truly are. Galactosemia C. There are no maternal antibodies, and this disorder consists of multiple subtypes. Sometimes they didn't even explain the abbreviation originally and it was somehow implied that the reader should know it, even if it is unheard of. The majority of cases result from a sporadic mutation. The edema often follows an upper respiratory infection (URI). Signs and symptoms are those characteristic of anemia and thrombocytopenia. A strict gluten-free diet for life is necessary and results in complete reversal of intestinal damage. Routine immunizations and also yearly influenza vaccination, 23-valent polysaccharide pneumococcal vaccine at 2 years of age, and meningococcal vaccine should all be given. Phlebotomy is also used to keep the Hct < 60%. It is important to focus on establishing whether cholestasis exists and whether the problem is isolated to the liver. Rotavirus vaccines a. Cataracts f. Newborn screening programs can diagnose several IEM before symptoms develop; however, the specific disorders that are screened differ from state to state. Proteinuria A. There are about 150 different types of HPV, some of which cause significant health problems such as genital warts and certain cancers (e.g., cervical, vaginal, anal, penile, and oropharyngeal). Early localized disease, or late disease with arthritis only, is typically treated with doxycycline (for children ≥9 years of age), amoxicillin, or cefuroxime. These symptoms may overlap with an adult presentation of HIV infection, but it would be very unusual for a young child with HIV infection to present in the first few years of life with cytomegalovirus (CMV) retinitis, shingles, or central nervous system (CNS) lymphoma, all which are acquired immunodeficiency syndrome (AIDS)–defining illnesses in adults with HIV infection. Unless the patient is septic, antibiotics should be held until blood cultures are obtained and the bone has been aspirated for culture. Spinal muscular atrophy, type 1 E. 602 5. Proteins are converted to peptides and amino acids. Pyloroplasty may be performed to improve gastric emptying. Retinopathy of Prematurity (ROP) A. A 3-year-old boy presents to the emergency department for evaluation of acute right upper quadrant abdominal pain. Patients are also given oral phosphate binders and vitamin D analogues to prevent renal osteodystrophy. Types of immunizations 1. Neonates and infants: evaluation of eye fixation, tracking of light or objects, and pupillary responses b. Spina Bifida IV. Mothers of infants with congenital myotonic dystrophy have myotonia, an inability to relax contracted muscles, which manifests as difficulty releasing a hand grip during a firm handshake. Presentation classically includes abscess formation, and history of infection of the bones, skin, 768 109. Other symptoms such as cough, abdominal pain, and headache with visual aura are not part of the DSM-5 criteria, although abdominal pain and headache may be present in a depressed teen. Abnormal EMG (used only when diagnosis is not certain) 5. The bond between CO and hemoglobin is more than 200 times stronger than the bond 708 between oxygen and hemoglobin. Common features include short stature, intellectual disability, hepatosplenomegaly, and skeletal abnormalities. Studies show thrombocytopenia, prolongation of PT and aPTT, reduction in clotting factors (especially fibrinogen and factors II, V, and VIII), elevated fibrin degradation products (positive D-dimer assay), and fragmented and helmet-shaped RBCs on blood smear. An intact (normal) cold caloric response is reflected by eye deviation to the ipsilateral side, with nystagmus to the contralateral ear. Categories 1. EMG is sometimes performed and demonstrates brief, small-amplitude muscle potentials with an incremental response during high-frequency stimulation. H—heart defects such as conotruncal defects and aortic arch abnormalities 3. The reticulocyte count reflects the number of immature RBCs in the circulation and, therefore, the activity of the bone marrow in producing RBCs. The usual percentage of RBCs that are reticulocytes is 1% (normal absolute count = 50,000 cells/mm3). Potential poison dose is calculated for the worst-case scenario. Obtaining a history of abuse from a young child is difficult. A 4-month-old female infant is brought to your office by her parents, who are concerned about some behaviors they have witnessed. A 12-year-old boy who has three renal cysts on a renal ultrasound that was performed for the evaluation of microscopic hematuria. General concepts a. Infant mortality can be as high as 40% because of respiratory problems. Respiratory Distress Syndrome (RDS) VII. Evaluation for renal osteodystrophy (i.e., bone disease secondary to renal failure) 5. Pierre Robin sequence a. In this patient, the fracture has resulted in a break in the skin (open fracture), and the ends of the fracture override (overriding ends). Extraintestinal manifestations are less common during childhood and are noted in Table 10-4. Immunology Overview X. Other findings include a normal or increased reticulocyte count. There is a presumed genetic cause, and some cases may have an autosomal dominant inheritance pattern. Abdominal pain occurs in the periumbilical or epigastric area and may radiate to the back. The MPH should be noted on the growth chart. Other sites of involvement include the trunk, retroperitoneum, mediastinum, and paratesticular and perianal regions. Klippel–Feil syndrome is defined as congenital fusion of the cervical vertebrae. Liver involvement of variable clinical severity is a constant finding, including cirrhosis with portal hypertension. Feeding refusal with irritability or constant hunger may indicate esophagitis. All four forms of SMA are caused by mutations in the survival motor neuron gene (SMN1) on chromosome 5. Radiographs of the cervical spine may identify abnormalities of the cervical spine and should be obtained in anyone presenting late or failing to improve with physical therapy. A fluctuant midline mass is present anywhere along the spine, but most commonly in the lumbosacral region. SIDS prevention includes firm bedding and sleeping supine. Pathology of the spinal cord shows degeneration and loss of anterior horn motor neurons and infiltration of microglia and astrocytes. Stress and anxiety reduction may provide long-term relief. Thrombocytopenia is defined as a decreased number of platelets, generally 90%. Treatment is by monthly intramuscular vitamin B12 injections. Major causes of hypercortisolism a. Movement Disorders IX. The more common causes include the following: 1. Liver Abnormalities and Hepatitis A. The answer is C [II.A.1.a]. Seventy to eighty percent of children with migraines have at least one affected parent. During the first 7 weeks of gestation, the gonadal tissue remains undifferentiated. Gastric emptying study (or scintigraphy) uses a radioactive marker (e.g., technetium 99m) mixed into age-appropriate foods to measure the rate of gastric emptying. Tension headaches occur toward the end of the day, whereas headaches from increased ICP occur in the morning. Despite the presence of petechiae, the platelet count is normal (i.e., a nonthrombocytopenic purpura). Control of inflammation a. Treatment options include observation, bracing, and surgery, and management decisions are based on the magnitude of the curve and the remaining growth potential. Common Genetic Disorders V. It is difficult to predict the level of bilirubin from the extent of jaundice on physical examination. The evaluation of an ALTE should include attempts to identify an underlying cause, and workup may include an electroencephalogram to rule out a seizure disorder, an electrocardiogram to rule out a dysrhythmia such as long QT syndrome, electrolytes, a barium esophagogram or pH probe study to rule out gastroesophageal reflux disease, and a sleep study. Elevated WBC count 2. There is no abnormality in the hypothalamus or pituitary gland. For the steroid-resistant patient a prolonged course of tacrolimus plus steroids may induce a remission. Cardiac inflammation often leads to severe valvular dysfunction, which may require intervention immediately or many years after the event. Cardiac involvement with conduction abnormalities and dilated cardiomyopathy D. The most common enteric pathogens which are associated with reactive arthritis include Salmonella, Shigella, Yersinia, Campylobacter, and Clostridium difficile. This patient has features consistent with systemic-onset juvenile idiopathic arthritis (JIA). Prognosis. The incidence is inversely related to gestational age with greater than 90% of infants 90% of cases) involves proximal esophageal atresia with a distal tracheoesophageal fistula (see Figure 4-5). Acquired torticollis arises later in childhood and is much less common than congenital torticollis. General Considerations II. Migraine headaches are frequently unilateral (although commonly bilateral in children) and may begin in the periorbital area and spread to the forehead and occiput, whereas tension-type headaches are often generalized or bitemporal. Asthma IX. SGA, failure to thrive, and small stature 3. Duodenal atresia is a congenital obstruction of the duodenum caused by failure of the lumen to recanalize at 8–10 weeks' gestation. The answer is B [III.F.3, III.F.4.c, and Table 9-2]. BECTS involves nocturnal focal seizures occasionally with secondary generalization. Cystic fibrosis (See Chapter 9, section IV.B.) 2. Sandifer syndrome is a characteristic torticollis with arching of the back caused by painful esophagitis. Secondary infections may result from anaerobic and aerobic organisms, such as Staphylococcus aureus, Pasteurella multocida, and Streptococcus species. Without surgical intervention, this process usually leads to perforation within 36–48 hours. Primary prevention 1. Immunosuppressive therapy (e.g., corticosteroids, cyclosporin) may also help. This patient's eye findings are termed the setting-sun sign, or a tonic downward deviation of the eyes. It is important to note that patients with β-thalassemia minor may be very easily misdiagnosed as having iron-deficiency anemia and treated inappropriately with iron. Patients with nephrotic syndrome and fever should therefore be treated empirically with antibiotics pending culture results. Usually results in impaired vision 2. Fetal phenytoin syndrome D. Imaging a. Rationale for vaccine: Influenza can be a very serious illness that may result in hospitalizations and even death, regardless of a child's medical history. Peak incidence is at 2–4 months of age. Most cases of scoliosis (80%) are idiopathic. He now acts normal except for his cold symptoms, according to his parents. Gaucher cells (foamy macrophages) are found on bone marrow biopsy. Type 2 Diabetes Mellitus (Type 2 DM) A. This patient's shortness of breath may be caused by congestive heart failure or a pericardial effusion. SS hemoglobinopathies a. Metabolic storage disorders (e.g., Canavan syndrome, gangliosidoses) d. Disorders of Puberty A. Genetic testing for mutations associated with nephrotic syndrome may be indicated in the child who presents with nephrotic syndrome before their first birthday and in the steroid-resistant child. Cervical hydrosyringomyelia (accumulation of fluid within the central spinal cord canal and the cord itself) 3. BAER/ABR measures electroencephalographic (EEG) waves generated in response to clicks via electrodes placed on the infant's scalp. The incidence is approximately 1 in 3000 live births. The earlier the onset, the longer the duration of the abnormal stimulus, and the more blurry the image, the more severe the vision loss. Legg–Calvé–Perthes (Perthes) disease is idiopathic avascular necrosis of the femoral head. A group of autosomal recessive disorders caused by defects in lipid metabolism. Six months ago her weight was at the 50th percentile, and now her weight is at the 25th percentile for age. Age of onset is between 18 months and 7 years. Laboratory studies, intravenous access, and fluid resuscitation are indicated as for upper GI bleeding. Perinatal history. The cause is a type 1 hypersensitivity reaction. Tension headaches are bifrontal or diffuse, dull, aching headaches that are often associated with muscle contraction. FIGURE 18.1 Differential diagnosis of red eye in older infants and children. There are three subtypes of hemolytic uremic syndrome (HUS), a Shiga toxin–associated form (the most common form in childhood), pneumococcal- associated form, and an atypical form caused by medications or complement system dysregulation which is often inherited. This patient's disease is more common in males than in females. Coccidioidomycosis is treated with antifungal medications (fluconazole). About 20,000 children are hospitalized annually in the United States with influenza, and around 40% of children who die from influenza have no significant past medical history. Neck flexor muscle weakness b. The most common toxic exposures involve commonly used household products. Bacterial tracheitis presents with fever and stridor (rather than a muffled voice), and drooling and neck hyperextension are unlikely to be present. The diagnosis is based on the findings of albuminocytologic dissociation in the cerebrospinal fluid and by decreased nerve conduction velocity on electrophysiologic studies. Symptoms vary with the type of RTA and with the patient's age. Acute Abdominal Pain A. Endoscopy is performed to assess the degree of damage. Treatment principles include controlling symptoms, reducing recurrences, and optimizing nutrition. If you have an exam of pediatrics, you can read this book few days before exam date. The goal of the emergency phase of intravenous rehydration is to restore or maintain intravascular volume to ensure perfusion of the vital organs. Careful family history 3. If there are signs of a diffuse tubular disorder (manifested by hypokalemia, hypophosphatemia, and aminoaciduria), the patient should be evaluated for Fanconi syndrome by performing more extensive testing of other tubular functions. 650 VI. Hemarthroses (involving the knees, elbows, and ankles most commonly) and deep soft tissue bleeding are the hallmarks. The headaches are bilateral, dull, and achy, but mild. Normal defecation frequency varies with age, with an adult pattern developing by about 4 years of age. Migraine without aura is the most common form of migraine in children. This is typically present by 1 month of age. Signs on examination may include jaundice, hepatosplenomegaly, ascites, increased abdominal vascular markings, caput medusae, spider hemangiomas, and clubbing. Dietary deficiency is unusual, except during early infancy. Photophobia or phonophobia occurs. 9. Mild, episodic constipation can usually be addressed by ensuring the diet contains adequate soluble fibers, and by increasing the amount of water and sorbitol-containing juices. Platelet function assay g. Treatment includes attempts at reducing entry of stomach contents into the esophagus and relieving symptoms. The level should be plotted on the Matthew–Rumack nomogram to determine the potential for hepatitis. Soy formulas, while appropriate for galactosemia, still contain a full amount of branch-chain amino acids, and similar to regular formulas would overwhelm the patient's ability to metabolize the branched chain amino acids, resulting in toxic exposure as well. Infant and Child Cardiopulmonary Resuscitation (CPR) II. The parents remind you that this is his second febrile seizure. Screening of family members of patients with type 1 DM through national screening programs is encouraged. Diet. Evaluation of delayed puberty. Complications include skull deformity and facial asymmetry (plagiocephaly) if torticollis is not treated promptly. Now he is ready for discharge from the neonatal intensive care unit and will go home on 0.25 L/minute of oxygen by nasal cannula. Respirations may be absent or irregular, and the victim may cough up pink, frothy material. Normal Developmental Milestones II. Mechanical injury to the mucosa from vomiting (i.e., Mallory–Weiss tear) or from foreign body or caustic ingestion may cause UGI bleeding. Motility agents increase the LES tone or increase gastric emptying. Categorization of short stature (Figure 6-2) 1. Ataxic or agonal breathing (irregular respirations with no particular pattern) indicates medullary

injury and impending brain death. Clinical features (Table 11-2). Epidural steroids may be considered. There is a risk of cerebral edema if deficit replacement occurs too quickly in patients with hypernatremic dehydration. Diagnostic criteria includes the following: a. Children with systemic-onset JIA have a 50% chance of having a severe, erosive arthritis. This patient presents with an acute exacerbation of her chronic asthma and therefore would benefit from a short course of systemic corticosteroids. The typical victim is found dead in the morning in bed after being put to sleep at night. Encopresis is defined as the developmentally inappropriate release of stool, unrelated to an organic etiology. Pulmonary metastases identified at the time of diagnosis are resected if still evident after initial chemotherapy. RDS is respiratory distress caused by a lack of surfactant, most frequently in preterm infants. Pain is localized and increases with hyperextension. Brown Ryan J. Seronegative spondyloarthropathies. Plasmapheresis removes the patient's plasma along with the presumed antimyelin antibodies and is performed over a 4- to 5-day period. Immunologic response usually follows streptococcal pharyngitis by 2–7 months. Common Fractures Review Test Answers and Explanations Chapter 18 Ophthalmology I. Normocytic, normochromic anemia (normal RBCs in size, color, and shape; normal MCV) 2. 88. Frequent, recurrent, or prolonged febrile seizures do pose a risk and may require additional treatment with daily anticonvulsant prophylaxis or the use of rescue medications (e.g., rectal diazepam). Types of Genetic Tests III. Acquired microcephaly is caused by a cerebral insult in the late third trimester, perinatal period, or first year of life. A pseudocyst is a collection of fluid rich in pancreatic enzymes that arises from pancreatic tissue. Clinical features include diarrhea, malabsorption, and FTT. Valvulitis is frequent and typically causes insufficiency of the left-sided valves (mitral and aortic). Infection/sepsis is also a common cause of cyanosis due to respiratory depression and lung pathology. These may be classified as “major” and “minor” features (Jones criteria). The usual mechanism of injury is an upward force on the arm, such as pulling a toddler upward by the hand or wrist to help the child stand or clear an obstacle. False-positive guaiac results may occur because of ingested iron, rare red meats, beets, and foods with high peroxidase content, such as cantaloupe, broccoli, and cauliflower. Management of symptomatic infection is supportive. The renal impairment is caused by toxin binding to renal vascular endothelial cells. The most common cause of hypothyroidism is Hashimoto thyroiditis [see section IX.B.2.b]. Laboratory findings (Table 13-4) f. High-dose inhaled corticosteroid as needed for symptom relief For each description of a patient with asthma, select the most appropriate initial pharmacologic therapy. Prader–Willi syndrome (see Chapter 5, section IV.E.2) 3. Carditis and meningitis require intravenous ceftriaxone. Oculocephalic maneuver (doll's eyes). 443 Review Test 1. Guaiac is a colorless dye that changes color from the peroxidase activity of hemoglobin in the presence of hydrogen peroxide developer. Osteogenic sarcoma is a malignant tumor that forms osteoid or new bone. Empiric oral antibiotics are administered to prevent infectious contamination of the orbit from organisms from the maxillary sinus. Optic nerve damage in children with sickle cell disease 4. common cause of viral parotitis, but other viruses can be implicated. The clotting cascade is depicted in Figure 13-2. Analysis of the synovial fluid for cell count, Gram stain, and culture is important. Dacryocystitis (infection of the nasolacrimal sac) c. Caustic agents. A congenital cervical spine abnormality (e.g., Klippel–Feil syndrome) may cause congenital torticollis, but this is uncommon. Initial stabilization of hypovolemia and anemia is essential. SIDS is the death of an infant younger than 1 year whose death remains unexplained after a thorough case investigation that includes autopsy, death scene evaluation, and review of the clinical history. Surgical decompression is necessary to avoid avascular necrosis and to obtain synovial fluid for analysis. Patients with maple syrup urine disease cannot adequately metabolize branched-chain amino acids; therefore, offering parenteral nutrition with protein would continue the toxic exposure. In some cases, patients may not be able to tolerate oral feedings for weeks, and TPN becomes essential to prevent protein catabolism. Water-soluble vitamins include vitamin C and the B-complex vitamins (thiamine, riboflavin, niacin, pyridoxine, folic acid, cobalamin, biotin, and pantothenic acid). 486 Answers and Explanations 1. Before the advent of effective therapy, the outlook for patients with SLE was historically poor and uniformly fatal. Limb length discrepancy, which may cause limp and pain 4. Craniosynostosis 1. Newborns present with severe respiratory distress and scaphoid abdomen due to displacement of the abdominal contents into the thorax. Compared with OAE, BAER is a more accurate test, but it requires costly equipment, trained operators, and sometimes sedation. Steroids are indicated for patients who present with severe abdominal pain, poorly controlled joint pain, or nephritis. Laboratory findings include varying degrees of anemia and a blood smear showing polychromatic RBCs. 3. The answer is A [, section XVI.B]. 94. Fever and irritability 2. Blepharitis is eyelid inflammation. Not only will this improve oxygen-carrying capacity, but this will contribute to iron repletion. Frequency averages 4×/day during the first week of life, 2×/day by 1 year of age, and 1×/day by 4 years of age. It is typically used for 2– 3 months if the diagnosis is made by 6 weeks of age. Two α-globin genes are deleted. Hypogonadism 11. Shaken baby syndrome may occur in a child younger than 2 years of age who is violently shaken. The differential diagnosis is extensive and is listed in Table 17-3. CO interferes with oxygen delivery and utilization. The usual cause is S. Family history, especially parental growth and pubertal histories, is important. Fetal sonography is highly sensitive in detecting spinal defects. It is important, however, to be aware that the age of onset and subsequent course of hormonal and physical changes during puberty are variable. A lecithin-to-sphingomyelin (L:S) ratio greater than 2:1 and the presence of phosphatidylglycerol (a minor phospholipid in surfactant) are indicators of fetal lung maturity. Pulmonology. Diagnosis of MCAD is made by the presence of abnormal urine organic acids and an abnormal acylcarnitine profile. Case based reviews questions and their answers at the end of all chapters that makes it easy to revise things and test yourself. Pathophysiology of GERD 1. Human-to-human transmission has been described. See section IV.D.3 for management of adrenal crisis. Febrile seizures do not require inpatient observation. Irritability and developmental regression are common in infants and toddlers. Synovial fluid with a WBC count > 50,000–100,000 cells/mm^3 with neutrophilic predominance is suggestive of septic arthritis. Hemostasis requires normal function of three important elements: blood vessels, platelets, and soluble clotting factors. Red flags indicating more serious pathology may include an abnormal neurologic examination, a new and unusual headache (especially when described as “a sudden worst headache of one's life”), a patient with an immunocompromised state, fever, or stiff neck, and blurring of optic disc margins on examination. Juvenile Dermatomyositis (JDM) A. Coma is a state of unawareness of self and environment in which the patient lies with the eyes closed, without purposeful movement or sleep–wake cycles, and is unarousable by external stimuli. Type 1 DM is an autoimmune disease due to genetic and environmental factors. Antibiotics are indicated for severe, acute necrotizing pancreatitis. The most common of these germinomas are yolk sac tumors (60%), followed by teratomas (15%) and, rarely, seminomas and embryonal carcinoma. Enzyme-linked immunosorbent assay (ELISA), which has a relatively high sensitivity. Acute cerebellar ataxia of childhood D. This patient's presentation is most consistent with which of the following diagnoses? Older children and adults are more likely to have symptomatic infection. Children typically present between 2 and 5 years of age with gait problems and weakness, and they are often wheelchair dependent by around their 12th birthday. The examination is performed by placing the hips in flexion at 90° with the feet flat on the table, heels apposed to the buttocks, while the infant is supine. A prolonged, throbbing, unilateral headache starts in the supraorbital area and radiates to the occiput. These children are able to sit and walk. Seizure Disorders of Childhood VI. This is called “postural roundback,” and it is managed with physical therapy and postural exercises. Disease prevention a. Celiac disease is most common in regions where wheat is a staple in the diet, and associated human leukocyte antigen (HLA) genotypes are found (e.g., North America, Middle East, Europe, Australia). Cushing syndrome. Bilateral involvement is common. Cardiac complications are quite rare and may include heart block and myocarditis. Etiology is generally unknown, but a lead point such as Meckel diverticulum, polyp, intestinal duplication, Peyer patch, or lymphoma may act to draw the proximal intestine inward. Examination of a comatose 13-year-old boy in the emergency department is significant for bilaterally nonreactive and dilated pupils. Adaptation of the intestine depends on the length and quality of the residual small bowel and on the presence of an intact ileocecal valve. When prescribing medication, health care professionals are advised to consult the product information sheet (the manufacturer's package insert) accompanying each drug to verify, among other things, conditions of use, warnings, and side effects and identify any changes in dosage schedule or contraindications, particularly if the medication to be administered is new, is infrequently used, or has a narrow therapeutic range. The etiology is unknown. Hypersensitivity Disorders V. Pigmentary Disorders VII. Opacification of the lens (cataract), severe uncorrected refractive error, significant differences in refractive errors between the eyes (anisometropia), and vitreous opacities (hemorrhage) all lead to poor visual stimulation, and therefore abnormal visual development. Tyrosinemia type I C. Dental history. An anterior–posterior radiograph of the neck will show a “steeple sign” characteristic of subglottic narrowing, while a “thumbprint sign” on lateral neck radiograph is associated with epiglottitis. Noninfectious Disorders of the Respiratory Tract Review Test Answers and Explanations Chapter 10 Gastroenterology I. β-Thalassemia is the result of mutations of the β-globin chain. Downward lens subluxation C. Psoriatic arthritis (see also Table 16-4). E —ear anomalies, including abnormal outer ear shape, hearing loss, and ossicular and temporal bone abnormalities 7. Abnormal muscle biopsy (used only when diagnosis is not certain) E. Fatal drowning refers to a person who dies any time as a result of drowning. The most common cause of pneumonia in older children and adolescents is atypical infection with Mycoplasma pneumoniae and Chlamydia pneumoniae. Patients are unable to sustain protrusion of the tongue (chameleon tongue). Weakness is often manifested as an inability to move the limbs against gravity. Causes are multiple. Incidence is approximately 1 in 10,000 live births, with a male predominance. Sudden infant death syndrome, or SIDS, prevention (“back to sleep”), for example 2. It is important to realize that the growth spurt in boys occurs late in puberty as compared with girls in which the growth spurt occurs early in puberty. When central hypotonia is suspected, it is necessary to consider: a. Without treatment, around 85% die within the first year of life. On examination, you note a temperature of 39.2°C (102.6°F) and a salmoncolored maculopapular rash on the trunk. Excessive bleeding after trauma may occur. The narrow pedicle, which suspends the small bowel and the base of the superior mesenteric vessels, can easily twist, leading to ischemia and midgut volvulus. Protein intake should be at the RDA for age. The differential diagnosis is extensive and includes congenital or traumatic musculoskeletal problems such as fractures; infections such as osteomyelitis or pyelonephritis; neoplasms; and rheumatologic conditions. Premature closure of the metopic suture leads to a triangular-shaped head and prominent longitudinal central ridge on the forehead (termed trigonocephaly). Glycogen storage disease type 1 D. This infant is presenting with nasolacrimal duct (NLD) obstruction. The infant is admitted to the neonatal intensive care unit, but later he dies of Escherichia coli sepsis. Placement of a gastrostomy tube often accompanies a Nissen fundoplication in infants. Death usually occurs by 5 years of age. The patient has one unprovoked seizure and the probability of further seizures occurring over the next 10 years (i.e., the patient has another diagnosis that predisposes to an increased risk of seizures, such as a brain tumor) c. The Hypotonic Infant A. Patients with Down syndrome are at risk for both 763 80. Evidence suggests that activated charcoal improves clinical outcomes, especially if given within 1 hour after an ingestion. Congenital torticollis is commonly associated with asymmetry of the head and face (plagiocephaly). Replacement of vitamins A, D, E, and K is necessary. 192 XI. Becker muscular dystrophy E. Antinuclear antibodies are likely to be positive. Which of the following statements is correct regarding coccidioidomycosis? Reactive arthritis is triggered by an enteric or sexually transmitted pathogen (classically, Chlamydia trachomatis). Clinical presentation provides a clue to the diagnosis, and no laboratory or imaging study is diagnostic. Instead, any increased stool losses should be replaced on a “milliliter per milliliter” basis. Enteroviral meningitis is characterized by low-level pleocytosis, with normal chemistries and a negative Gram stain. Dandy–Walker malformation X-linked hydrocephalus Infantile botulism Spinal muscular atrophy type 1 (Werdnig–Hoffman disease) Juvenile myasthenia gravis Congenital myotonic dystrophy For each patient, select the most likely diagnosis. In most cases, no surgical intervention is needed for the treatment of a varicocele and reassurance only may be given. Tension headaches are rare in young children, and 473 therefore, other diagnoses (e.g., migraines) should be preferentially considered. coli can occur at any age (see Chapter 7, Table 7-6). Enteral therapy. However, 95% have a chromosomal translocation between chromosomes 11 and 21 (a similar translocation to that noted in PNET brain tumors). The outlook is guarded. Patients with infantile botulism have a symmetric descending paralysis. Kyphosis, stiff joints, and joint contractures 4. Constipation from GI smooth muscle dysfunction e. S. DKA when ill or noncompliant 4. Neurologic examination is normal. Early introduction of cow's milk can replace the need for vitamin D supplementation. Plagiocephaly 1. Organizational skills training and play therapy have not been shown to be effective as a first-line treatment at this age. Investigation for the causes of CKD 2. Ewing sarcoma is a sarcoma characterized as a small, round, blue cell tumor (undifferentiated, monomorphous cell appearance). Some patients have associated otitis media. Children younger than 3 years 699 2. The answer is D [IV.E.3]. pertussis, with lower rates of vaccineassociated fever, seizures, and local reactions. The daily recommended intake of vitamin D in the first year of life is 200 IU. Foreign body aspiration may cause stridor or cough and should be considered as a cause, because in as many as 50% of foreign body aspirations, choking is not witnessed. Potter syndrome a. All of these causes are independent of the HPGA. Infants born to mothers taking phenytoin during pregnancy are at increased risk for specific malformations. Hypercortisolism. CRP is usually elevated. Delay in securing the airway emergently could potentially lead to airway obstruction secondary to edema and inflammation. Diagnosis is on the basis of clinical presentation, findings on liver biopsy, and exclusion of other causes of cholestasis. Hepatitis A immune globulin, given as “predeparture” passive immunization to some visitors to high risk areas C. An LP is necessary if meningitis is suspected at any age. Local massage, if there is no evidence of infection b. Corticosteroids are used when cholinesterase inhibitors fail. 11. The oxygen–hemoglobin dissociation curve is shifted to the left. Skin involvement 1. Although previously recommended, low-dose prophylactic antibiotics to decrease the risk of urinary tract infection in low risk patients is not indicated. Amoxicillin–clavulanate D. Clinical findings are similar to those of STEC-HUS. The healthier the child appears, the more likely the pain is functional. This congenital obstruction of the jejunum or ileum is caused by a mesenteric vascular accident during fetal life. DDAVP induces vWf release from endothelial cells and is used for mild to moderate bleeding and for prophylaxis before surgery. Most infants will not ingest this volume until 4 months of age or older. Diagnosis may be made by increased urinary glycosaminoglycans, by enzyme assays, and by genetic testing. A 3-year-old girl is brought to your office by her parents, who report that she has had 2 months of intermittent high-spiking fevers with temperature up to 39°C (103°F), which occur nightly and return quickly to normal. You admit him for intravenous rehydration because of his dehydration. Defects in Amino Acid Metabolism VII. Intellectual disability and neurologic defects, including ataxia 2. Order a stat head computed tomography scan. The lungs are clear except for mild wheezing at the bilateral lung bases. Anticipatory Guidance Review Test Answers and Explanations Chapter 2 Behavioral and Developmental Pediatrics I. Initial symptoms may be subtle, with abdominal pain or decreased growth being the only findings. Females with full mutation may have behavioral problems and developmental delays, but most have normal intelligence quotient (IQ). Infants with spinal muscular atrophy have hypotonia but also have characteristic tongue fasciculations. These murmurs are unaffected by the patient's position. The differential diagnosis of hematuria is outlined in Figure 11-2. Spine A. You suspect that the infant has a urinary tract infection (UTI). Stool evacuation is necessary to begin recovery, and is performed using polyethylene glycol solutions, magnesium salts, mineral oil, enemas, or manual disimpaction. Incidence is 1 in 5000–10,000 live births, with a male predisposition. If more than 100 mg/kg of iron is ingested 6. Most states screen for sickle cell anemia and other hemoglobinopathies, because early intervention in a comprehensive treatment program significantly decreases morbidity and mortality. Absence of the expected cortisol suppression seen in an overnight dexamethasone suppression test (i.e., dexamethasone given in the evening normally suppresses the following morning's physiologic rise in cortisol) 4. There are two clinical types of protein intolerance: a. Vision screening for ophthalmologic disorders begins at birth and is detailed in Chapter 18. During a well-child examination for a 12-year-old female, her mother refuses the human papilloma virus (HPV) vaccine, stating that her daughter “does not need the vaccine now because she is not going to be sexually active for a very long time.” Which of the following statements about HPV and the vaccine is true? Management includes fusion of C1 and C2 if instability is severe. Lumbar lordosis then develops. often referred to as a brief resolved unexplained event (BRUE). The onset of disease is in early infancy when the infant presents with a hyperactive startle and loses eye contact. Surgery may be required if the diagnosis is made beyond 6 weeks of age, if the hips are bilaterally dislocated, if the hips are not reducible on physical examination, or if the Pavlik harness fails to stabilize the hip. Pathogenesis. Replacement of normal diet with commercial enteric formulas has been shown to induce and maintain remission in many children with CD and is in wide use worldwide. Symptoms and signs: petechial conjunctival hemorrhage, preauricular lymphadenopathy, and a pseudomembrane along the conjunctiva 2. Antistaphylococcal antibiotics are given to patients who are suspected to have infection. Blood pressure monitoring and management are critical. Parainfluenza virus is the most likely cause. 545 XI. Patients with the acute onset of hyponatremic or isonatremic dehydration generally have their fluid and electrolyte deficits replaced over 24 hours. Autosomal dominant and autosomal recessive Alport syndrome may also lead to ESRD. A 6-year-old girl is brought to the clinic for a routine health maintenance visit. Treatment of early disease includes oral doxycycline for children ≥8 years of age, oral cefuroxime or oral amoxicillin. The wrist is held flexed and hyperextended at the metacarpal joints (choreic hand). Ataxia is a disturbance in smooth coordination of movements and often manifests as unsteady gait. Inflammation of the joints triggered by an infection, typically an enteric or sexually transmitted pathogen. Because you cannot read long books in few days and you would not be able revise all of the topics. If peritonitis is suspected, paracentesis for Gram stain, culture, and cell count of the ascites fluid is indicated. He has been previously well, and inspection of his immunization records reveals that all are up-to-date for his age. These two diseases involve different regions and different functions of the factor VIII molecule. Syverson, MD, Pediatric Rheumatology, Assistant Clinical Professor of Pediatrics, Sanford Children's Hospital, University of North Dakota School of Medicine, Fargo, North Dakota Brian G. | Philadelphia : Wolters Kluwer, [2019] | Series: Board review series. This patient likely has bacterial tracheitis and should be started on antistaphylococcal antibiotics. Studies show RBC fragmentation seen as “burr” cells, “target” cells, and irregularly shaped cells on the blood smear, along with thrombocytopenia. This patient had an apparent life-threatening event (ALTE) now more 358 8. All patients with short stature who are more than 2 SDs below the mean or who have a growth velocity less than 5 cm per year are considered to have a pathologic growth disorder until proven otherwise. A 6-month-old female infant with a 2-week history of vomiting is brought to your office by her parents. The VUR is likely caused by a short submucosal tunnel in which the ureter inserts through the bladder wall. In adulthood, typical myotonic features include myotonic facies (atrophy of masseter and temporalis muscles), ptosis, a stiff, straight smile, and an inability to release the grip after handshaking (myotonia). Watanabe, MD, FAAP, Pediatric Liver and Intestinal Transplantation, Children's Hospital Los Angeles, Los Angeles, California, Associate Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Lauren J. The pathophysiologic mechanism is unknown. Although diphenhydramine and corticosteroids may be useful in severe allergic reactions, epinephrine is the principal treatment. It declines and gradually disappears by 6–9 months of age. Urinary Tract Infection (UTI) Review Test Answers and Explanations Chapter 12 Neurology 6 I. Chromosomal disorders 1. Anatomy and Physiology of the Respiratory System II. Which of the following statements regarding VUR is correct? Table 5-3 Typical Clinical Features of Inborn Errors of Metabolism (IEM) General symptoms: Lethargy or coma Poor feeding Intractable hiccups Unusual odor (especially when acutely ill): Odor: Mousy/musty Sweet maple syrup Sweaty feet Boiled cabbage Neurologic: IEM: Phenylketonuria Maple syrup urine disease Isovaleric or glutaric acidemia Tyrosinemia type I 181 Hypotonia Unexplained developmental delay Unexplained and difficult to control seizures Ophthalmologic: Cherry-red macula, cataracts, or corneal clouding Gastrointestinal: Unexplained vomiting Hepatomegaly Splenomegaly Metabolic: Hypoglycemia may be associated with fatty acid oxidation disorders or carbohydrate disease Elevated NH_3 without acidosis is suggestive of urea cycle defects Elevated NH_3, metabolic acidosis, and elevated anion gap are suggestive of organic acidemias Urinary ketones in a newborn are unusual and may indicate an IEM such as maple syrup urine disease NH_3 = ammonia. Which of the following is the best serum laboratory screen for the most likely diagnosis? FFR management is based on cleaning out the fecal mass, softening the stool, and educating the patient and family. Appropriate fluids for use in the emergency phase include isotonic crystalloids, such as normal saline or lactated Ringer solutions in boluses of 20 mL/kg. Complications of ventriculoperitoneal shunts include shunt infection and shunt obstruction. a. Atlantoaxial instability is an unstable joint high in the cervical spine and does not manifest as congenital torticollis. Headaches that occur on awakening in the morning are more characteristic of headaches resulting from increased intracranial pressure. An otherwise healthy newborn who develops an acute severe illness within the first few hours to weeks of life b. The early onset of gonadotropin-mediated (i.e., mediated by FSH and LH) puberty. Taking a prenatal multivitamin preparation that includes folic acid has decreased the incidence of SB in the newborn. Hematuria is defined as the presence of red blood cells (RBCs) in the urine. The differential diagnosis of FTT is outlined in Figure 1-1. DTaP vaccine 1. 539 VII. Ninety percent of poisonings are accidental. Treatment is the same regardless of whether the drowning occurred in salt or fresh water. Mutations can be inherited or sporadic. Acanthocytosis of erythrocytes (abnormal star-shaped red blood cells) is seen in abetalipoproteinemia. Recurrence risk decreases with increasing patient age. This patient has a high likelihood of developing carditis. Acute cerebellar ataxia is the most common cause of ataxia in children. With the abovementioned syndromes, a high index of suspicion is important because spinal cord injury may occur with dislocation or significant instability. Patients may be asymptomatic or have hematuria, UTI, or nephrolithiasis. Diagnostic studies include low serum copper and ceruloplasmin, as well as low copper in tissues. She is often reluctant to walk because of swelling of her ankles and knees. Staphylococcus aureus C. Clinical features include lung hypoplasia, abnormal limb positioning, and characteristic “Potter facies” (compressed facial appearance, with large ears flattened against the head). Epileptic syndromes are epileptic conditions characterized by a specific age of onset, seizure characteristic, and EEG abnormality. However, in SCFE, the Klein line may not cross the epiphysis at all. Electrolytes demonstrate hypernatremic, hyperchloremic metabolic acidosis. Spina bifida (SB) is a general term that refers to any failure of bone fusion in the posterior midline of the vertebral column. Nontraumatic causes include iris neovascularization (associated with diabetes mellitus, intraocular tumors, and retinal vascular diseases), clotting disorders, and iris tumors (e.g., melanoma, juvenile xanthogranuloma). 5α-Reductase deficiency. Dogs (80% of bites), cats, rodents, other wild or domesticated animals, and humans (2– 3% of bites) may all cause a bite injury. These small, mobile, and painless nodules occur on the bony prominences of the extensor surfaces of the extremities. Diagnosis is based on demonstration of deficient enzyme activity and/or by genetic testing. Systemic Lupus Erythematosus (SLE) V. 353 FIGURE 9.3 Features of cystic fibrosis. Hgb and age a. Activated charcoal should be considered for all poisonings. The hyperoxia test helps to distinguish between respiratory and cardiac causes of cyanosis. Frequent follow-up is essential, and growth velocity, physical examination, bone age, and laboratory tests (17-OHP, PRA, androgens, etc.) should be monitored carefully. However, the iron level in β-thalassemia minor is normal or elevated. Radiographs are of limited usefulness but may reveal dilated loops of the bowel, or pneumoperitoneum if perforation has occurred. Any pathologic condition that causes abnormal retinal stimulation can cause amblyopia. There is a large spectrum of phenotypes, with some patients who are largely asymptomatic and others who are transfusion-dependent starting in infancy. Perkins, MD, PhD, FAAP, Associate Dean for Graduate Medical Education, Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Robert K. Treatment options include corticosteroids, haloperidol, valproic acid, or phenobarbital. Fixed kyphosis beyond 50° is considered pathologic. For a child 6–12 months of age, an LP should be seriously considered if the child is deficient in vaccination for Haemophilus influenzae type b (HIB) or Streptococcus pneumoniae or when immunization status cannot be determined. An autosomal dominant disorder caused by mutations in FGFR3. Acyanotic Congenital Heart Disease IV. Up to 20% of survivors will develop permanent disabilities (e.g., loss of limbs, deafness, central nervous system damage). Testicular enlargement is usually the first sign of puberty (≥4 mL as measured with an orchidometer or >2.5 cm in length). NLD obstruction is characterized by watery eye with increased tear lake, matted eyelashes, and often mucous in the medial canthal area. Betaine therapy (lowers homocysteine levels and reduces risk for thromboembolism) c. Prognosis is especially poor if metastases are present at diagnosis. The answer is C [, section IV.I]. This clinical presentation is consistent with infantile spasms. Thorough physical examination should include a funduscopic examination, assessment for midline defects (central incisors, cleft palates, etc.), assessment of thyroid size, evaluation for stigmata of genetic syndromes (e.g., web neck, shield chest, and abnormal carrying angles are suggestive of Turner syndrome; see Chapter 5, section IV.C.1), scoliosis screening, and Tanner staging (see Chapter 3, section I.A.2). Type of vaccine: inactivated vaccine c. The likely diagnosis is cystic fibrosis (CF). The classic electrolyte presentation is a hyperchloremic metabolic acidosis with a normal serum anion gap. Initial steps include a detailed history and physical examination, serum electrolytes and glucose, arterial blood gas (ABG; ±100% oxygen test—see below), complete blood count (CBC), and chest radiograph (CXR). Lower GI bleeding 1. Clinical findings may include short stature, anemia, failure to thrive, polyuria and polydipsia, lethargy, and rickets. Treatment depends on the age of the patient at the time of diagnosis. Child abuse includes physical abuse, psychological abuse, neglect, and sexual assault. Studies show severe hypochromia and microcytosis, elevated reticulocyte count, target cells and poikilocytes (abnormally shaped red blood cells) on the blood smear, and elevated unconjugated bilirubin, serum iron, and lactate dehydrogenase (LDH). Spinal muscular atrophy (SMA) a. Pulmonary surfactant is the surface-active material that decreases alveolar surface tension and prevents atelectasis. Dietary counseling may be required to increase nutritional iron through ironrich foods (e.g., meats, legumes, prunes, iron-fortified cereals). Chemical conjunctivitis is the most common cause of red, watery eyes in the first 24 hours of life. After the first year of life, individuals with Hurler disease present with developmental delay, coarse facies, corneal clouding, and dysostosis multiplex. Acute cerebellar ataxia rarely occurs in children older than 10 years. History is critical to confirm abuse. Hip IV. Nyong'o 640 I. Generally observed as a sporadic occurrence in an otherwise healthy family. Characteristic cutaneous findings, particularly over sun-exposed areas, include the following: a. Babies born to mothers with these antibodies are at risk for neonatal lupus. The answer is C [V.F.2]. Spondylolisthesis occurs when the vertebral body involved in the spondylolysis slips anteriorly (i.e., subluxation of the vertebra). The parents deny any travel or medication use. Up to 10% of deliveries occur beyond 42 weeks of gestation. Hepatoblastoma is the most common type of liver tumor in childhood. 115. Hypogonadism is absent in both disorders. Ultrasound is the best imaging screen and demonstrates fluid in the joint capsule. STEC-HUS is the most common subtype seen in childhood. Erythropoietin levels are high. This patient's clinical presentation and electroencephalogram (EEG) are consistent with absence epilepsy of childhood. Appropriate bolus fluids in the emergency phase of intravenous rehydration should include 20 mL/kg of one half–normal saline solution. Factor IX deficiency—hemophilia B (Christmas disease). IEM are a heterogeneous group of diseases that can present in a variety of ways. | Coller, Ryan, editor. Figure 13-1 presents the differential diagnosis of anemia on the basis of the previously discussed classification schemes. It is necessary to determine if the etiology is organic or whether FFR exists. The answer is C [, section II.D.6]. Classification. Down syndrome is also associated with mental retardation; however, the facial characteristics include upslanting palpebral fissures, epicanthal skin folds, and a protruding tongue. The diagnosis is often made by prenatal fetal ultrasound, noting the abnormal position of the stomach in the thorax and rightward deviation of the heart (in cases of left CDH). Disadvantages: it is only available for children 2 years of age or older without contraindications to the vaccine. A small pseudocyst may resolve on its own. DNA testing to identify the gene can be performed to confirm the diagnosis. Constitutional symptoms are common and include fatigue, anorexia, malaise, low-grade fevers, and weight loss. The book is dense with information, but the bullet-point format and tables help keep it organized. Other manifestations include a. Ideally, the history should be obtained with open-ended questions from an interviewer trained in sexual abuse evaluation. Abnormal muscle innervation, breastfeeding, early administration of oral erythromycin, and decreased production of nitric oxide synthase have all been implicated. Antenatal history may reveal decreased fetal movements and polyhydramnios caused by poor swallowing in utero. Toxic intermediates produced when glutathione is depleted bind directly to hepatocytes, causing hepatocellular necrosis. SMA therefore presents with hypotonia, difficulty feeding, and tongue fasciculations. Necrotizing enterocolitis (NEC) 1. Other types of scoliosis are neuromuscular (associated with cerebral palsy) or syndromic (associated with skeletal dysplasias and connective tissue disorders). Meningococcal vaccine (MCV) a. Cholestasis occurs in 1 in 10,000–15,000 live births, and more than 50% are caused by neonatal 391 hepatitis or biliary atresia [see sections XI.D.2 and XI.D.3]. Congenital diaphragmatic hernia (CDH) 1. Macrocephaly, long face, prominent jaw, protruding ears. Surgery is necessary to drain and debride a subperiosteal abscess if fever and swelling persist despite 48 hours of intravenous antibiotics. Gait and cognition are not affected. Clinical features of pathologic reflux (GERD) 1. Bone Mineral Disorders XI. Prognosis is best in younger patients. Disease of the rectum only is called ulcerative proctitis. For recessive conditions, there may be an affected sibling, and for X-linked disorders, there may be an affected brother or maternal uncle. Infants present with brisk deep tendon reflexes. Fever of Unknown Origin (FUO) IV. Nasolacrimal duct (NLD) obstruction 1. Young children are typically bitten on the head and neck, whereas older children are bitten predominantly on the extremities. Plants (6–7% of all ingestions) g. Appendicitis is obstruction and inflammation of the appendix. It resolves spontaneously without treatment in over 80% of cases by age 9–12 months, and so observation only would be the most appropriate management at this time. Clinical features include abdominal distension, lack of meconium passage, vomiting, and bowel perforation. Epidemiology: the most common of the UCDs b. Cosmetics and personal care products (most common toxic exposure) b. Inflammatory disorders of the intestinal mucosa, such as Crohn disease (CD) [see section IX] and colitis, may also result in protein loss. Maintenance therapy most often includes an osmotic laxative (such as polyethylene glycol) to soften and lubricate the stool. Stool calprotectin is a very good screen for IBD. Progressive weakness leads to loss of ambulation before 12 years of age. She is therefore at risk for severe and disseminated disease. Clinical features a. Which of the following statements regarding the suspected diagnosis is correct? Evaluation 1. The risk of abuse is greatest, however, in children with the following characteristics: 1. Teratomas are tumors containing more than one of the three primary germ cell layers (i.e., ectoderm, mesoderm, and endoderm). Abdominal ultrasound and liver biopsy are performed in rapid sequence to rule out other causes of cholestasis. To transmit Lyme disease, an infected tick must be attached to the skin for at least 36-48 hours. Rheumatic fever is a delayed, nonsuppurative, autoimmune complication occurring 2– 4 weeks after an upper respiratory infection with group A β-hemolytic streptococcus (GABHS) (Streptococcus pyogenes). Flaccidity or no movement may suggest severe spinal or brainstem injury. Nonabuse causes of retinal hemorrhages include birth trauma, leukemia, increased intracranial pressure, malignant hypertension, bacterial endocarditis, immune thrombocytopenic purpura, and rarely, cardiopulmonary resuscitation. Meningocele requires surgical repair. Periorbital violaceous heliotrope rash, which may also cross the nasal bridge b. 2. Prognosis of OTC deficiency is variable, depending on the severity of hyperammonemic episodes. 4. Individuals with Niemann–Pick disease may present with hepatomegaly and seizures, but hypoglycemia and E. On examination, you note that he appears very ill and prefers to sit leaning forward on his hands with his neck hyperextended. For example, if a boy has a father who is 5 feet 9 inches in height and a mother who is 5 feet in height, then the MPH is 5 feet 7 inches ± 4 inches. A gastrostomy tube also enables venting of the stomach in cases of uncomfortable bloating. The prognosis is generally favorable and depends on the severity of the presentation. Chlamydia trachomatis infection presents at 4–10 days of life, often with purulent or serous discharge. In severely cyanotic infants, intubation and mechanical ventilation may be necessary until a diagnosis is made and definitive treatment is initiated. 638 CHAPTER 18 639 Ophthalmology Omondi L. Congenital glaucoma is very different from adult glaucoma. Porphobilinogen D. Decorticate posturing (flexion of arms and extension of legs) is seen in cortical and/or subcortical abnormalities with preservation of brainstem function. The displaced and angulated supracondylar fracture in this patient is an orthopedic emergency because of the risks of compartment syndrome and neurovascular injury. Diagnosis is made by the following: 1. The examination describes a healthy child with a benign family history. Peak ages are younger than 5 years and during adolescence. Patients with Turner syndrome have left-sided heart lesions (e.g., coarctation of the aorta). More than 750,000 people die annually from hepatitis B and its complications. RBC = red blood cell; U/A = urinalysis. These patients usually present at 4–5 years of age. There are three different subtypes of HUS, which differ in their known etiology, treatments, and prognoses: Shiga-like toxin HUS, pneumococcal HUS, and atypical HUS. Hyperoxia test in infants with heart disease. Significant colony counts depend on the culture method: a. This patient's condition is inherited in an autosomal recessive pattern. Normal Visual Development and Amblyopia A. Immunosuppressive medications 1. On physical examination, a truncal macular rash is evident, and a grade 3/6 loud holosystolic murmur is audible at the apex and axilla. Visual development 1. Tang, MD, FAAP, Developmental-Behavioral Pediatrics, Palo Alto Medical Foundation, Adjunct Clinical Assistant Professor of Pediatrics, Stanford School of Medicine, Stanford, California Jessica Tenney, MD, FAAP, FACMG, San Francisco, California Division of Medical Genetics, Assistant Professor of Pediatrics, UCSF School of Medicine, Rujuta B. Login Sign Up Forgot your password? Bagged urine cultures, especially in uncircumcised infants, are unreliable. Precocious puberty 1. Patients with grade 4 or 5 VUR should be referred to a pediatric urologist for consideration of ureteral reimplantation. May be associated with congenital muscular torticollis (see Chapter 17, section II.A.1), in which fetal head constraint during the third trimester can result in both trauma to the sternocleidomastoid and deformations of the skull. In these cases, the biliary system becomes overwhelmed by the increased bilirubin load. Blepharitis is one of the most common causes of red eye. A normal cerebrospinal fluid (CSF) profile usually has less than 10 white blood cells (WBCs). Growth is closely monitored, and patients may require recombinant human growth hormone if their growth fails to normalize with other medical interventions. Surgery to remove the disk is necessary only if symptoms persist or if the 614 neurologic examination is abnormal. Dark urine 4. Duodenal atresia and stenosis a. Therefore, their use is recommended only after the diagnosis of rheumatic fever is certain. An autosomal dominant disorder caused by mutations in the gene that codes for fibrillin. Hypoglycemia XIV. aureus infection. The remainder of the examination is normal. The precise cause of ROP is unknown; however, vascular endothelial growth factors are posited to be involved. Children are most susceptible to amblyopia during the first 3–4 months of life, the 642 period of critical visual development. Signs and symptoms include abdominal mass, abdominal pain caused by torsion of the tumor together with the ovary, and vaginal bleeding. Fever and irritability are common. OTC deficiency a. Henoch–Schönlein purpura Psoriatic arthritis Systemic-onset juvenile idiopathic arthritis Reactive arthritis Inflammatory bowel disease-associated arthritis Systemic lupus erythematosus Kawasaki disease Dermatomyositis Oligoarticular juvenile idiopathic arthritis Rheumatic fever Lyme disease For each patient, select the most likely diagnosis. Scoliosis is a fixed lateral curvature of the spine of greater than or equal to 10°. Malnutrition 1. CBC may show an elevated hematocrit as a result of hemoconcentration resulting from 425 the hypoproteinemia. Typically, patients with ankylosing spondylitis are human leukocyte antigen B27 (HLA-B27) positive and have involvement of the hips and sacroiliac joints. Vaccination is recommended for both males and females to decrease the contagious risk and to lower the risk of complications, including cervical, anal, oropharyngeal and penile cancers. PCV13 is composed of 13 pneumococcal serotypes. Type of vaccine: oral, live vaccine c. Classification 1. Takayasu arteritis. Decreased complement (C3 and C4) is seen especially with active disease and represents immune complex–mediated complement activation. Pancreatic lipase is a necessary enzyme that hydrolyzes triglycerides for emulsification. Causes may differ in boys and girls but, in general, include exposure to exogenous sex steroids (found in some skin lotions or foods), McCune– Albright syndrome, gonadal tumors, adrenal tumors, and nonclassic CAH; see section IV.C.3.a.(3). Essentials of Pediatric Nursing. The child in the description has suffered anaphylaxis, a severe, potentially life-threatening, systemic reaction. Laboratory studies to collect forensic evidence should be performed if the abuse occurred within 72 hours of presentation, and should include cultures or serologic testing for sexually transmitted infections (STIs), including human 702 immunodeficiency virus (HIV). Clues suggesting an organic cause include delayed meconium passage (>48 hours after birth), onset of constipation during infancy, history of pelvic surgery, encopresis before 3 years of age, and inability to toilet train. It may be the result of infectious or noninfectious causes. Management includes oral macrolide antibiotics. Malrotation and midgut volvulus 1. It is most common in children 5–15 years of age, reflecting the age group most susceptible to streptococcal throat infections. The cause is herpes simplex virus type 1 (HSV-1) and typically represents the initial exposure to HSV-1 virus. Usually, the child will start to use the arm within 15 minutes of the reduction, but use of the arm may occasionally be delayed for up to 24 hours. Outcome depends on surgical resectability and chemosensitivity. On examination, you note weakness in his lower extremities and absent lower extremity deep tendon reflexes. On the other hand, prepubertal patients whose HPGA has not yet been activated, and patients with peripheral precocious puberty (PPP) in which peripherally produced sex steroids suppress pituitary gonadotropin secretion, would be expected to have a flat response (i.e., no increase in LH secretion) on injection of synthetic GnRHa. c. 22. Quick Relief Medication: SABA as needed for relief of symptoms. Peak incidence occurs at 5–9 months of age. Clinical features include fever and pain, induration, tenderness, warmth, and erythema of the involved area. Iron is given with vitamin C (e.g., orange juice) to enhance intestinal iron absorption. Crigler–Najjar type 2 is an autosomal dominant disorder in which 90% of enzyme activity is absent. h. She has had headaches three times per week for the past several months, and her parents are concerned that she may be having migraines. The content of the well-child visit includes 1. 481 X. Mycophenolate, in particular, is being used more often as an alternative to cyclophosphamide to treat lupus nephritis. Lactose breath hydrogen testing to rule out lactose intolerance is a reasonable option, because lactose intolerance may be present in as many as 10% of otherwise asymptomatic children. Management a. Over 80% of cases resolve spontaneously within 9–12 months. Disorders of the Adrenal Gland V. Short stature is discussed in Chapter 6, section I. Patients can be transitioned to oral antibiotics once the child has shown clinical improvement and sensitivities are available. Without functional cholesterol biosynthesis, steroid hormone production is defective. RBC casts are diagnostic of glomerular bleeding, which usually occurs in acute or chronic glomerulonephritis. The seizure lasts 38°C), in whom CNS infection, metabolic imbalance, and history of afebrile seizure are excluded. There are two types of vaccines. Which of the following is correct regarding the cause and management of the probable diagnosis? ORT may be an effective, safe, and inexpensive alternative to IV rehydration therapy. Inflammation of the gallbladder with transmural edema that may be associated with gallstones or, less commonly, without stones (termed acute acalculous cholecystitis) b. Antibiotic treatment of streptococcal pharyngitis or impetigo does not reduce the risk of poststreptococcal glomerulonephritis, although the risk of rheumatic fever is reduced. Clinical features include a ruddy facial complexion with a normal-sized liver and spleen. Abdominal CT scan is useful in diagnosing 377 complications, such as abscess, necrosis or pseudocyst (described below). SCFE is bilateral in 30% of cases, and patients with hypothyroidism are especially more likely to develop bilateral disease. Jaundice occurs in 5% of patients, and it is associated with low levels of the enzyme glucuronyl transferase. Autosomal dominant and X-linked mutations have been found. Neither calcium nor magnesium abnormalities play a role in SCFE. Intussusception is the most common cause of bowel obstruction after the neonatal period in infants younger than 2 years. Young children may refuse to walk for fear of falling. Long-acting inhaled β_2-agonist as needed for symptom relief plus daily low-dose inhaled corticosteroid E. Biliary atresia is a progressive fibrosclerotic disease that affects the extrahepatic biliary tree. Hypoglycemia Review Test Answers and Explanations Chapter 7 Infectious Diseases I. Opiate overdose E. Vomiting of nonbilious, milky fluid starts during the second or third week of life. It became a hugely frustrating exercise. Objects used to burn may be branded to the skin (e.g., irons and cigarettes). Antidotes exist for a relatively small number of compounds (Table 20-3). CD may involve any segment of the GI tract, from the mouth to anus. Key point: All girls with short stature should have a karyotype, as the physical features of Turner syndrome can be subtle. Peripheral hypotonia is dysfunction of lower motor neurons (i.e., spinal motor neurons, peripheral nerves, and the neuromuscular junction). If the disease progresses or if peritonitis develops, cholecystectomy is indicated. Myopia. Elemental iron (4–6 mg/kg/day) is prescribed orally for mild to moderate anemia. Fulminant acute-type AIHA occurs in infants and young children and is preceded by a respiratory infection. FGR is a diagnosis made during gestation and is classified based on the pattern of intrauterine growth parameters. Type of vaccine 1. Tourette syndrome 1. Patients are usually normal at birth but will show features of this disorder in the first few years of life. Prognosis for those who do not respond to chemotherapy or relapse is very poor. Dog bites 1. Treatment requires chemotherapy and either surgery or surgery and radiation for local control. Fever and anorexia are usually present. Chronic infection does not occur, although the disease may relapse with return of symptoms in up to 25%. A direct Coombs test is weakly positive. If clinical suspicion is high, consider celiac testing. Congenital myotonic dystrophy is an autosomal dominant muscle disorder that presents in the newborn period with weakness and hypotonia. Rationale for vaccine: Diphtheria, tetanus, and pertussis all may cause serious disease, especially in young infants. The answer is E [, section IV.A and Table 18-2]. Assessment of growth and development b. Surgery may be required if there is no improvement within 3– 9 months. The mutation results in polymerization of Hgb within the RBC membrane when the RBC is exposed to low oxygen or acidosis. Bone pain, erythema, swelling, and induration, which may develop in the area over the osteomyelitis 3. The male newborn earns 1 point for a heart rate 200 repeats b. Fecal elastase has a high sensitivity and specificity to detect pancreatic insufficiency. Circumcision VI. LCH is characterized by infiltration of organs with Langerhans cells. Short-acting inhaled β_2-agonist as needed for symptom relief C. Infants with galactosemia develop vomiting and diarrhea after feeding with either breast milk or cow's milk–based formulas, because both types of feeding contain galactose. Physical examination should be comprehensive and also should focus on growth (i.e., length/height, weight, head circumference, and body mass index). Normal depth perception (stereopsis), similar to normal visual development, may be impaired because of the following: a. Improper eye alignment, such as uncorrected strabismus [see section IX] b. Kayser–Fleischer rings, which represent copper deposition in Descemet's membrane, are pathognomonic for Wilson disease. Renal Tubular Acidosis (RTA) X. Organic CAP is caused by a disease process or disorder and occurs in one-third of cases. Primary polycythemia (polycythemia vera) is an extremely rare cause of polycythemia during childhood. I'll definitely be returning to this book again and again. Clues suggesting FFR include the development of constipation after toilet training and identification of a sentinel event (e.g., change in psychosocial environment, inappropriate toilet training methods, abuse), in the face of continued normal growth 382 and development. Disadvantages: requires injection c. Coller I will forever be grateful for the support, mentorship, and encouragement by the late Dr. David Rimoin, a wonderful friend and leader who touched so many learners and patients. Diabetes Insipidus (DI) XII. Household contacts of immunodeficient patients should not receive OPV vaccine, as it is shed in the stool. Hepatitis B infection is caused by HBV, a DNA virus. Child development should correlate with the nature of the injury. Proteinuria IV. Patients may develop hepatic adenomas that are at increased risk for malignant transformation to hepatocellular carcinoma. Bleeding into the iliopsoas muscle may be especially severe as a result of delayed recognition of the bleeding and the potential for significant blood accumulation. In some children, the cause of seizures is known (Table 12-2); however, in many, the cause remains unknown. Genetic Disorders and Inborn Errors of Metabolism. Muscular dystrophy B. Turner syndrome D. Pituitary or hypothalamic involvement may lead to growth retardation, diabetes insipidus, hypogonadism, and panhypopituitarism. Treatment includes transfusions of RBCs and platelets as needed, and bone marrow transplant from an HLA-compatible donor, if available. Anaphylaxis to a vaccine or its constituents b. These ticks are especially prevalent in woodlands and fields in the New England states and parts of the Pacific coast and Midwestern United States. Unilateral ptosis or cranial nerve III palsy accompanies this headache. Symptoms may include jaundice, hepatomegaly, hepatic failure, and cirrhosis. Congenital microcephaly is associated with abnormal induction and migration of the brain tissue. Some patients have an inability to concentrate the urine or may present with hematuria (5%) during adolescence. Malrotation is common in patients with heterotaxy, a condition in which the internal organs are abnormally arranged in the chest and abdomen, and may be associated with small bowel atresia, Hirschsprung disease, and intussusception. Conjunctival cultures and Gram stain are not performed routinely for mild to moderate infections, and patients are usually treated empirically. The growth pattern should be normal, and no pubic hair should be apparent. (Consider GH deficiency in patients with a single central maxillary incisor or with cleft palate.) c. There is no single confirmatory test for Sydenham chorea. Testing for total serum hemolytic complement (CH50) will evaluate for normal functioning of the classic complement pathway (question 143). Four percent of patients with IBD present before 5 years of age. SB occulta is suggested by finding any skin abnormality overlying the spine and may be confirmed by spinal radiographs. Early onset of pubic or axillary hair growth without the development of breast tissue or enlarged testes. Atlantoaxial instability C. Outpatient management for older nontoxic children with suspected UTI may be appropriate. RBC transfusion may be required for severe anemia associated with cardiovascular compromise. Total colectomy is associated with infertility risk in females. Hirschsprung disease. Adolescent Health Screening III. Dystrophin levels are normal. 505 IV. The most common cause of hypercortisolism is iatrogenic, as a result of prolonged use of steroids [see section IV.E]. Sequelae of GERD a. Nausea and vomiting may occur. Pneumocystis jiroveci B. 475 D. Table 10-2 Organic Causes of Chronic Abdominal Pain Constipation Peptic ulcer disease Carbohydrate intolerance (e.g., lactose, fructose, sorbitol) Inflammatory bowel disease Pancreatitis Celiac disease Helicobacter pylori infection Parasitic infection (e.g., Giardia lamblia infection) Genitourinary disorders (e.g., pyelonephritis, hydronephrosis) Congenital structural abnormalities of the gastrointestinal tract (e.g., malrotation, intestinal duplication, hernia) 381 VII. Most vaccine side effects are mild to moderate in severity and occur within the first 24– 48 hours after administration (e.g., local inflammation and low-grade fever). Defects extrinsic to the RBC that cause hemolysis 1. Renal abnormalities, including renal tubular acidosis c. Down syndrome (see Chapter 5, section IV.B.1), skeletal dysplasias (Chapter 5, section IV.H.), and Klippel–Feil syndrome [see section II.A.3] are associated with atlantoaxial instability. Adult defecation ranges from 3×/day to 3×/week. Additional screening tests depend on the age of the child and may include lead level, hemoglobin, lipid levels, blood pressure, and hearing and vision assessment. Severe classic forms have developmental regression and death in childhood. Most patients should have a bone age radiograph, as well as laboratory testing for morning levels of 17-hydroxyprogesterone (17-OHP), as some patients with nonclassic congenital adrenal hyperplasia (CAH) may present with premature adrenarche. Pancytopenia may be congenital or acquired. Table 4-3 Causes of Fetal Growth Restriction Type I: Early interference with fetal growth from conception to 24 weeks' gestation Chromosomal anomalies (including trisomies 21, 13–15, and 18) Fetal infections (TORCH) Maternal drug use (chronic alcoholism, heroin, and cocaine) Maternal chronic illness (hypertension, severe diabetes mellitus) Type II: Intrauterine malnutrition from 24 to 32 weeks' gestation Inadequate intrauterine space (multiple pregnancies, uterine tumors, uterine anomalies) Placental insufficiency from maternal vascular disease (renal failure, chronic essential hypertension, collagen vascular diseases, pregnancy-induced hypertension) Small placenta with abnormal cellularity Type III: Late intrauterine malnutrition after 32 weeks' gestation Placental infarct or fibrosis Maternal malnutrition Pregnancy-induced hypertension Maternal hypoxemia (lung disease, smoking) TORCH = toxoplasmosis, other (syphilis), rubella, cytomegalovirus, herpes simplex virus. Vaccination during the preteen years is recommended to facilitate the development of an immune response before the initiation of sexual activity. General management principles 705 1. Approximately 80% of patients fully recover. There are no contraindications owing to underlying medical conditions, unless the child has a history of anaphylaxis to a previous influenza vaccination. Pharmacotherapy a. Reduction in stomach volume by the Nissen procedure requires a period of adaptation and expansion of the stomach that is best assisted with tube feedings. The infantile, juvenile, and adolescent forms are autosomal recessive and lead to ESRD in childhood or young adulthood, and there are often significant associated extrarenal manifestations in 10–15% of patients, including retinal degeneration, cerebellar vermis hypoplasia, occipital encephalocele, hepatic fibrosis, situs inversus, bronchiectasis, and skeletal defects. The clinical features of homocystinuria and Marfan syndrome overlap considerably; however, this patient most likely has homocystinuria based on the presence of cognitive delay and a marfanoid body habitus with fingers of normal length (i.e., no 198 arachnodactyly). Respiratory Distress VI. Cheyne–Stokes breathing (alternating apnea and hyperpnea) suggests bilateral cortical injury and is associated with herniation, specifically displacement of the diencephalon (thalamus and hypothalamus). Glycolytic enzymatic defects of RBCs include glucose-6-phosphate dehydrogenase deficiency and PK deficiency. After receiving at least one Tdap, the vaccine should be given every 10 years thereafter for continued protection. The answer is C [IX.E and Table 11-2]. Microcephaly 1. In addition, he has short palpebral fissures and a long, smooth philtrum with a thin upper lip. A sexually active 16-year-old boy with arthritis and conjunctivitis. Surgery is rarely required. To request permission, please contact Wolters Kluwer at Two Commerce Square, 2001 Market Street, Philadelphia, PA 19103, via email at or via our website at shop.lww.com (products and services). An enlarging subdural hematoma can cause uncal herniation with a unilateral dilated, nonreactive pupil. Cerebral infarct B. Severity of amblyopia depends on when the abnormal stimulus began, the length of exposure to the abnormal stimulus, and the severity of the blurred image. Passive movement of the elbow (examiner moves the extremity) can result in neurovascular injury and should not be performed if a supracondylar fracture is suspected. On physical examination, the child is well hydrated but has a large anterior and posterior fontanelle and a persistent downward deviation of both eyes. Polyhydramnios in utero in two-thirds of cases, due to inability of the fetus to swallow amniotic fluid b. Evaluation and diagnosis. Alcohol intoxication less frequently causes coma and more often leads to somnolence. Meconium Aspiration Syndrome (MAS) VIII. By injecting synthetic GnRHa into a patient, the LH response, and to a lesser degree the FSH response, can be used as an assessment of the activation of the HPGA. Same is the case with its pediatrics book. CNS = central nervous system; RDS = respiratory distress syndrome; MAS = meconium aspiration syndrome; PPHN = persistent pulmonary hypertension of the newborn; TTN = transient tachypnea of the newborn; IDM = infant of diabetic mother. Short-acting inhaled β_2-agonist as needed for symptom relief plus daily long-acting β_2-agonist. The patient is diagnosed with an epilepsy syndrome (e.g., childhood absence epilepsy) 3. The abdomen is not distended, and bowel sounds are present. Most cases are inherited in an autosomal dominant fashion. Boys are more likely to be affected. A trial of nonstimulant medication is also not a first-line treatment. Severe thoracic curves (>70°) can be associated with restrictive or obstructive lung disease. Congenital forms of RTA are caused by mutations in various transporters in the proximal or distal tubular cells. Pathophysiology and etiology. RTA should be considered in patients who present with a non–anion gap hyperchloremic metabolic acidosis. Klinefelter syndrome B. Genetic syndromes (e.g., Russell–Silver syndrome, Prader–Willi syndrome; see Chapter 5, sections IV.E.5 and IV.E.2, respectively) d. Scheuermann kyphosis is a rigid kyphosis greater than 50°, in which at least three consecutive vertebrae are wedged. It is a common cause of acute otitis media, pneumonia, and invasive bacterial infections (bacteremia and meningitis) in the pediatric population. Table 1-2 Rules of Thumb for Expected Increase in Height 24 Age 0–12 months (average birth length is 50 cm) 13–24 months 2 years–adolescence Expected Height Increase 25 cm/year Birth length increases by 50% at 12 months of age 12.5 cm/year 6.25 cm/year Birth length doubles by age 4 years Birth length triples by age 13 years Table 1-3 Rules of Thumb for Expected Increase in Head Circumference Age 0–2 months (average birth head circumference is 35 cm) 2–6 months By 12 months Expected Head Circumference Increase 0.5 cm/week 0.25 cm/week Total increase = 12 cm since birth FIGURE 1.1 Differential diagnosis of failure to thrive (FTT). Galeazzi sign, which assesses asymmetry of the apparent femoral length. The intestine of patients with gastroschisis may be affected by abnormalities in intestinal vascular supply or by inflammation due to exposure to the amniotic fluid. pneumoniae infection. Conjunctivitis and Red Eye IV. It can cause nasal congestion and rhinorrhea for several days. You are planning on testing him for human immunodeficiency virus (HIV) infection and are reviewing common infections and the clinical presentation of children with HIV infection. UGI study with small bowel follow-through can reveal mucosal ulcerations, narrowed lumens, thickened walls, and fistulas. False negatives may result if the urine is very dilute. Any part of the body may be affected. Gastroschisis is a congenital fissure of the anterior abdominal wall, usually located to the right of the umbilical cord stump (in the right paraumbilical area). The most common causes include viral, bacterial, and allergic conjunctivitis, as well as blepharitis (eyelid inflammation). Neurologic deficits are usually not present or are only very mild. Prenatal diagnosis is common. In juvenile myasthenia gravis, remission of symptoms can be as high as 60% after thymectomy. Note that there is usually no testicular enlargement because these patients do not have an increase in FSH, which would stimulate seminiferous tubule enlargement [see exceptions in section II.B.2.d.(3).(c)]. Salmonella should be considered in patients with sickle cell anemia. Urine-reducing substances are positive. Environmental triggers. This child may have enterohemorrhagic E. Botulism immune globulin improves the clinical course. Treatment includes administration of vitamin K. Increased nutritional support with concentrated calories, use of mediumchain triglyceride–containing formulas, and provision of fat-soluble vitamins A, D, E, and K are indicated. This is excessive glucocorticoid production caused by excessive ACTH production by a pituitary tumor, such as a microadenoma. The skull is most commonly involved. Diagnosis is by identifying the typical histologic features on biopsy of skin or bone lesions. Drug Allergy VIII. Treatment should be initiated when chorea is leading to significant impairment of motor function. Such patients have negative urine dipsticks for blood. However, because spondylolysis is a stress fracture, plain films may not detect the fracture. Both surgery and antibiotics are needed. 95. There is some degree of correlation between the mutation and clinical phenotype. Febrile seizures are defined as any seizure that accompanies a fever owing to a non-CNS (central nervous system) cause in patients between the ages of 6 months and 6 years. Passive immunization involves delivery of preformed antibodies to individuals who have no active immunity against a particular disease, but have either been exposed to or are at high risk for exposure to the infectious agent. Examples include diphtheria, tetanus, and acellular pertussis (DTaP); hepatitis A (Hep A) vaccine and hepatitis B vaccine (HBV); inactivated polio vaccine (IPV); Haemophilus influenzae type b (HIB); inactivated influenza; pneumococcal and meningococcal vaccines. Which of the following is the most likely cause of pneumonia in this adolescent? Clinical features include constipation, vomiting, and abdominal distension. Bone scan and chest CT scan are performed to evaluate for metastatic disease. The end result is decreased pulmonary compliance, increased airway resistance, increased pulmonary artery pressures, and impaired gas exchange. The goal of the emergency phase is to restore or maintain the intravascular volume to ensure perfusion of vital organs. Ring sideroblasts result from the accumulation of iron in the mitochondria of RBC precursors. Renal biopsy to establish the diagnosis or to determine a management approach is not indicated for most patients with nephrotic syndrome. The following fractures are considered highly suggestive of abuse: 1. Past medical history should include prenatal history, medications, infections, and family history of liver disease or jaundice. Type III is the most severe type among those who survive the neonatal period. RBCs originating in the glomerulus are dysmorphic in character, often with blebs in the RBC membrane. Craniofacial findings include macrocephaly, prominent forehead, low nasal bridge, and midface hypoplasia. This patient has a 25% likelihood of developing a subsequent malignancy. Substance Abuse V. The different stages of pubertal development are defined as follows. Which of the following statements regarding his maintenance fluid and electrolyte requirements is correct? A 2-month-old boy is brought to your office for evaluation with a 2-day history of poor feeding and vomiting. Chronic supraphysiologic steroid use (usually greater than 2 weeks) b. The prognosis for complete recovery is poor. Vitamin B_{12} deficiency leads to a macrocytic anemia with a mean corpuscular volume (MCV) >95. The HPGA releases gonadotropin-releasing hormone (GnRH) from the hypothalamus, which binds to receptors in the pituitary gland and causes the release of folliclestimulating hormone (FSH) and luteinizing hormone (LH). A patient must fulfill five or more of nine specific criteria almost every day for at least 2 weeks to make a definitive diagnosis of major depressive disorder. Disease onset typically occurs during the summer months. Disease affecting the entire colon is known as pancolitis. Intravenous immune globulin therapy for a presumed diagnosis of Kawasaki disease B. Pathophysiology 1. Eradication of GABHS infection a. Macrocephaly 1. CNS complaints include listlessness, irritability, seizures, and decreased consciousness with encephalopathy. Information about the toxin should include the type or name of toxin, toxin concentration (if known), and the route of exposure. Noonan syndrome with right-sided heart defects). Table 4-2 Frequent Problems of Preterm Infants Disrupted mother–father–infant interaction Perinatal asphyxia Hypothermia Hypoglycemia Hypocalcemia Respiratory distress syndrome (hyaline membrane disease; surfactant deficiency syndrome) Fluid and electrolyte abnormalities Indirect hyperbilirubinemia Patent ductus arteriosus Intracranial hemorrhage Necrotizing enterocolitis Infections Retinopathy of prematurity Bronchopulmonary dysplasia Anemia Neurodevelopmental impairment 134 III. Young, MD, Associate Medical Director, Children's Health Clinic, Cedars-Sinai Medical Center, Los Angeles, California, Assistant Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California 18 CHAPTER 1 19 Pediatric Health Supervision Kathy L. These patients present similarly to patients with the more common 21-OH deficiency, except that they are hypertensive and hypokalemic. Documentation of low serum vitamin B_{12} level is diagnostic. The infant has infantile Tay–Sachs disease, a devastating 197 6. 450 CHAPTER 12 451 Neurology Rujuta B. Rationale for vaccine: Hep A is the most common viral cause of hepatitis worldwide, although it is asymptomatic in up to 70% of infected children younger than 6 years. The 100% oxygen test (hyperoxia test): ABG is performed before and after administration of 100% oxygen. Although FFR is the most likely cause, a thorough history and physical examination (including a complete neurologic examination) must be conducted to identify the presence of an organic etiology. This syndrome places children at risk for a lethal ventricular arrythmia known as torsades de pointes (question 104). Bronchoscopy with alveolar lavage may be performed when aspiration is strongly suspected. Laboratory evidence of aseptic meningitis The response options for statements 9–14 are the same. Hydrocephalus is an elevation in intracranial pressure (ICP) due to a disturbance in the flow of cerebral spinal fluid (CSF). Dermatomyositis typically presents with proximal muscle weakness, characterized by a positive Gowers sign (difficulty standing from the sitting position and, as a result, having to “climb” up the thighs for support). It might be useful to observe the infant while feeding in the emergency department. Muscle and nerve biopsy d. A cherry-red spot may also be seen. Whole-bowel irrigation (WBI) is rapid, complete emptying of the intestinal tract accomplished using polyethylene glycol (an osmotic agent) and an electrolyte solution (to prevent electrolyte imbalance). 457 458 II. African American children may develop secondary sexual characteristics earlier than other children. It is successful in 90–95% of cases. Caloric irrigation. Hypercalciuria is a common cause of hematuria; however, a flank mass would not be a presenting sign. Pericarditis and pericardial effusions are less common. Parenteral antibiotic therapy for a presumed diagnosis of meningococcemia pending blood culture results 3. Meningocele. Clinical features, diagnosis, and management are found in Table 18-2. There are many types of epilepsy syndromes recognized. History, clinical presentation, and positive viral culture or direct fluorescent antibody staining of vesicular fluid are the basis of diagnosis. Blood loss during birth may contribute to the anemia. 597 VII. Etiologies are classified as either congenital or acquired (Table 1-4). Clinical features (Table 5-3) 1. Platelet count is normal. Scoliosis alone should cause no pain unless the curve is very great. Corticosteroids are often used for severe anemia and are continued until hemolysis diminishes. Female Reproductive Health Issues 3 VII. As mentioned earlier, it is one of the best selling short book of pediatrics with many positive reviews. Ophthalmologic consultation to evaluate for a bacterial corneal ulcer is necessary if the abrasion is associated with contact lens use. Head circumference > 95% for age. The answer is E [V.F.1]. Wright, MD, FAAP, Director, Wright Foundation for Pediatric Ophthalmology, Director, Pediatric Ophthalmology Research and Education, Cedars-Sinai Medical Center, Los Angeles, California, Clinical Professor of Ophthalmology, Keck School of Medicine at the University of Southern California, Los Angeles, California Sharon L. Lee T. Patients with thalassemia minor have normal to elevated RBC counts as opposed to iron deficiency in which the RBC count is low to normal. Common signs and symptoms include chronic laryngitis,

hoarseness, wheezing, the development of vocal cord nodules, and 369 subglottic stenosis. Patients may rarely be asymptomatic at the time of diagnosis. Sydenham chorea (St. Vitus dance) 1. Complete history should include questions about recent trauma, prior abdominal pain or surgery, infections, and medication use. Vocal tics can be simple (e.g., coughing, grunting, humming, clearing the throat) or complex (e.g., echolalia, which is the repetition of heard words or phrases). Specific Helminth Infections XVII. Child protective services or law enforcement agencies must be notified if there is a suspicion of abuse. The clinical presentation may be oliguric (diminished urine output) or nonoliguric (normal urine output). Rh hemolytic disease occurs when the mother, who has no Rh antigen (maternal Rh negative), produces antibodies to the Rh antigen on her fetus's RBCs (fetal Rh positive). Otoacoustic emission (OAE) measures sounds generated by normal cochlear hair cells that are detected by a microphone placed into the external auditory canal. She is referred to you for further evaluation. As compared with adults, infants require more protein in their diet (2.2 g/kg during infancy, decreasing to 0.8 g/kg during adulthood). Curves are right thoracic 90% of the time. (See also Chapter 3, section IX.C.1.) 2. Patients with both disorders have a decreased upper-to-lower segment ratio. Bronchoscopy would be indicated if foreign body aspiration was suspected, which is less likely in a 5-month-old. Serotonin (5-HT), calcitonin gene related peptide (CGRP), inflammatory markers (substance P, vasoactive intestinal peptide), the trigeminal nerve and changes in cerebral blood flow all appear to play a role. The WBC count and ESR are normal or only slightly elevated. Severe: spontaneous bleeding (5% factor VIII protein activity) 3. Finally, my pediatrician colleagues and dear friends at UCLA, Dr. Kate Perkins and Dr. Deb Lehman, whose partnership and friendship I will always cherish. Clinical findings are related to extravascular hemolysis. One α-globin gene is deleted. In an injured brainstem, movement of the head does not evoke any eye movement. The urea cycle is responsible for the metabolism of excess nitrogen into urea. History 1. coli sepsis are not typical features of these diseases. Central (or isosexual) precocious puberty (CPP) 1. Four α-globin genes are deleted. Acute pancreatitis a. Assess for prematurity or intrauterine growth retardation (IUGR) or small for gestational age (SGA). CREST syndrome refers to a form of scleroderma with less extensive involvement, manifesting with calcinosis, Raynaud phenomenon, esophageal involvement, sclerosis of the skin, and telangiectasias. Imaging. Western blot, if the ELISA is positive or equivocal, to confirm the diagnosis. There are no chronic sequelae of the joint, skin, and CNS manifestations of rheumatic fever. An anterior–posterior radiograph of the neck will show a “steeple sign.” E. Treatment includes supportive care and botulism immune globulin. Kernicterus has been reported in neonates. Short Stature A. Endocrinopathies that cause short stature 1. Some forms of RTA result in nephrocalcinosis, which in turn may lead to polyuria from urinary concentrating defects. Physical therapy may be necessary for several weeks or longer to aid recovery. Hypergonadotropic hypogonadism. To meet their metabolic demands and facilitate growth, patients require very high caloric intakes. Other features include cataracts and cardiac conduction abnormalities. As with males, serum α-fetoprotein is elevated in yolk sac tumors in females. Mild developmental delay is expected. The constellation of clinical signs and symptoms that include fatigue, muscle weakness, and a heliotrope rash in a girl between 5 and 10 years of age is suggestive of dermatomyositis. Aggressive treatment of hyperammonemia is required to prevent neurologic sequelae. A 5-day-old male infant is brought to the emergency department in coma. Causes of FGR are listed in Table 4-3. Breast buds are usually the first sign of puberty, although in 15% of girls, pubic hair develops first. PPSV23 is composed of polysaccharide capsular antigens from 23 pneumococcal serotypes. Newborns are considered LGA if their birth weight is above the 90th percentile for gestational age. Which of the following is correct? Dandy–Walker malformation is a combination of an absent or hypoplastic cerebellar vermis and cystic enlargement of the fourth ventricle, which blocks the flow of CSF. Children with mild maple syrup urine disease may also present with developmental delay, but their urine has a sweet maple syrup odor. In girls, physical examination shows breast development, pubic hair, and rapid growth. Nutritional support is very important, as patients commonly have failure to thrive with difficulty gaining weight, for which high-calorie diets are prescribed. Each has its own seizure type, severity, and prognosis. Glaucoma may be initially misdiagnosed as NLD obstruction because of the presence of tearing, but is distinguished by the presence of a normal red reflex in patients with NLD obstruction. Vomiting and lethargy are common. Diagnosis is usually based on the clinical presentation. Lower motor neurons control movement in the arms, legs, chest, face, throat and tongue. Radiographic imaging of the abdomen may reveal radiopaque substances. Clinical features are initially caused by the invasion of B. This X-linked disorder has clinical features similar to those of hemophilia A and occurs in 1 in 50,000 males. See Table 10-3 for organic causes of constipation. Joint laxity E. Ocular enlargement, tearing, photophobia, enlarged cornea, corneal clouding, and a dull red reflex are common. Any child who has had a severe systemic reaction to insect venom should be prescribed intramuscular epinephrine, and an emergency plan should be developed, providing instruction on what to do in the event of a future reaction. Positive Barlow maneuver. Pancreatic insufficiency, biliary obstruction, and prolonged diarrhea may result in diminished ability to absorb vitamin K. Presentation is similar to PA and MMA. Aspiration pneumonia is a frequent complication resulting from a diminished gag reflex. Characteristic findings include multiple café-au-lait macules, axillary and inguinal freckling, cutaneous and/or plexiform neurofibromas, iris hamartomas (Lisch nodules), and osseous dysplasia. The extremities are the third most common site of involvement (18% of cases). J. Tics must be present ≥1 year, although their severity and frequency waxes and wanes. Patients with neurocysticercosis have Taenia eggs or adult tapeworm identified in their stools only 25% of the time. Abdominal rigidity is seen with a peritoneal process. Cranial remodeling helmet therapy was a common treatment, but the American Academy of Pediatrics (AAP) updated its position in 2011 stating that helmet therapy was unnecessary for most babies with plagiocephaly. Cardiorespiratory monitors have not been shown to prevent SIDS. A newborn may be LGA with a birth weight above the 90th percentile for gestational age but not have an absolute birth weight above 4000 g. IBD is an important cause of occult and frank GI bleeding. Jejunoileal atresia a. The antistreptolysin O titer is positive in 90% of children after a respiratory infection but in only 50% of patients who have had skin infections. Observation only is needed for most children. History of abuse as a child 3. False-negative results may occur with ascorbic acid (vitamin C) ingestion. Management includes enzyme replacement therapy, bone marrow transplant, and supportive care. Migraine headaches 1. The most extensive state screening panels screen for 50 disorders or more. Bile acid deficiency d. Clinical features of FAP 1. 191 X. IV ceftriaxone and IV doxycycline A 2-year-old boy is being evaluated for failure to thrive. Treatment is supportive, including cool compresses and topical NSAID drops. Fetal and neonatal mortality remains high (50% or greater) in infants in whom the condition is diagnosed before 151 25 weeks' gestation. Therefore, the presence of apocrine odor does not indicate that a patient is in puberty. History of diabetes, hypertension, BMI > 95th percentile (if 2–8 years old) or 35 G. Diagnosis a. General considerations. Pancytopenia and Aplastic Anemia A. Glasgow Coma Scale provides a standard measure to monitor the level of consciousness (see Chapter 20, Table 20-1). Cranial nerve dysfunction is another important feature. Inflammatory Bowel Disease (IBD) X. Symptoms are nonspecific and can be similar to those of a UCD. The mean age of onset is 6 years. Patients often have enlarged gonads, but their secretion of sex steroids is independent of the HPGA. Unconjugated (indirect) bilirubin is combined with glucuronide by the enzyme uridine 5'-diphospho (UDP)–glucuronyl transferase in the liver to form monoand diconjugates (conjugated or direct bilirubin). Male sexual differentiation is initiated by the SRY gene located on the short arm of the Y chromosome. | BISAC: MEDICAL / Pediatrics Classification: LCC RJ48.2 .P385 2019 | DDC 618.9200076—dc23 LC record available at This work is provided “as is,” and the publisher disclaims any and all warranties, expressed or implied, including any warranties as to accuracy, comprehensiveness, or currency of the content of this work. The 4-year-old boy's asthma would be characterized as intermittent asthma. 698 V. A history of neonatal hypoglycemia, prolonged jaundice, and/or microphallus suggests hypopituitarism resulting in growth hormone (GH) deficiency. Thyroid Disorders X. Treatment is supportive and includes cool compresses and topical nonsteroidal anti-inflammatory drug (NSAID) drops. However, in some cases additional diagnostic evaluation is required, focusing on ensuring normal anatomy and confirming suspected abnormalities. Age of onset is variable depending on the type of IEM. This results in the development of the labia, the clitoris, and the lower two-thirds of the vagina. Physical examination is normal in most victims. Which of the following statements regarding rehydration of this child is correct? Diabetes melltus (DM) is the second most common chronic disease of childhood, affecting at least 1 of 500 children. Infants may present with the following: a. A 3-week-old uncircumcised male infant presents with a 2-day history of very poor feeding. Parent with total cholesterol > 240 mg/dL or known dyslipidemia d. Nonorganic CAP, also known as functional abdominal pain (FAP), occurs in two-thirds of cases and is more common in females and in children older than 5 years. Neurologic defects are present and depend on the level of the lesion, varying from complete paraplegia (above L3) to preserved ambulation and variable bladder or bowel incontinence (S3 and below). Prognosis 482 1. Although the degree of renal insufficiency in infancy may range from mild to severe, ARPKD is progressive and ultimately all patients require renal transplantation. Group B streptococcus D. Hypocalcemia is not a feature of RTA. Deep tendon reflexes are absent, and spinal magnetic resonance imaging is normal. Echocardiography typically shows evidence of carditis, such as decreased ventricular function, valvular insufficiency, or pericardial effusion. Urine toxicology screen, serum electrolytes, and metabolic panel should also be evaluated. Normal intelligence with delayed acquisition of motor milestones 4. Female puberty. Onset of bone marrow failure occurs at a mean age of 7 years. If a thoracic curve reaches 50°, there is always progression, even after skeletal maturity. Constipation is often the presenting symptom, followed by weakness, cranial nerve findings, poor abilities to feed, and paralysis. gonorrhoeae, such as 1% tetracycline and 0.5% erythromycin, tend to be less irritating to the conjunctiva. In girls, check serum FSH, LH, and estradiol levels. Table 20-3 Selected Toxins and Their Antidotes Toxin Acetaminophen Anticholinergic agents Benzodiazepines Black widow spider envenomation Carbon monoxide Coral snake envenomation (Eastern U.S. or Texas coral snake) Cyanide Antidote NAC Physostigmine Flumazenil Antivenin Latrodectus mactans Oxygen Antivenin Micrurus fulvius Cyanide antidote kit (contains amyl nitrite, sodium nitrite, sodium thiosulfate) Hydroxocobalamin (vitamin B12) Digitalis glycosides Heavy metals (e.g., mercury, manganese, copper, gold, nickel, zinc, lead, arsenic) Inducers of dystonia Benztropine Inducers of methemoglobinemia Iron Isoniazid Methanol; ethylene glycol Narcotics Organophosphates; carbamate pesticides Pralidoxime (for organophosphates) Pit viper snake bite (rattlesnake, water moccasin, copperhead envenomation) β-Blockers; calcium-channel blockers Sulfonylurea oral hypoglycemic agents Digoxin-specific Fab antibodies d-Penicillamine (for lead, mercury, arsenic, copper) Dimercaprol (British anti-lewisite [BAL] in oil) for all the heavy metals and lewisite (chemical weapon) DMSA (for lead and possibly mercury, arsenic, other metals) EDTA, calcium (for lead, nickel, zinc, manganese) Diphenhydramine Methylene blue Deferoxamine Pyridoxine (vitamin B6) Ethanol, fomepizole Naloxone Atropine Crotalidae polyvalent, antivenin, Crotalidae polyvalent Fab antibodies Glucagon Octreotide Glucagon EDTA = ethylenediaminetetraacetic acid; DMSA = dimercaptosuccinic acid; NAC = N-acetylcysteine. Infants may present with jaundice and anemia. Children with an immunocompromising condition or functional/anatomic asplenia should get a second dose 5 years after the first PPSV23. The majority (75%) occur in females. Respiratory syncytial virus is the most common cause of bronchiolitis. Surgical repair consists of ligation of the fistula and anastomosis of the two esophageal segments. Serologic testing (enzyme-linked immunosorbent assay and Western blot analysis) is recommended for confirmation of disease. Supportive care includes nutrition, fat-soluble vitamin supplementation, and, once bile flow is reestablished, ursodeoxycholic acid. Parotitis IX. While patients with Kawasaki disease may develop aseptic meningitis, the next best step in management is to treat with intravenous immune globulin (IVIG) and high-dose aspirin to prevent cardiac sequelae. Assessment of growth and nutrition 4. Absence seizures last less than 10 seconds and have a very characteristic EEG pattern, showing a generalized 3-Hz spike-and-wave abnormality. An imaging evaluation is important in this infant because of both the pyelonephritis and the positive family history. The Cobb angle, which measures the degree of scoliosis, is measured by drawing a line along the superior aspect of the most angulated vertebrae at the top of the curvature, and drawing another line along the inferior aspect of the lowest most angulated vertebrae of the curvature. Medical history is significant for a 4-day episode of diarrhea 2 weeks before her current presentation. Dermatomyositis may present with constipation from smooth muscle dysfunction and constitutional symptoms, but abdominal pain is not a feature of the disorder. A type I fracture is within the physis. Severe pain, tearing, and photophobia b. In infants with cyanotic congenital heart disease, the PaO_2 usually increases minimally (less than 10–15 mm Hg) and will almost never exceed 150 mm Hg. E. They report that he has struggled in school, requiring some special educational assistance since kindergarten, but has been otherwise healthy. Motility assessment of each eye involves having the child follow a target in all directions. Disorders of the Complement System Review Test Answers and Explanations Chapter 16 Rheumatology I. Bleeding is painless, intermittent, and often streaky. This is ideally achieved through a family-centered medical home model where the pediatric care team works in partnership with the family to ensure that all health-related needs are met. By the 10th week of gestation, the midgut enters the abdomen. Before the serum iron level is known, a test dose of deferoxamine may be administered. Gastrointestinal Bleeding A. Table 13-1 Clinical Features of Anemia Mild Pallor (noted especially on skin and on mucous membranes) Diminished attention Moderate Weakness and fatigue Decreased exercise tolerance Irritability Tachycardia Tachypnea Anorexia Systolic heart murmur Severe Congestive heart failure Cardiac dilation Shortness of breath Hepatosplenomegaly Spoon-shaped nails Table 13-2 Features of Glucose-6-Phospate Dehydrogenase (G6PD) Deficiency Epidemiology Pathophysiology Triggers of hemolysis Clinical features Laboratory findings Mediterranean, Arabic, Asian, and African ethnic groups G6PD enzyme is critical for protecting the RBC from oxidative stress; deficiency results in RBC damage when the RBC is exposed to oxidants Infection Fava beans Drugs (e.g., sulfa, salicylates, antimalarials) Symptoms occur 24–48 hours after exposure to oxidant Hemolysis occurs, resulting in abdominal pain, V/D, fever, and hemoglobinuria followed by jaundice; HSM may be present Hemoglobinuria Elevated reticulocyte count 500 Smear shows “bite” cells, “hemighosts” and Heinz bodies Diagnosis Treatment Low levels of G6PD in RBCs Transfusions as needed; splenectomy is not beneficial V/D = vomiting, diarrhea; RBC = red blood cell; HSM = hepatosplenomegaly. 426 VII. 8. Hepatic function tests, amylase and lipase, and testing for pregnancy and sexually transmitted diseases should be performed, if indicated. Although he has a petechial rash, his platelet count will be normal. Inhaled mucolytics should be considered. In addition to traumatic brain injury, the most common non-traumatic cause of coma in children is infection. Soft Tissue Tumors Rhabdomyosarcoma is the most common soft tissue sarcoma in childhood, and is a malignant tumor of the same embryonic mesenchyme that gives rise to skeletal muscle. It should be noted that other conditions may result in elevated amylase, including gastritis, repeated vomiting (including self-induced vomiting), and renal failure. Carbon monoxide also interferes with cellular oxidative metabolism. Focal spike discharges in the centrotemporal region is seen on EEG. Etiology and pathophysiology 1. A—anal atresia 3. Choice D refers to extra heart sounds, such as clicks transmitted to the aorta in aortic stenosis or bicuspid aortic valves. Older children usually present with the following: a. PCV13 includes the 13 serotypes that account for most invasive pneumococcal diseases. Tumors at sites other than the head and neck or the genitourinary tract carry an intermediate prognosis with survival about 70%. Obtain serum salicylate level at least 6 hours after ingestion. The conversion of testosterone to dihydrotestosterone (DHT) by 5αreductase occurs in the skin of the external genitalia. Therapy is based on a multidisciplinary, team approach that attempts (1) to prevent or minimize inflammation and end-organ dysfunction, (2) to treat complications, and (3) to provide psychosocial and family support, as follows: 1. Meconium plug may occur not only in infants with cystic fibrosis or Hirschsprung disease but also in normal infants. Early-onset oligoarticular juvenile idiopathic arthritis (JIA) B. It is useful to obtain the renal ultrasound in the first day or two in the hospital to assess for severe hydronephrosis that might require urgent urologic consultation. Absence of a region on the paternally derived chromosome 15 is the cause of Prader–Willi syndrome, a disorder associated with neonatal hypotonia, hypogonadism, almond-shaped eyes, and short stature. Upper Extremity II. Lyme Disease VIII. Prognosis is dependent on staging and histology. It's as close as you can get to a reference book while being in the 500-page range. Congenital myotonic dystrophy a. Although the cause is unknown, the following are risk factors that predispose to the development of nonorganic CAP: 1. A direct Coombs test is strongly positive. Dietary recommendations include frequent small meals and thickening of feeds using infant cereal or a commercial thickener, which decrease the symptoms of GERD by making the stomach contents more viscous. Prescription medications B. This autoimmune disorder of the proximal small intestine is characterized by intolerance to gluten, which results in mucosal damage. Hirschsprung disease is the most common cause of organic constipation in an otherwise healthy child (see Chapter 4, section XII.D.5 for details). Management consists of fluid resuscitation, perioperative antibiotics, and most often an appendectomy, often by laparoscopy. The lower esophageal sphincter (LES) is a contracted circular muscle at the distal end of the esophagus that relaxes in a coordinated fashion with the peristaltic wave to allow esophageal contents to enter the stomach. Ongoing losses should be replaced on a “milliliter for milliliter basis” concurrent with the replacement of deficits. The degree of pain varies from severe (acute slip) to mild (chronic slip). The throat should be examined with a tongue depressor to rule out a retropharyngeal abscess. Growth disturbances are defined as growth outside of the usual pattern. Patients are asymptomatic without anemia unless exposed to severe hypoxemia. Adverse effects of immunization 1. Visual acuity is poor at birth, in the range of 20/200, as a result of physiologic immaturity of the visual cortex responsible for visual processing. α-Thalassemia results from defective α-globin chain synthesis, and βthalassemia results from defective β-globin chain synthesis. Terms such as “near drowning,” “dry or wet drowning,” “secondary drowning,” or “active and passive drowning” are no longer used. It develops in otherwise healthy adolescents and is managed with referral to physical therapy and orthopedics. The clinical diagnosis may be challenging, as short stature may be the only clinical manifestation and diarrhea may not be present in all patients. A “steeple sign” on a radiograph of the neck is consistent with the diagnosis of croup, not epiglottitis (which demonstrates a “thumbprint sign” on lateral radiograph of the neck). Urinary Tract Infections (see Chapter 11, section XIV) XIII. As a result, the intestine of gastroschisis patients may develop narrowed or atretic segments and cause bowel obstruction. α1-Antitrypsin deficiency 7. The answer is C [Chapter 16, section II]. Prenatal exposure to alcohol can cause a wide range of findings, with FAS being the most severe. Retinoblastoma X. Congenital aplastic anemia is also known as Fanconi anemia. Long QT syndrome may be congenital or acquired from electrolyte abnormalities (hypokalemia, hypomagnesemia) or medication side effects. FTT may involve all growth parameters, although weight gain is generally the most affected. By comparison, the ratio in normal male infants is typically 90% of cases). Migraine, tension-type, and cluster are common primary headaches. As the patient bends forward, a hump, representing rib or transverse process rotation of the curved spine, may be seen (forward bending test; Figure 17-1). The child's mother should also be examined, as she will often have intellectual disability, as well as the typical physical exam features of myotonic dystrophy. Table 10-5 Differential Diagnosis of Gastrointestinal Bleeding Neonate (Birth– 1 Month of Age) Swallowed Infant/Young Child (1 Month–2 Years of Age) Anal fissure Preschool Age (2– 5 Years) Infectious colitis 389 School Age (>5 Years) Infectious colitis Adolescent (10– 21 Years) Infectious maternal blood Allergic colitis Necrotizing enterocolitis Hirschsprung disease Volvulus Allergic colitis Infectious colitis Hirschsprung disease Intussusception Meckel diverticulum Intestinal duplication Vascular malformation Juvenile polyp Meckel diverticulum Hemolytic uremic syndrome Henoch– Schönlein purpura Swallowed blood (e.g., epistaxis) Mallory–Weiss tear 390 Juvenile polyp Inflammatory bowel disease Swallowed blood (e.g., epistaxis) Mallory–Weiss tear colitis Juvenile polyp Inflammatory bowel disease Mallory– Weiss tear Peptic ulcer/gastritis XI. Chorea is also likely to be found on examination of this patient. Symptoms typically develop after fructose- and sucrose-containing foods are 187 introduced into the diet. Opiate ingestion causes 488 constricted pupils. There are strong genetic influences, but inheritance has not been found to fit into classic Mendelian patterns (autosomal or X-linked). Which of the following is correct regarding the likely 194 6. Outlook is poor. For the latter two groups, the nephrotic syndrome is a chronic illness in which the side effects of medications have to be weighed against the complications of the disease. Parent, grandparent, aunt/uncle, or sibling with myocardial infarction, angina, stroke, or coronary artery bypass graft/stent/angioplasty at 42 weeks from the first day of the last menstrual period. In many cases the diagnosis of GERD is a clinical one, based on the patient's signs and symptoms. Synovial fluid WBC counts may be elevated above the normal reference range, but are less than that seen in septic arthritis (see Table 17-4). Insulin-like growth factor 1 (IGF-1) and insulin-like growth factor binding protein 3 (IGF-BP3) are indirect measures for GH deficiency. Long-term complications include delayed growth and puberty, cardiomegaly, hemochromatosis, cor pulmonale, pulmonary hypertension, renal insufficiency, gallstones, poor wound healing, avascular necrosis of the femoral and humeral heads, osteopenia, retinopathy, and diminished cognitive and school performance. Myasthenia gravis is an autoimmune disorder that presents with generalized weakness, fatigability of muscles, ptosis, and diplopia. Ataxia telangiectasia is characterized by immunodeficiency which may cause recurrent sinopulmonary disease, leading to chronic lung disease with bronchiectasis and pulmonary fibrosis. Examples of RBC membrane disorders include hereditary spherocytosis and hereditary elliptocytosis, and examples of RBC enzyme disorders include glucose-6-phosphate dehydrogenase (G6PD) deficiency and pyruvate kinase (PK) deficiency. Saulsbury, MD, Head, Division of Immunology and Rheumatology, Department of Pediatrics, University of Virginia Health Systems, Charlottesville, Virginia, Professor of Pediatrics, University of Virginia School of Medicine, Charlottesville, Virginia Nirali P. Intermittent high-spiking fevers, joint pain and swelling, lymphadenopathy, and hepatosplenomegaly are common features. Neonatal history is often significant for feeding and respiratory problems. Innocent Cardiac Murmurs III. Spine III. Miller. Her current signs of fever, truncal rash, and swollen lips B. burgdorferi transmission becomes substantial. trachomatis pneumonia. Glucose-6-phosphate dehydrogenase deficiency (G6PD) is the most common RBC enzymatic defect. Empiric antibiotic therapy should be started in symptomatic patients with a suspicious urinalysis while culture results are pending. Commonly due to maternal diabetes. Although a plain radiograph may not show bone changes early, soft tissue swelling (displacement of fat planes) can often be detected. Chorea may start as hand clumsiness and progress to choreoathetoid movements with emotional lability. Cluster headaches. Risks for other types of cancer in patients with Turner syndrome are likely similar to that of the general population. A 9-year-old girl with asthma is brought to the office for the first time. Chest radiography will demonstrate decreased lung volumes with bilateral lobar consolidation. Inappropriate polycythemia may be caused by benign and malignant tumors of the kidney, cerebellum, ovary, liver, and adrenal gland; excess hormone production (e.g., corticosteroids, growth hormone, androgens), and kidney disease. Treatment with intravenous immune globulin can transiently improve symptoms. Rationale for vaccine: HPV is the most common sexually transmitted disease in the United States, affecting the majority of sexually active adults. Hgb H disease. An MRI of the head should be performed in all boys, and in girls with any neurologic symptoms (e.g., headaches or seizures) or with rapid pubertal changes. The answer is B [, sections V.F.3.d and VI.C.3]. Rhee, MD, Pediatric Ophthalmologist, Departments of Pediatrics and Surgery, Associate Professor of Ophthalmology, Medical College of Ohio, Toledo, Ohio Marta R. Low levels are associated with nonspecific symptoms such as headache, flulike illness, dyspnea with exertion, and dizziness. His lungs are likely colonized with Staphylococcus aureus. The maleto-female ratio is 4:1. Henoch–Schönlein purpura is an IgA-mediated vasculitis that presents with a palpable, purpuric rash on the buttocks and lower extremities, large joint arthralgias, renal involvement, and GI bleeding from complications such as intussusception and bowel perforation (see Chapter 11, section V.F.3 and Chapter 16, section I). Fat-soluble vitamins include vitamins A, D, E, and K. Disorders of the Hair VIII. The male-to-female ratio is 2:1. Skeletal disorders 1. A variety of neurologic problems can give the appearance of an unsteady gait. A history of parental consanguinity may be seen in patients with autosomal recessive disorders. Which of the following statements is most accurate regarding the probable diagnosis? Morbidity is related to functional disability and calcinosis. The maintenance sodium requirement is approximately 2–3 mEq/kg/day for infants and children. The diagnosis may be confirmed by skull radiographs and head CT scan. Calcinosis, or calcium deposition, in muscle, fascia, and subcutaneous tissue occurs in up to 40% of children with this disorder. May be accompanied by automatisms, such as eye blinking, chewing, and incomprehensible utterances. Febrile seizures are the most common childhood seizure type, occurring in 2–4% of all children. This can be caused by increased production, blockage of flow, or decreased absorption of CSF. R—renal dysplasia 6. Avascular necrosis of the femoral head b. Acidosis should be confirmed by a venous blood gas. Intravenous access with two large-bore peripheral lines should be established, and a rapid fluid bolus of 20 mL/kg of normal saline solution should be given as needed. Submersion time > 5 minutes Resuscitation delay > 10 minutes CPR required Abnormal neurologic examination or seizures Arterial blood pH < 7 700 VI. It presents with chorea (involuntary, brief, purposeless movements of the limbs and upper body) and emotional lability. It is published by boards review series. Radiographic studies may include a barium enema, rectal biopsy to rule out Hirschsprung disease, and manometry studies. 91. Girls may present with candidal vulvovaginitis. The answer is E [Table 9-2 and III.F.3]. During a seizure, or while in the postictal period, the patient's gait may be irregular and unsteady. The diagnosis is made clinically based on presumptive evidence of rheumatic fever and the exclusion of other likely causes of chorea. 536 FIGURE 14.1 Clinical features of neuroblastoma. These are acids or alkalis with corrosive potential. Type 2—insulin resistant 3. 607 CHAPTER 17 608 Orthopedics Ryan J. Significant instability requires surgical stabilization. Emesis may not always be present, or it may be so severe that it results in suboptimal calorie retention and FTT. downslanting palpebral fissures. Teratogens causing SB include valproic acid, carbamazepine, phenytoin, colchicine, vincristine, azathioprine, and methotrexate. Pathophysiology and etiologies of malabsorption a. Ehlers–Danlos syndrome, classic type a. Cardiac disease often includes pulmonary outflow obstruction alone, or as part of tetralogy of Fallot. Radiopacities may be seen on abdominal radiographs, and dense metaphyseal bands may be seen on radiographs of the knees and wrists (lead lines). Doses >150 mg/kg are associated with toxicity. In DMD, patients are wheelchair dependent by 12 years of age and often die in their late teens from respiratory failure or cardiomyopathy. Most patients with MCD respond to therapy with corticosteroids. There is a self assessment test of 100 questions at the end of this book which is also a great asset for testing yourself for USMLE. Intussusception is the telescoping or invagination of a more proximal portion of the intestine into a more distal portion. Which of the following syndromes is associated with mental retardation and these physical findings? In addition to a history and physical examination, the following are generally indicated: 1. Most poisons are ingested, although poisons may also be inhaled, spilled on the skin or into the eyes, or injected intravenously. Treatment includes ethosuximide, valproic acid, or lamotrigine. Toxic-appearing children with high fever and children with dehydration should also be admitted to the hospital for initial IV antibiotics and hydration. Either depriving the normal eye of clear vision through occlusion or causing blurriness by an eye drop forces the use of the amblyopic eye. Barium upper gastrointestinal (UGI) study examines the anatomy of the esophagus, stomach, and duodenum. ESRD often occurs in males with X-linked Alport syndrome as early as adolescence, but the age of onset of ESRD differs between kindreds. Urinary reducing substances (a nonspecific finding) 3. Most MPSs are inherited in an autosomal recessive manner, with the exception of Hunter syndrome that is X-linked. Autosomal dominant polycystic kidney disease (ADPKD) or adult polycystic kidney disease 1. It commonly presents at 3–13 years of age. 112. The most common cause of primary NS in children is minimal change disease (MCD), which accounts for 95% of NS cases among young children and 50% of cases in older children and adolescents. Henoch–Schönlein purpura B. Neonatal conjunctivitis (ophthalmia neonatorum) 1. The lumen of the appendix is obstructed by either a fecalith or by lymphoid tissue, causing appendiceal distension and ischemia. All children with pyelonephritis, all children with recurrent UTI, prepubertal males, and girls younger than 2 years of age with cystitis should have an imaging evaluation, which should include a renal ultrasound and consideration for a VCUG. The incidence is 1.5–4 in 1000 live births, with a slight male predominance. Bordetella pertussis is the cause of whooping cough. 180 FIGURE 5.3 Summary chart of inborn errors of metabolism. Kyphosis (roundback) is defined as anterior–posterior (AP) curvature of the thoracic spine. Crystals may be indicative of renal stone disease. Caloric intake should also be at the RDA for age. A negative antistreptolysin O titer would rule out the diagnosis of poststreptococcal glomerulonephritis in this patient. Infants have feeding intolerance and vomiting. (Simple transfusion of RBCs is not indicated. In boys, consider adrenal tumors, Leydig cell tumors (presenting with asymmetric testicular enlargement), nonclassic CAH, β-human chorionic gonadotropin (β-HCG)–producing tumors, McCune–Albright syndrome, and testotoxicosis. Setting-sun eyes, a tonic downward deviation of both eyes caused by pressure from the enlarged third ventricles on the upward gaze center in the midbrain 3. Infections of the Skin VI. 502 TEC = transient erythroblastopenia of childhood; SS = sickle cell; URI = upper respiratory infection; Hgb = hemoglobin; RBCs = red blood cells. It affects 10% of the population. Pathophysiology a. The response options for statements 11 and 12 are the same. Management. Other vasculitides. Introduction of cow's milk before 12 months of age has been associated with iron-deficiency anemia and is therefore not recommended. As with any child, overall carbohydrate intake should be moderate and excess simple sugars should be avoided. The chorea may begin as clumsiness of the hands. Constipation may be functional or organic. It is also associated with Beckwith– Wiedemann syndrome and familial polyposis, and is more common in children born prematurely. Recurrence is unusual unless vigorous activities are resumed too soon. Unless otherwise stated, the severe/classic form of each disorder is discussed below. X-rays may show dysostosis multiplex (a constellation of bony abnormalities that include a J-shaped sella turcica; malformed, ovoid, or beaklike vertebrae; short and thickened clavicles; and oar-shaped ribs). Antibiotics are contraindicated and may worsen the clinical course. Persistent Pulmonary Hypertension of the Newborn (PPHN) IX. These genetic mutations cause chronic, excessive activation of complement, which also leads to platelet activation, endothelial cell damage, and systemic thrombotic microangiopathy. Insect Venom Allergy 7 VI. Management of any trauma victim begins with management of the airway. This book is for the quick review for USMLE and other exams. Feeding problems, failure to thrive, vomiting, and diarrhea b. Diarrhea See Chapter 7, section XI. Protein intolerance leading to malabsorption 1. Patterns of bruising may help determine the type of object used to inflict the trauma (e.g., distinctive marks are left by belt loops, buckles, hangers, and hands). The nose and oropharynx should be inspected for non-GI sources of bleeding. Emesis is the most common presentation. Behavioral and developmental pediatric. Maintenance fluid calculations for this child take into account both sensible and insensible losses. 617 III. Corticosteroids are the initial drugs of choice. Autosomal recessive 446 C. The answer is A [, section I.E.2]. The diagnosis of microscopic hematuria may be made if ≥6 red blood cells (RBCs) are noted per high-power field on three or more consecutive urine samples. Inheritance is usually autosomal dominant. In boys, check serum FSH, LH, testosterone, and β-HCG levels. Silent carrier. Although 4–5% of school children may have microscopic hematuria on a single voided urine sample, only 0.5–2% have persistent microscopic hematuria on retesting. Renal disease occurs in up to one half of patients. Short course of oral steroids may be needed for exacerbations. When turning the head of an unconscious patient, the eyes normally look straight ahead and then slowly drift back to midline position, because the intact vestibular apparatus senses a change in position. Inadequate caloric absorption is the least common cause of FTT. Facial findings include a prominent forehead and hypoplasia of the midface region. Normal growth 1. It may occur early (within 24 hours after birth), within the first week of life (classic form), or late (1–3 months after birth). Genital and perianal examination should be performed last and should include inspection of the hymen, vagina, and perianal areas (penis, scrotum, and perianal areas in males), with notation of any discharge, injury, or bleeding. 465 V. Her growth was normal until 2 years of age, when her height started to fall off the growth curve, steadily decreasing to below the fifth percentile. If a single lesion or organ is involved, local curettage may be used. This patient's presentation is most consistent with Guillain–Barré syndrome. The head is tilted toward the affected side and the chin is rotated away from the affected side. Treatment should be immediately initiated with an intravenous first-generation cephalosporin. The answers are E, A, and D, respectively [, Figure 14-1]. Approximately 30% of patients will have a recurrence. Mumps is most commonly associated with parotitis but may also cause meningoencephalitis and orchitis. An autosomal dominant disorders caused by mutations in genes that code for type V collagen. All of these features are seen in homocystinuria but are not expected to be seen in Marfan syndrome. More severe patients are classified as type II or III and have CNS manifestations. GH stimulation testing (with agents such as clonidine or arginine) may be needed. Hurler syndrome (MPS type I) is an autosomal recessive disorder caused by α-liduronidase deficiency. Low reticulocyte count reflects bone marrow suppression or failure and can be seen with RBC aplasias, viral suppression, medication effect, and pancytopenia associated with aplastic anemia. Increased urine copper excretion c. Sexual maturity rating scales are found in Chapter 3, Figures 3-1, 3-2, and 3-3. Vaccine for serotype B is recommended for children ≥ 10 years of age at increased risk of serotype B infection (e.g., community outbreak, functional/anatomic asplenia, and complement pathway abnormalities). No neurologic deficits are present. FIGURE 12.3 Causes of headache. Elevated serum anion gap E. This disease is a large-vessel vasculitis. Severe disease, seen in 10% of cases, presents with more than six stools per day, fever, anemia, leukocytosis, and hypoalbuminemia. Folic acid deficiency a. Arthritis may be associated with either ulcerative colitis or Crohn disease. Clinical features 1. Two types of audiometric tests are used for the newborn. Diplopia secondary to decreased extraocular movements may be the only manifestation. Visual development is most critical during the first 3–4 months of life. Headaches in Childhood VII. Patients should therefore be monitored for signs of hydrocephalus and spinal cord compression. Diagnosis is on the basis of physical examination (the arm being held in external rotation and an inability to touch the contralateral shoulder) and radiographs (especially an 610 axillary view) of the glenohumeral joint to visualize the dislocation and exclude fractures. Serologic markers a. The setting-sun sign is suggestive of hydrocephalus. NLD obstruction is failure of complete canalization of the lacrimal system that results in obstruction to tear outflow. These unabsorbed sugars are detected as reducing substances in the stool by a positive Clinitest reaction. The rash of fifth disease is a “slapped cheek” rash followed by a lacy reticulate rash on the extremities. Crigler–Najjar type 1 is an autosomal recessive disorder in which almost 100% of enzyme activity is absent. Given that she has bloody diarrhea, she is not likely to have Giardia lamblia. Inherited aHUS occurs with both autosomal dominant and autosomal recessive inheritance patterns, although not all patients have identifiable mutations. Cardiac defects may occur (ventricular septal defects are most common). A major discrepancy between a child's present growth percentile and the targeted MPH percentile suggests a pathologic state. Although infantile spasms are frequently cryptogenic (unknown etiology), there are a number of associations, including hypoxic–ischemic injury, tuberous sclerosis, malformations of cortical development, Down syndrome, neonatal hypoglycemia, meningitis, trauma, phenylketonuria, intraventricular 468 hemorrhage, pyridoxine deficiency, and infection. Specific immunizations 1. coli and for Shiga toxin is informative. A male infant born at 26 weeks' gestation was diagnosed with surfactant deficiency syndrome. In particular, unstimulated gonadotropin testing should be done with ultrasensitive or pediatricspecific assays, and not routine LH and FSH assays that are designed for use in adults. Myelomeningocele is the herniation of the meninges and spinal cord tissue through the bony cleft, most commonly in the lumbosacral region. The tumor arises from the coccyx and presents as a soft tissue mass. Time of occurrence, if known, is very important. Malrotation occurs when normal bowel rotation is interrupted. Genetic testing for spinal muscular atrophy (SMA) or myotonic dystrophy F. General assessment. I'll definitely be returning to this book again and again.2- VERY GOOD AND UNIQUE FOR PEDS.3- One of the best books for a brief and deep review of Pediatrics, I love it too much.Check the best step 2 CK books :Kaplan USMLE Step 2 CK Lecture Notes 2019 5-book setKaplan USMLE Step 2 CK Lecture Notes 2019 {Pediatrics}Master the Boards USMLE Step 2 CK 5th Edition 2019First Aid for the USMLE Step 1 2019, Plus New UpdatesPathoma Videos, PDF Books, Study Note 2018 Publisher's Note: Products purchased from 3rd Party sellers are not guaranteed by the Publisher for quality, authenticity, or access to any online entitlements included with the product. Now featuring more than 400 board-style questions with complete answers and explanations, BSR Pediatrics, Second Edition prepares medical students for success in pediatric rotations, shelf exams, and the UMSLE Step 2 with a comprehensive overview of general and subspecialty pediatrics presented in the popular Board Review Series outline format. At least five attacks that last between 15 minutes and 3 hours c. This patient's clinical features point to a likely diagnosis of pneumonia secondary to infection with Chlamydia trachomatis, which is a common perinatally transmitted cause of pneumonia in infants 1–3 months of age. Disadvantages include the induction of vaccine-related polio, a rare complication. Referred pain from arthritis involving the spine E. Chronic and progressive symptoms such as developmental delay or regression of developmental milestones, intellectual disability, or seizures 2. Skin eruption with multiple vesicular lesions 645 2. Treatment includes meticulous and prompt local wound care. Imaging studies (CT or MRI) are performed to determine the extent of local extension and to prepare for surgical excision. Treatment includes vitamin K, FFP, and platelets as needed. Iatrogenic. supports the diagnosis of Lyme disease. 355 6. Small feedings by nasogastric tubes and assisted ventilation are provided if needed. Diabetic embryopathy a. High-resolution chromosomal studies to evaluate for a suspected genetic disorder, which include chromosomal microarray and fluorescent in situ hybridization (FISH). Perinatal asphyxia, intraventricular hemorrhage, and meningitis are other causes of infantile spasms. MDI therapy combines long-acting (basal) insulins with short-acting (bolus) insulins. The answer is E [V.C]. Skin hyperpigmentation 4. The clinical course for the child with MCD follows periods of relapses and remissions. The cause is unknown; however, it is associated with previous retinoblastoma, Paget disease of bone, radiation therapy for cancer, and fibrous dysplasia. Organic CAP is treated on the basis of the specific diagnosis. Immunizations are among the most important components of well-child care and disease prevention. Girls present with typical secondary sexual characteristics or virilization. Drowning A. Gastrointestinal Bleeding XI. History of premature birth, low birth weight, neonatal separation from parents, or multiple births 4. Hypotonia is a reduction in muscle tone in which there is decreased resistance of movement during passive stretching of muscles and a decrease in spontaneous activity. Compound heterozygotic disease can occur with Hgb C or β-thalassemia, leading to Hgb SC or sickle β-thalassemia disease, respectively. Physical examination reveals hypertension and periorbital and peripheral edema. Normal intraocular pressure in infants is 10–15 mm Hg. Infants with congenital glaucoma have intraocular pressures exceeding 30 mm Hg. 2. Varicella vaccine a. Nutritional management includes assurance of adequate caloric intake and avoidance of high phosphorus, high sodium, and high potassium foods. Early disease is further broken down into early localized and early disseminated disease. Surgery may be required for patients who fail conservative and medical management. As a result, live vaccines should generally be avoided in patients with compromised immunity (e.g., cancer, congenital or drug-induced immunodeficiencies, acquired immune deficiency syndrome). Chronic symptoms usually resolve within 1– 2 weeks. An abnormal response suggests pontine injury. Gastritis, esophagitis, and ulcers should also be treated with methods to decrease acid production, such as H_2-blockers or proton pump inhibitors. The skin on the back (usually lumbosacral region) is epithelialized, and a hairy patch or dimple often covers the area. Neonatal myasthenia is a transient muscle disorder caused by the transplacental passage of acetylcholine receptor antibodies. Because TLESR is induced during administration of barium through a nasogastric tube, the sensitivity for GERD is high (85–90%) but the specificity is low (50%). Disorders of the cervical spine 1. Decreased fat absorption may lead to growth failure and vitamin deficiencies. Delay in diagnosis may result in irreversible vision loss and even death. Patients should undergo nasogastric suctioning and fluid resuscitation before surgical resection and anastomosis of the atretic segment of the small bowel. Chest radiography usually demonstrates hyperinflation and atelectasis. Surgery to open outflow channels is almost always required. Rheumatic Fever VII. On examination, you find that he is jaundiced and has hepatomegaly. A 6-month-old male has had hypotonia, facial weakness, areflexia, and a history of feeding problems since birth. Monozygotic twins have a >50% concordance rate, whereas dizygotic twins have only a 30% concordance rate. Risk factors associated with SIDS may be found in Chapter 9, section IV.E.2.b. C. β-Thalassemia minor (heterozygous β-thalassemia or β-thalassemia trait) causes a mild asymptomatic anemia with Hgb levels 2–3 g/dL below ageappropriate norms. Physical examination reveals symmetric weakness at the ankles and knees, with normal strength at the hip joints. Rogido, MD, FAAP, Division of Neonatal-Perinatal Medicine, Assistant Professor of Pediatrics, Emory University School of Medicine, Atlanta, Georgia Annette B. Which of the 444 following statements regarding this patient's diagnosis is correct? Hyphema is blood within the anterior chamber. 651 VII. Stool studies should be performed to rule out infectious enteropathies. Octreotide (vasopressin) can be used to vasoconstrict varices. 410 II. Specific causes of conjunctivitis. Review of her growth records reveals very poor growth consistent with failure to thrive. Autoimmune hemolytic anemia (AIHA) occurs when antibodies are misdirected against the RBCs. a. Diagnosis is by a urine mucopolysaccharide spot test, by an enzyme assay, and by genetic testing. Reprinted with permission from Kyle T, Carman S. Corneal ulcer (rare) c. This patient's clinical presentation and ethnicity are consistent with IgA nephropathy (Berger disease), the most common form of chronic glomerulonephritis in the world. Onset is most common between 5 and 13 years of age. Despite this finding, about 50% of children complain of low-back pain or discomfort in their legs. Bilateral involvement is present in 70% of patients. Bordetella pertussis E. Galactosemia should always be considered in the differential diagnosis of any newborn who develops hypoglycemia and has hepatomegaly. General concepts of hepatic injury 1. For the most up-todate recommendations on the childhood immunization schedule, please visit www.cdc.gov. Homocystinuria E. Bone marrow transplant is curative and is the therapy of choice. Oral rehydration therapy (ORT) 1. Laboratory studies include an elevated white blood cell (WBC) count, ESR, and C-reactive protein (CRP). Kernicterus caused by extremely high bilirubin levels occurs almost universally. The peak age is 1–3 years. Miller Fisher syndrome, a variant of Guillain–Barré syndrome, is characterized by ophthalmoplegia, ataxia, and areflexia. Bone Tumors VIII. The voiding cystourethrogram (VCUG) should be postponed until the urine culture is subsequently shown to be negative. Slow deterioration of pulmonary function (e.g., hypoxemia and hypercarbia) may occur during the first 12–24 hours. Membranoproliferative glomerulonephritis E. Acyclovir, an antiviral that has activity against herpes viruses, is not indicated for enteroviral infection. BRS Pediatrics (Board Review Series) 2nd Edition [PDF]Free Download link Join Our Premium Membership Only For $ 15 JOIN NOW for $ 15 [sociallocker][/sociallocker]Features of the (Download BRS Pediatrics (Board Review Series) 2nd Edition pdf)::Now featuring more than 400 board-style questions with complete answers and explanations, BSR Pediatrics, Second Edition prepares medical students for success in pediatric rotations, shelf exams, and the UMSLE Step 2 with a comprehensive overview of general and subspecialty pediatrics presented in the popular Board Review Series outline format. These diseases affect the integrity of blood vessels and may present with bleeding. 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This self-limited condition lasts for less than 24 hours. Gamma glutamyl transpeptidase (GGTP) and 5′-nucleotidase (5NT) are also elevated in biliary disease. Mitochondrial Disorders These disorders affect multiple organ systems and may arise from mutations in the mitochondrial DNA (present in multiple copies per cell) or the nuclear DNA. Radiographic studies are not routinely indicated unless warranted by the history or physical examination. Deep venous thrombosis, pulmonary embolism, or stroke 4. A complete recovery is expected. Neither neuroimaging nor EEG is needed unless the neurologic examination is abnormal. Serious side effects, including those that are life-threatening or result in permanent disability, are rare (e.g., vaccine-related polio after OPV). Although most acute abdominal pain is self-limited, the pain may result from a serious medical or surgical cause and may require prompt surgical evaluation. Characteristic increasing weakness occurs later in the day and with repetitive or sustained muscle activity. The use of antibiotics is controversial in treating diarrhea due to HUS-associated bacteria. The EEG shows biphasic spike and sharp-wave discharges in the midtemporal and central regions. Table 17-3 Differential Diagnosis of a Painful Limp Mnemonic: “The Joint STARTSSSS HOTT” S Septic arthritis T Transient synovitis A Acute rheumatic fever R Rheumatoid arthritis T Trauma Other Causes of Limp D Developmental hip dysplasia D Discitis L Legg–Calvé–Perthes L Limb length discrepancy L Lyme disease Fracture Strain or sprain S Sickle cell disease Pain crisis (vaso-occlusive crisis) Osteomyelitis S Systemic lupus erythematosus S Steroid use (chronic) S Slipped capital femoral epiphysis (SCFE) H Henoch–Schönlein purpura O Osteomyelitis T Tuberculosis T Tumor Osteosarcoma Leukemia 622 Table 17-4 Septic Arthritis Versus Transient Synovitis Organisms Clinical features Acute Ill-appearing Fever Limp Refusal to bear weight Flexion, abduction, external rotation of the hip Typical joints involved Laboratory findings Elevated CRP (>2 mg/dL) Elevated WBC Blood culture positive 30–50% All laboratory results may be normal Joint aspirate Gram stain may have organisms Treatment Intravenous antibiotics Septic Arthritis Staphylococcus aureus Streptococcus pyogenes Streptococcus pneumoniae Peak 1–3 years Acute or subacute Well-appearing Mild viral prodrome Limp Refusal to bear weight Flexion, abduction, external rotation of the hip Hip, knee, ankle Elevated ESR (>40 mm/hour) Mildly elevated CRP Mildly elevated WBC Blood culture negative >50,000–100,000 cells/mm3 Gram stain without organisms Surgical decompression Rest Transient Synovitis Typically sterile inflammationPossibly postviral Peak 2–7 years Hip Mildly elevated ESR 50°. This patient's clinical presentation with nail dystrophy and skin changes between the toes is most consistent with tinea unguium (onychomycosis) and tinea pedis. Maternal systemic lupus erythematosus nephritis B. Increased risk for congenital malformations occurs in infants born to mothers with type 1 or type 2 diabetes, especially when the Hgb A1C level is >12 just before conception and/or in the early first trimester. Conjugated (direct) hyperbilirubinemia is a marker for cholestasis. Further evaluation by Child Protective Services for possible nonaccidental injury D. Abnormal U/L ratio suggests disproportionate short stature [see section I.D.2.b]. Polycythemia is defined as an increase in RBCs relative to total blood volume. SGA is a diagnosis based exclusively on the birth weight. Disproportionate short stature is defined as short stature in patients who are short 203 legged with an increased U/L ratio, suggesting rickets or a skeletal dysplasia. 462 IV. In neonatal myasthenia, treatment is supportive because the disorder is self-limited. Systemic scleroderma. Blunt trauma is the most frequent cause. Immunodeficient patients should not receive live vaccines including OPV, MMR, rotavirus, and varicella vaccines. Chronic use of drugs, such as steroids or stimulants for attention deficit/hyperactivity disorder, that result in significant appetite suppression and poor weight gain may lead to short stature. In contrast, patients with early-onset oligoarticular JIA are usually female, present between 1 and 5 years of age, have a high risk of developing chronic uveitis, and do not have involvement of the sacroiliac joints. Germ Cell Tumors (Germinomas) XI. In December 2016, the Food and Drug Administration (FDA) approved a new treatment (Nusinersen) to improve motor function in children with SMA. Reprinted with permission from Tecklin JS. Treatment is based on the suspected cause. Management includes enzyme replacement therapy and supportive care. Treatment of yolk sac tumors involves radical orchiectomy and, if necessary, retroperitoneal lymph node dissection. Chromosomal disorders: Prader–Willi and Down syndromes 4. Wilson disease is an autosomal recessive disorder of copper metabolism that results in copper accumulation in various tissues, including the liver, brain, and cornea. The two major causes in children are folic acid and vitamin B12 deficiencies. Chronic illness b. By 12 weeks, this process is complete, except for penile growth, which continues to term. Ovarian tumors most commonly include yolk sac tumors, teratomas, and dysgerminomas. If there are atypical features or concern for other causes of ataxia, however, neuroimaging is necessary (e.g., to rule out acute life-threatening causes such as tumors or hemorrhage in the posterior fossa). Infants often appear otherwise well. Recent dietary changes include the introduction of cereals, fruits, and herbal tea with honey. A group of autosomal recessive disorders of glycogen metabolism that result in glycogen accumulation in various organs. Lymphomas IV. Metronidazole is used in the treatment of CD, especially for perianal involvement. Vertigo from migraines, acute labyrinthitis, and brainstem tumors may lead to unsteady walking. The treatment of congenital torticollis, including head positioning during sleep and physical therapy, should be started as soon as the diagnosis is made to prevent this complication. Table 11-2 Types of Renal Tubular Acidosis (RTA) Type of RTA Distal RTA (Type I) Proximal RTA (Type II) Characteristic Features Causes or Associations Clinical Presentation Treatment Inability of the distal renal tubular cells to excrete acid (H+) Isolated inherited defect Associated with nephrotic syndrome, sickle cell anemia, connective tissue disorders Associated with toxins, drugs (amphotericin) Vomiting Growth failure Acidosis Nephrocalcinosis and nephrolithiasis Small doses of oral alkali Impaired bicarbonate reabsorption by the proximal renal tubular cells Isolated inherited defect Intoxication (heavy metals) Prematurity Drugs (gentamicin) Associated with more global defects in tubular reabsorption (Fanconi syndrome*) Vomiting Growth failure Acidosis Muscle weakness Large doses of oral alkali 431 Type IV RTA Transient acidosis in infants and children Hyperkalemia is the hallmark Associated with renal disorders such as obstructive uropathy and interstitial nephritis Diabetes mellitus Associated with mineralocorticoid deficiency states * Patients may be asymptomatic or may present with failure to thrive Furosemide to lower serum potassium; oral alkali Findings associated with Fanconi syndrome: proximal RTA, hyperphosphaturia, aminoaciduria, glucosuria, and potassium wasting. This procedure is often performed laparoscopically and may be done 378 electively after the acute episode resolves. Exploration for malrotation and other luminal obstructions is also performed at the time of surgical correction. Clinical features of Type I OI 1. Hunter syndrome is similar but does not have corneal clouding. Epileptic syndromes 1. The antiepileptic medications usually used include ethosuximide, valproic acid, or lamotrigine. Neither corticosteroids nor mucolytics are effective treatments for C. The most commonly used screening test for Wilson disease is a low serum ceruloplasmin, which is very suggestive of the disorder. Inflammatory bowel disease (IBD)–associated arthritis. These are likely due to a combination of genetic, developmental, and environmental risk factors. Table 20-4 Stages of Acetaminophen Toxicity 2 3 Time After Ingestion 30 minutes– 24 hours 24–72 hours 72–96 hours 4 4 days–2 weeks Stage 1 Signs and Symptoms Asymptomatic, or vomiting and diarrhea Gastrointestinal symptoms resolve; at 36 hours, hepatic transaminases begin to increase Hepatic necrosis, jaundice, hypoglycemia, lactic acidosis, hepatic encephalopathy, coagulopathy, and renal failure Resolution of symptoms, progressive liver damage requiring liver transplantation, or death Table 20-5 Stages of Iron Toxicity Stage 1 Time After Ingestion 1–6 hours 2 3 6–12 hours 12–36 hours Signs and Symptoms Abdominal pain, vomiting, diarrhea, GI bleeding Shock from bleeding and vasodilation Fever and leukocytosis Resolution of stage 1 symptoms Metabolic acidosis 711 4 2–6 weeks Circulatory collapse Hepatic and renal failure DIC Neurologic deterioration Late sequelae includes pyloric or intestinal scarring with stenosis DIC = disseminated intravascular coagulation; GI = gastrointestinal. The answer is E [I.B and I.C]. Preventive care 1. Benign epilepsy with centrotemporal spikes (BECTS), which is also known as benign rolandic epilepsy a. Measuring the erythrocyte sedimentation rate will confirm the diagnosis. The causative organism is not isolated in up to 30–50% of cases. Recurrence rate is very high after bowel resection. Categories a. A high index of suspicion is necessary because the symptoms may be subtle and the conditions can be life-threatening. Also known as papular acrodermatitis, GCS most often occurs in children between the ages of 6 months and 12 years and is associated with hepatitis B, Epstein–Barr virus, enterovirus, and echovirus infections. Complications 1. 96. Internal rotation, flexion, and abduction are usually reduced in the affected hip. Infection is acquired from the vaginal canal during birth or from hand-to-eye contamination from infected individuals. A 3-year-old boy with fever, bilateral knee swelling, and a “salmon-colored” rash on the trunk and proximal extremities. Smith–Lemli–Opitz syndrome. G. When RBCs are present on microscopic examination, careful examination of RBC morphology and identification of other urine elements, such as casts or bacteria, may be extremely helpful in determining the cause of the hematuria. Acute renal failure may result. LH in boys stimulates the testicular Leydig cells to produce androgens, which in turn are responsible for penile enlargement and the growth of axillary, facial, and pubic hair. Diagnosis requires 595 evidence of recent streptococcal infection and either two major criteria or one major plus two minor criteria. Langerhans Cell Histiocytosis (LCH) A. Bilateral ptosis is the most common presenting sign. Protein intolerance is characterized by diarrhea, vomiting, and colicky abdominal pain that occur after exposure to dietary protein. Patients usually acquire neurocystercercosis from ingestion of Taenia eggs, not from ingesting raw pork, but via the fecal oral route after contact with someone infected with the adult tapeworm, excreting eggs in their stool. Malrotation is an anatomic abnormality of intestinal rotation that allows the midgut to twist around the superior mesenteric vessels (i.e., volvulus). Appendicitis a. Chronic draining ears may indicate LCH involving the mastoid. Abdominal complaints include colicky pain, constipation, anorexia, and vomiting. To my parents, siblings, and close friends, for believing more in me than I ever could myself. BRS Pediatrics pdf Review: It is written by Llyod J. von Willebrand disease. When peripheral hypotonia is suspected, it is necessary to consider the following: a. A voiding cystourethrogram reveals grade 2 vesicoureteral reflex (VUR), but her renal ultrasound is normal. Of the vaccines currently recommended for children younger than 6 years, only some forms of influenza vaccine contain small amounts of thimerosal. Constipation is the classic first symptom of botulism. The answers are C, B, D, E, and A, respectively [, section IV and Table 7-3]. Septic arthritis of the hip is an orthopedic emergency. A 1-week-old male infant is brought to the emergency department because of vomiting and decreased feeding for 3 days. Fluorescein staining of the corneal epithelium is performed to evaluate for an abrasion of the corneal tissue. ACTH stimulation test is the test of choice and measures adrenal cortisol reserve by comparing the baseline cortisol level with the cortisol level 1 hour after ACTH injection. Most IEM are inherited in an autosomal recessive fashion, but several are X-linked recessive. It is more common in males. Clinical findings of B12 deficiency include hyporeflexia, ataxia, and a smooth red tongue, in addition to the classic symptoms and signs associated with anemia (fatigue and pallor). 139 FIGURE 4.2 Differential diagnosis of respiratory distress in neonates. This drug is one of the most common medications ingested by children and adolescents. Histoplasmosis, another endemic fungal infection, is endemic in other parts of the country, including the Mississippi River Valley. C3, ANA, and antistreptococcal antibodies are indicated during an initial evaluation to rule out causes of NS other than MCD. First-degree AV block is characterized by PR prolongation without failed AV conduction. He also has trident-shaped hands and bilateral short femurs and upper arms. Children have a slightly painful limp with decreased internal rotation and abduction of the hip. With treatment, 90% of patients survive to adolescence, but many have functional disabilities. Neurologic symptoms follow, including weak cry and suck, loss of previously obtained motor milestones, ophthalmoplegia, and hyporeflexia. A 14-year-old girl with arthritis, alopecia, leukopenia, and “a bad sunburn” on her face. Special diet (methionine restricted, cystine enhanced) b. You perform a laboratory evaluation to look for evidence of infection. Spondylolysis is a stress fracture in the pars interarticularis (i.e., the bone that connects the superior and inferior articular facets of a vertebral body) usually secondary to repetitive hyperextension of the spine as occurs in gymnastics, tennis, and diving. *Polyarteritis nodosa. Duration of antibiotic treatment is 4–6 weeks. Topical agents (clotrimazole) are not very effective for nail infection, but rather, systemic therapy (terbenafine) is indicated. Hereditary elliptocytosis is another autosomal dominant defect in the structure of spectrin that may or may not result in hemolysis. Herniated intervertebral disk is much more common in adults, but may occur in adolescents. SS disease is caused by a single amino acid substitution of valine for glutamic acid on the number 6 position of the β-globin chain of Hgb. The 8-month-old infant has infantile botulism, an environmentally acquired disorder of the neuromuscular junction in which botulinum neurotoxin blocks release of acetylcholine at the neuromuscular junction. Patients should be followed, and if progressive enlargement of the tissue is noted, they should be referred to a pediatric endocrinologist. Some common causes include the following: 1. Abdominal pain and chronic diarrhea may be caused by inflammatory bowel disease. Rehydration solutions should not contain KCl until her urine function is evaluated. Reactive arthritis. (Physical characteristics of child abuse are described further in Chapter 20, section VI.) b. An elevated lead level or elevated erythrocyte protoporphyrin is the basis of diagnosis. Abdominal examination a. Prevalence of epilepsy in children is 0.5–0.8%. Organic acidemias. Glycogen storage diseases (GSDs). The peak ages of onset are less than 1 year and between 9 and 11 years. Glucocorticoids are the mainstay of therapy for children with SLE. 590 d. The signs and symptoms of anemia are listed in Table 13-1. The infant may have isolated or multiple malformations, including caudal regression syndrome (sacral agenesis), neural tube defects, renal malformations, cardiac defects, or craniofacial abnormalities. 210 211 III. Surgical correction is with a partial pyloromyotomy, in which the circular pylorus muscle fibers are transected. Although edema of the hands and abdominal pain can be seen in Henoch–Schönlein purpura, fevers and mucous membrane findings are not usually present. Serum electrolytes, including calcium and magnesium, ammonia, lactate, and pyruvate levels should be drawn to evaluate for a metabolic disorder. A 15-year-old boy who has mild hearing loss, mild hypertension, hematuria, and proteinuria. If stool losses continue, these losses should not be replaced until all the deficit fluids are replaced. Older children and adults may have recurrent calculi, muscle weakness, bone pain, and myalgias. Rationale for vaccine: HIB was a serious cause of invasive bacterial infection, including meningitis, epiglottitis, pneumonia, and sepsis, before vaccine licensure in 1985. However, it may occur in children with predisposing conditions such as sickle cell disease, cystic fibrosis, or prolonged TPN therapy. Rationale for vaccine: Infection with Neisseria meningitidis can cause meningitis and sepsis, which is fatal in up to 15% of children. Allergic conjunctivitis is suggested by severe itchiness and is usually seasonal. Eye misalignment (strabismus: see section IX). Scoliosis is typically asymptomatic. Omphalocele is frequently associated with other congenital anomalies, including congenital heart defects (most commonly tetralogy of Fallot and atrial septal defects), Beckwith–Wiedemann syndrome, and chromosomal disorders (trisomy 13 or, less frequently, trisomy 18; see also Chapter 5, section IV.E.5). Screening for Helicobacter pylori with stool antigen or breath testing should be reserved for children with symptoms of dyspepsia or epigastric pain, because the asymptomatic carriage rate of H. 704 VIII. It commonly presents with a nonthrombocytopenic purpuric or petechial eruption on the buttocks or thighs with abdominal pain, arthritis, and glomerulonephritis. Slipped capital femoral epiphysis (SCFE) is a slipping of the femoral head off the femoral neck. This is commonly associated with polyhydramnios due to the fetus' inability to swallow amniotic fluid. Jaundice may be a late finding. 770 Index Note: Page numbers followed by “f” indicate figures and “t” indicate tables. The cause is adenovirus, types 8, 19, and

37. Two years postmenarche is considered skeletally mature. Carbon monoxide displaces oxygen from the hemoglobin molecule, forming carboxyhemoglobin (CO-Hb), which can no longer carry oxygen. Treatment involves containment or ensuring that the femoral head is positioned within the acetabulum to facilitate its remolding and reossifying. Neurofibromatosis type 1 A 6-year-old boy complains of a 4-day history of low-back pain and difficulty walking. Early intervention for hearing impairment is associated with better developmental language outcomes, and should be initiated no later than 6 months of age. ORT is inappropriate for patients with severe life-threatening dehydration, for patients with paralytic ileus or GI obstruction, and for patients with extremely rapid stool losses or repeated severe emesis losses. Some patients with IBD and joint symptoms may be HLA-B27 positive and have involvement of the axial skeleton in a pattern indistinguishable from ankylosing spondylitis. 504 III. Pain usually improves within 3 days with analgesics and rest. If shaken baby syndrome is suspected, a dilated ophthalmoscopic evaluation for retinal hemorrhages should be performed (see Chapter 18, section V.A). Incidence is 4–10 in 100,000 live births. Photophobia from corneal inflammation (keratitis) caused by a hypersensitivity reaction to the virus (occurs in one-third of patients) 3. One-third of testicular tumors in childhood are benign, unlike in adults, in whom almost all testicular tumors are malignant. Initial assessment must include a detailed history and physical examination. Late complications of radiation therapy include pathologic fractures at the tumor site, retarded bone growth, limb length discrepancy, functional impairment, and secondary malignancy. Shock and cardiovascular collapse may occur as bowel ischemia progresses. This screening test is routinely performed in newborn nurseries before infant discharge. influenzae, adenovirus, and picornavirus. Reprinted with permission from Pillitteri A. Gastroesophageal reflux disease (GERD) is a pathologic state associated with gastrointestinal (GI) or pulmonary symptoms and sequelae. Serum lipase level elevation is more specific for acute pancreatitis and remains elevated longer than serum amylase levels. Oral agents. Complications and associations. The presence of RBC casts indicates a cause other than MCD. Minor features include fever, arthralgias, leukocytosis, increased ESR, and prolonged PR interval on electrocardiogram. Most bites occur in boys during the spring and summer months. α-Thalassemia is the result of deletions of the α-globin chain and occurs predominantly in Southeast Asians. Diagnosis is by finding decreased PK activity in the RBCs. 4. Erythema, swelling, and asymmetry of soft tissue folds may be present. Congenital glaucoma, if not detected and treated surgically early, leads to blindness. The prognosis for children with poststreptococcal glomerulonephritis is excellent, and affected children usually recover completely; renal failure 448 5. Intestinal malrotation may result in volvulus (loops of intestine twist if attached to a narrow band of mesentery) with restricted circulation to the rotated segment, leading to intestinal gangrene. It may still be helpful up to 72 hours after ingestion. Spinal MRI may be necessary in children younger than 3 years to rule out compressive lesions of the spinal cord, because the sensory examination in children of this age is often difficult to evaluate. Although patients may be more willing to move their legs than patients with septic arthritis, they may also hold their hip in a flexed, abducted, and externally rotated position, similar to the position seen in septic arthritis. Carbon monoxide poisoning a. Given that he is ill-appearing and has a petechial rash, sepsis with typical bacterial pathogens should also be considered. Marfan syndrome 4. Fetal alcohol syndrome E. 183 VI. On physical examination, he is nontoxic and moderately dehydrated and has a temperature of 38.3°C (101°F). You are called to the nursery to examine a “floppy” female infant born within the past 24 hours. Goals of therapy include normalization of the child’s activities, education of parents as to the nature of the disorder, and development of methods to empower the child and family to control the discomfort. A 12-year-old girl is brought to see you for evaluation of frequent headaches. Positive family history for IBD is found in approximately 15–20% of patients. These include certain vitamins, minerals, amino acids, fatty acids, and a carbohydrate source. Therefore, all of the pathways of the ventricular system communicate normally, and the problem occurs at the beginning or end of the pathway. Minor Jones criteria include fever, arthralgias, elevated erythrocyte sedimentation rate, and leukocytosis (not leukopenia). Symptoms usually begin with periumbilical pain, followed by vomiting. Diagnosis is on the basis of clinical findings and radiologic confirmation. General concepts about malabsorption 1. Symptoms are improved by sleep. This autosomal dominant disorder is characterized by paucity of intrahepatic bile ducts and multiorgan involvement. Intestinal symptoms include abdominal pain, postprandial cramping, diarrhea, and anorexia. Therapy is geared to abort organ destruction or dissemination. 188 VIII. IgA nephropathy D. Growth Abnormalities A. Type 1 Diabetes Mellitus (Type 1 DM) A. Trisomy 21 D. In addition to hypotonia, patients with congenital neuromuscular disorders may have additional findings such as bilateral ptosis, ophthalmoplegia, flat midface, high-arched palate, chest wall abnormalities (e.g., bell-shaped chest, pectus excavatum or carinatum), and bilateral cryptorchidism. Splenectomy cures the disorder, but to decrease the incidence of invasive disease caused by encapsulated bacteria, splenectomy is generally delayed until after 5 years of age. Occasionally, a sausage-shaped mass may be palpated in the abdominal right upper quadrant, representing the intussusception. Myocarditis is usually manifested by tachycardia out of proportion to the extent of the fever. Stool sample for Giardia intestinalis–specific enzyme immunoassay (EIA) C. Other complications include aplastic crises, most commonly associated with parvovirus B19 infection, and pigmentary gallstones. Hypopituitarism of any cause (e.g., brain tumors) d. Congenital clotting factor disorders. Epidemic keratoconjunctivitis is clinically similar to pharyngoconjunctival fever, but symptoms are confined to the eyes. The user interface is straightforward and simple, enabling the user to reach the desired content with just a few clicks Use Promo Code at Checkout: 50OFF Expires: April 30, 2022 (only 5 slots available) Pediatrics SECOND EDITION Editors Lloyd J. Intracranial causes a. Duchenne and Becker Muscular Dystrophies (DMD, BMD) X. AST is also found in skeletal muscle, red blood cells, and cardiac tissue. Urinalysis reveals hematuria with red blood cell casts and 2+ proteinuria. Renal vein thrombosis 11. Which of the following statements regarding this infant’s most likely diagnosis is most accurate? e. Treatment is immobilization after closed reduction. Diagnosis may be difficult in children, which contributes to a high rate of perforation (20–50%). Dietary changes in children often yield a rapid response, but noncompliance in adolescents is common and may lead to growth failure and delayed sexual maturity. Common signs and symptoms include fever, diaphoresis, and flushed appearance; tinnitus; vomiting; headache; lethargy, restlessness, coma, and seizures; hyperpnea; and dehydration. Inheritance is most commonly autosomal dominant. Presenting signs and symptoms of MCAD include hypoglycemia, encephalopathy and liver dysfunction. Clinical features depend on the CO-Hb level. This results in diminished digestion and absorption of nutrients. The answer is C [III.B.4.b]. Progressive muscular weakness starting at any time during childhood 2. Extracranial causes a. Head computed tomography (CT) scan to evaluate for acute CNS injury, such as hemorrhage or trauma. A previously healthy 3-year-old boy presents with lethargy, pallor, and bloody diarrhea. Alloimmune hemolytic anemia occurs when antibodies from someone else are directed at the patients’ RBCs and is most commonly caused by newborn Rh and ABO hemolytic diseases. Hereditary spherocytosis is the most common inherited abnormality of the 495 RBC membrane and occurs predominantly in persons of Northern European ancestry. Lyme Disease A. Patients can have microcephaly, cleft lip/palate, liver disease, syndactyly of the second and third toes and holoprosencephaly. High-risk areas reflect the natural habitat of ticks of the Ixodes species, which are the vectors for Lyme disease transmission. Any cause of muscle weakness or sensory loss, such as spinal cord lesions or acute disorders of the motor unit [e.g., Guillain–Barré syndrome, see section VII.D], can lead to an unsteady gait. Chemical conjunctivitis is most often secondary to 1% silver nitrate. Leukocoria IX. Antibiotic treatment with penicillin for streptococcal pharyngitis would have prevented this patient’s glomerulonephritis. Upper segment = total height minus lower segment c. Systemic symptoms of weight loss, anorexia, fever, and anemia may also occur. Adolescent girls: owing to poor diet, rapid growth, and loss of iron in menstrual blood 2. The cause is trauma, including injury from contact lens use. Kamil, MD, Pediatric Nephrologist at Cedars-Sinai Medical Center and Harbor-UCLA Medical Center, Health Sciences Clinical Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Deborah Lehman, MD, FAAP, Professor of Clinical Pediatrics, Assistant Dean for Student Affairs, David Geffen School of Medicine at UCLA, Los Angeles, California Calvin G. The initial presentation is usually a painless soft tissue mass. Recommendations for routine immunizations are updated annually. A 12-year-old boy presents with severe arthritis of the hips and sacroiliac joints. It is not otherwise part of the routine evaluation of children with short stature. Subcutaneous nodules, although rarely seen, are associated with severe cardiac involvement. Encephalopathy within 7 days after DTaP/Tdap vaccine c. Surgery to remove the primary tumor is performed by limb amputation or limb salvage procedures after initial chemotherapy. Similarly, although sickle cell disease also causes hematuria, it would be an uncommon presentation in a neonate. Atlantoaxial instability is caused by an unstable joint and is characterized by excessive mobility and potential for dislocation. Sulfasalazine and other 5-aminosalicylic acid (ASA) drugs are effective for mild disease, especially for UC, and can also prevent relapses. Infantile hydrocephalus The response items for statements 12–14 are the same. Diagnosis is confirmed with low serum vitamin B12 levels. Cervical and thoracic myelomeningoceles are not associated with hydrocephalus. Plasmapheresis lowers the level of AChR antibodies. Musculoskeletal anomalies include butterfly vertebrae and broad thumbs. To the maximum extent permitted under applicable law, no responsibility is assumed by the publisher for any injury and/or damage to persons or property, as a matter of products liability, negligence law or otherwise, or from any reference to or use by any person of this work. Although there are multiple definitions, FTT is a term most commonly used to describe a growth rate less than expected for a child, and is of particular concern when a child’s weight crosses two major percentile isobars on a growth chart. Note: We have removed this book from our servers due to copyright issue. Risk factors for first febrile seizure include having a first- or second-degree relative with a history of febrile seizures, neonatal hospital stays >30 days, developmental delay, attendance at daycare, high frequency of febrile episodes, peak body temperature and recent vaccination. If the nomogram predicts hepatitis, the antidote, N-acetylcysteine (NAC), a glutathione precursor, is indicated. Most cases are due to autosomal dominant mutations in genes that encode for chains of type I collagen. Chemotherapy may convert a previously unresectable tumor to one that is amenable to surgery. Bounding pulses are typically seen when there is significant reversal of blood flow, such as a large patent ductus arteriosus (PDA) or severe aortic regurgitation. If the patient is intravascularly volume depleted, the intravascular volume should be restored first with appropriate IV fluids, and then total fluid intake should be restricted to the patient’s insensible losses (approximately 300 $mL/m^2/day$) plus output (urine, stool) replacement. Brainstem injury C. 5th Ed. Philadelphia: Lippincott Williams & Wilkins, 2014. Infants of diabetic mothers have a greatly increased risk of renal vein thrombosis. Girls with Turner syndrome are usually diagnosed in childhood after an evaluation for short stature, or during adolescence after an evaluation for delayed puberty. This is a common and benign condition usually presenting in the first 2 years of life. Patients have mild microcytic anemia. Emotional lability (impulsivity, obsessive compulsive symptoms, aggression) is common and may precede signs of chorea. The pain does not interfere with pleasurable activities or sleep and has no consistent temporal correlation to activity, meals, or bowel patterns. Growth A. Antibiotic therapy should be avoided to prevent development of resistant organisms. Causes of prenatal-onset proportionate short stature include a. Amoxicillin is not recommended given that this is a viral infection. Patient monitoring should include daily weights, frequent blood pressure measurements, calculation of intake and output, and monitoring of electrolytes. Prior episode of bacterial meningitis B. Surgery to remove necrotic tissue within and around the pancreas may be indicated. Peripheral blood smear may show microcytic anemia, basophilic stippling, and red blood cell precursors. Pancreatic insufficiency and malabsorption are very common and require pancreatic enzyme replacement and the administration of fat-soluble vitamins (vitamins A, D, E, and K). This infant is also at risk for having significant vesicoureteral reflux because of the positive family history of kidney failure from reflux nephropathy and because of his history of prenatal hydronephrosis. Polymerization of Hgb results in a distorted RBC shape (sickled) that leads to decreased RBC life span (hemolysis) and occlusion of small vessels, resulting in distal ischemia, infarction, and organ dysfunction. Median life expectancy is in the 50s. Autosomal dominant disorder caused by mutations in NF1. TPN may be required if a large segment of the bowel is resected. Postictal state from status epilepticus B. Etiologies a. As with enteropathy, hypoproteinemia, edema, and FTT may develop. Mild unconjugated bilirubinemia occurs associated with stress or poor nutrition. Gastroesophageal Reflux IV. Management includes restricting galactose intake and a lactose-free diet. Therefore, children who receive the HIB vaccine at 15 months of age or older do not require additional doses, no matter how many doses were given before 15 months of age. Although abdominal ultrasound or CT scan often aids in diagnosis, the patient may undergo an appendectomy without having a definitive diagnosis in an attempt to prevent perforation. Vaccine is inactivated. No treatment is needed for patients who have wellcompensated hemolysis. Allergic conjunctivitis is most typically seasonal, often accompanying seasonal allergic rhinitis. Chronic pancreatitis b. Shock III. Allergic conjunctivitis 1. Lee Miller 12 Preface We created this text to provide medical students and physicians with a comprehensive, yet hopefully very manageable, overview of the basic principles of pediatric medicine. Classic signs include cough, coryza, conjunctivitis, and Koplik spots (an enanthem with white lesions on a reddened base on the inner cheek), followed by a classic rash (an exanthem described as morbilliform, starting on the face and moving caudally). A GnRH stimulation test may be warranted in addition to the following: a. Slipped capital femoral epiphysis (SCFE) is slipping of the femoral head off the femoral neck. The clinical presentation is variable. patient’s disorder? Evaluation a. 3 years of age = 1.3 3. Patients with infantile spasms typically present with brief, myoclonic jerks, lasting 1–2 seconds each, occurring in clusters of 5–10 seizures spread out over 3–5 minutes. Patients with saltwasting CAH have a metabolic acidosis, whereas patients with pyloric stenosis have a metabolic alkalosis.) 2. This is a myeloproliferative disorder seen typically in older adults. Females are twice as likely as males to develop dermatomyositis. Presence of AChR antibody titers G. Some children also have minor GI disturbances in motility, or in their sympathetic or parasympathetic nervous systems, which make them susceptible to nonorganic CAP. Classic clinical presentation of proximal muscle weakness with associated characteristic rashes 2. Your physical examination confirms these findings, along with an enlarged left cervical lymph node measuring 3 cm in diameter. Causes include inadequate dietary intake (e.g., from a strict vegetarian [vegan] diet), an inherited inability to secrete intrinsic factor (juvenile pernicious anemia), or an inability to absorb vitamin B12 (e.g., Crohn disease, short gut syndrome). The murmur disappears either when the child is supine or turns his or her head or when placing a finger over the internal jugular vein. The remainder of the physical findings would not be characteristic of pericarditis. A bruit would be unusual in a 7-year-old, but there are rare circumstances where a bruit may be appreciated. Diagnostic studies. Cytomegalovirus (CMV) retinitis B. Zinc E. Atlantoaxial instability (a complication of Down syndrome), aortic dissection (a complication of Marfan syndrome), or delayed puberty is not associated with achondroplasia. Antibiotics are not indicated because the infant likely has an inborn error of metabolism. 497 e. Treatment is based on tumor type and typically includes surgical resection, chemotherapy, and, sometimes, radiation therapy. >7 years of age = 1.0 d. Clinical features include hepatosplenomegaly, cytopenias (anemia, thrombocytopenia, and leukopenia), and bone disease (osteopenia, lytic lesions, and fractures). S trait is defined as having only one gene for Hgb S (heterozygous). Mucus-altering therapies are ineffective in this condition. Approach to Unsteady Gait A. Multiple fractures in different stages of healing e. The answer is A [VI.A, VI.B.4, VI.D, VI.E, VI.G, and Table 16-6]. Hemorrhagic conjunctivitis is a dramatic presentation of pediatric red eye in which the child presents with both conjunctivitis and subconjunctival hemorrhage. He presents to the emergency room with complaints of headache and a fever with a temperature of 103.5°F. Sexual behavior in young children raises red flags for abuse. 205 FIGURE 6.2 Differential diagnosis of short stature. Submersion-related injuries are the leading cause of death among children ages 1– 4 years in the United States. Patients with glycogen storage disease type 1 may have hepatomegaly and hypoglycemia, but they will not have urine-reducing substances, and they do not have a special propensity to get E. Rationale for vaccine: Pneumococcal (Streptococcus pneumoniae) disease causes more deaths in the United States each year than all other vaccine-preventable illnesses combined. Continued fructose ingestion can result in failure to thrive, as well as liver and kidney disease. Head growth abnormalities include abnormalities in size (microcephaly/macrocephaly) or shape (deformational plagiocephaly/craniosynostosis). FGR: Fetuses who develop at less than their expected growth potential, with an estimated fetal weight below the 10th percentile for gestational age, have FGR. Which of the following findings on physical examination or laboratory analysis would best correlate at this time with the finding of coronary artery aneurysms on echocardiogram? Cleaning agents c. Evaluation of pathologic short stature 1. History obtained from caregivers typically identifies the poison. The answer is B [V.A]. Risk of serious and life-threatening hemorrhage is lifelong. There is an association with cryptorchid testes. Treatment includes multiagent chemotherapy followed by surgical excision, when possible. The clinical presentation includes a diarrheal prodrome (often bloody and may be severe) followed by the sudden onset of hemolytic anemia, thrombocytopenia, and AKI. Cystinuria 5. Kwashiorkor is less common and is seen in the parts of the world in which starches are the main dietary staple. Defects in neuronal migration (e.g., gyral anomalies, agenesis of the corpus callosum) 4. FTT should be distinguished from isolated short stature, in which height is the most abnormal growth parameter (see Chapter 6, section I). History of mental illness 4. Daily blood glucose measurements using a glucose meter before all meals and at bedtime. Wilson disease should be considered in a child who presents with new-onset behavior problems and difficulty learning. Plasma ammonia concentration >200 μmol/L 3. Therapy includes treatment of hypertension, use of ACE inhibitors (even in patients who have proteinuria without hypertension), sometimes with the addition of an angiotensin receptor blocker to slow the progression of renal disease, and eventually renal transplantation. 1. A 14-year-old boy with mental retardation has been referred to you. Urticaria (Hives) VII. Management Historically, infection was the leading cause of death due to impaired splenic function. On examination, you note a normal respiratory rate and no evidence of respiratory distress but are surprised to hear inspiratory rales at the bilateral lung bases. Incidence is approximately 1 in 2500 live births. Figure 20-1 shows the link between some typical physical findings and associated toxins. Gastroesophageal reflux (GER) is the normal physiologic state in which stomach contents move retrograde into the esophagus. Fluid resuscitation, nasogastric suctioning, and broad-spectrum parenteral antibiotics should be administered. Further evaluation to rule out other causes of anemia is necessary in patients with anemia unresponsive to iron. Cholesterol and lipid screening 1. Simple virilizing CAH (i.e., only the glucocorticoid pathway is affected, resulting only in cortisol deficiency). The physical examination of this patient’s eyes, including both the negative oculocephalic test and bilateral dilated nonreactive pupils, is most consistent with brainstem injury. Most adolescents with kyphosis have flexible kyphosis, in which they can voluntarily correct the rounded area. Gastric perforation and peritonitis may follow acid ingestion. Two common types are tonic–clonic and absence seizures. In girls, consider adrenal tumors, virilizing ovarian tumors (arrhenoblastomas), feminizing ovarian tumors (juvenile granulosa tumors), nonclassic CAH, and McCune–Albright syndrome. Lead C. Prognosis 1. Definitive diagnosis is by histologic evaluation of tissue obtained by open biopsy. aureus, Streptococcus pneumoniae, and Streptococcus pyogenes are the most common organisms. Lower segment = pubic symphysis to the heel b. Many inherited enzyme deficiencies result in low testosterone levels (i.e., any enzyme deficiency in the pathway of androgen synthesis in Figure 6-5). Laboratory studies reveal that the patient is human leukocyte antigen B27 (HLA-B27) positive. Particular attention must be given to the patient’s hemodynamic status. Gottron papules, in which the skin over the metacarpal and proximal interphalangeal joints (knuckles) may become erythematous and hypertrophic 3. Differential diagnosis of a red, teary eye in newborns also includes the following: a. Hepatitis A infection is caused by HAV, a picornavirus. Sickle cell disease D. Classic presentation occurs after 5 years of age, with the onset of pubic hair growth, axillary hair growth, and apocrine odor. The treatment is to actually lower the bedtime insulin dose and not to raise it. Lead poisoning is typically a chronic ingestion; however, children may also present with acute lead intoxication. Etiology of hypogonadotropic hypogonadism a. This patient is likely to have had an infection of the skin or pharynx with a nephritogenic strain of group A β-hemolytic streptococcus 60–90 days before the current presentation. Management is aimed at eradicating B. Enterocolitis implies involvement of the colon, and typically presents acutely with diarrhea, rectal bleeding, mucus in the stool, abdominal distension, and irritability. A previously healthy 3-year-old girl presents with a 2-week history of progressive facial edema. Almost all (95%) are benign. To prevent SIDS or ALTE, co-sleeping is not recommended. If diagnosed with a UTI, this infant has an increased risk of having vesicoureteral reflux as compared with an infant without a UTI. Treatment of the subluxation is to reduce it by simultaneously flexing the elbow and supinating the hand. This leads to tighter binding of the remaining oxygen bound to hemoglobin and impaired release of oxygen to tissues. A previously healthy 13-year-old boy presents with a 2-week history of nonproductive cough and low-grade fever. Lovell and Winter’s Pediatric Orthopaedics. Therefore, with liver disease, synthesis of clotting factors is often diminished, with the vitamin K– dependent factors most severely affected [see section IV.C.1.a]. Gastric decontamination a. Anemia II. Electromyography is not helpful in making the diagnosis. Gastroesophageal Reflux A. Isovaleric acidemia. Etiology 1. Specific poisonings 1. Primary glomerular disease (e.g., IgA nephropathy) and systemic diseases (e.g., systemic lupus erythematosus) are less common causes of nephrotic syndrome in children. Collapse or shocklike state within 48 hours after prior vaccination 3. Improvement is often noted within 48 hours. Once the airway, breathing, and circulation are stable, further diagnostic workup may begin. The inflammatory process may involve all layers of the heart (pancarditis), including the epicardium, endocardium, myocardium, and pericardium. Sideroblastic anemia may be inherited or may be acquired as a result of drugs or toxins (e.g., isoniazid, alcohol, lead poisoning, chloramphenicol). Pneumococcal vaccines (PCV-13 and PPSV-23) a. 620 4. Acute acalculous cholecystitis is usually caused by infection (e.g., Salmonella, Shigella, Escherichia coli), but may also be seen after abdominal trauma, burns, or vasculitis. There is no association with malignancy in pediatric dermatomyositis (in contrast to adult disease, in which malignancy develops in 25% of patients). Abnormal Tearing V. Physical examination reveals mild hypertension, pale mucous membranes, abdominal tenderness, and a petechial skin rash on the trunk and extremities. Abnormalities in the development of the diaphragm can allow herniation of the abdominal contents into the thorax, which in turn impairs appropriate growth and maturation of the lungs. The onset of the headache is preceded by transient visual changes or unilateral paresthesias or weakness. This causes hyperventilation that may overcompensate for metabolic acidosis produced by 706 the salicylate (it is a weak acid), resulting in a respiratory alkalosis. Specific causes of abdominal pain 1. Congenital glaucoma is increased intraocular pressure occurring at or soon after birth. Head circumference is 2–3 standard deviations below the mean for age. Developmental Screening VIII. Bone scan, chest CT scan, and bone marrow aspiration and biopsy are performed to assess for metastatic disease. 503 II. Some patients with inflammatory bowel disease are also human leukocyte antigen B27 (HLAB27) positive and have involvement of the axial skeleton, which is clinically indistinguishable from ankylosing spondylitis. Bardet–Biedl syndrome. Intravenous immune globulin therapy for a presumed diagnosis of immune thrombocytopenic purpura C. Older children may swallow blood during an episode of epistaxis and can present with emesis that mimics a GI bleed. The pain may be varied in character, including paroxysmal, dull, sharp, or cramping. Children with perinatally acquired human immunodeficiency virus (HIV) infection are frequently asymptomatic in the first year of life but then develop recurrent common bacterial infections, including otitis media and pneumonia. Although vesicoureteral reflux may not be as serious as previously thought, the highest risk group for renal parenchymal damage is the child younger than 2 years with vesicoureteral reflux and pyelonephritis. Highly contagious and lasts for 2–3 weeks c. Activated charcoal does not bind to iron. In the United States, the incidence is as high as 1 in 100 individuals. Orbital floor fracture (“blow-out” fracture) 1. Treatment is dictated by the underlying cause of the NS. For severe episodes of hyperammonemia, hemodialysis and/or liver transplant may be indicated. A supracondylar fracture occurs most commonly in a child younger than 10 years who falls on an elbow or on an outstretched hand. Acquired Heart Disease VI. Gilliam, MD, FAAD, Pediatric Dermatology, Palo Alto Medical Foundation, Volunteer Assistant Clinical Professor of Pediatrics and Dermatology, UCSF School of Medicine, San Francisco, California Heather Hindo, MD, FAAP, Pediatrician, Asian Pacific Healthcare Venture, Inc., Los Angeles, California Rajashri Shuba Iyengar, MD, MPH, FAAP, FAAAI, Alto, California Division of Allergy and Immunology, Palo Alto Medical Foundation, Palo Elaine S. A type III fracture is in the physis and extends into the epiphysis. Delayed growth and puberty may also be present. Antibiotics are not effective for viral bronchiolitis. It is associated with chronic active hepatitis B infection, biliary atresia, glycogen storage disease type I, α1-antitrypsin deficiency, and hereditary tyrosinemia. Associated symptoms include nausea (especially on awakening), hoarseness, halitosis, and wheezing. Allergic Rhinitis III. Weight, height, head circumference (until 2 years of age), body mass index (starting at 2 years of age), and sexual maturity are monitored routinely during well-child care to assess for adequacy of growth and development. Which of the following treatments would be the most appropriate at this time? It may be a postinfectious inflammatory reaction. The incidence is 1:3500 live births. As many as 50% of all neonates or infants experience transient jaundice, the majority of whom have unconjugated (indirect) hyperbilirubinemia. Despite the success of these different medications in suppressing seizures, children often develop moderate to severe intellectual disability. V—vertebral defects 2. Patients may develop red-colored urine from the ingestion of exogenous pigments, such as those found in beets, and from medications, such as phenytoin and rifampin. Benign rolandic epilepsy is the most common partial epilepsy during childhood, accounting for 15% of epilepsy. Endoscopy with biopsy to detect inflammation may be indicated when the diagnosis is uncertain. Hypertrophic pyloric stenosis 1. Physical examination may reveal Murphy sign (i.e., palpation of the right upper quadrant during inspiration elicits intense pain and causes the patient to stop inspiratory effort). Hemorrhage may result from deficiency or dysfunction of any of these elements. is rare. The absorptive capacity of the bowel depends on the length and location of the resected segment and on the condition of the residual gut. Improper eye alignment b. Myasthenia gravis is caused by antibodies against the acetylcholine receptor (AChR) in the postsynaptic membrane of the neuromuscular junctions. Testicular tumors may be derived from germ cells or stromal cells; 70% of childhood testicular tumors are germinomas. A clue to fat malabsorption may be fecal fat globules seen on examination of stool using the Sudan stain, although the gold standard remains analysis for fecal fat on a 72-hour stool collection. Most children are hospitalized for initial treatment, although a relatively asymptomatic child with a reliable caregiver can be followed carefully as an outpatient. Random GH level should not be measured outside of the neonatal period, as most GH is released in a pulsatile fashion. If the patient were sexually active, a nucleic acid amplification test (NAAT) for gonorrhea and chlamydia would be indicated. Description: Second edition. Causes are extensive and involve multiple organ systems, because many conditions that produce neonatal respiratory distress are not primary diseases of the lungs (Figure 4-2). Fractures of the posterior or first ribs, sternum, scapula, and vertebral spinous process 3. In cases of CNS tumors, GH should not be used until the tumor has been treated. The spectrum of disease severity ranges from hip joint laxity to frankly dislocated hips. The answers are D, E, and B, respectively [, sections VI.B and VI.C]. PK deficiency is an autosomal recessive disorder that results in decreased production of PK isoenzyme leading to ATP (adenosine triphosphate) depletion and decreased RBC survival. The answer is B [, section III.B.2].The signs and symptoms of depression in an adolescent include behavioral, psychological, and physical manifestations that impair the teen’s ability to function normally. They note that during the past week, she has had brief jerking episodes, lasting 1–2 seconds each, with sudden arm extension followed by flexion of the head. Children with perinatally acquired HIV infection do not develop Kaposi sarcoma in childhood, which may be a presenting sign in adults with a new diagnosis of HIV infection. Relative polycythemia refers to an apparent increase in RBC mass caused by a decrease in plasma volume. Signs include cutaneous and internal organ bleeding. The answer is C [, Table 8-1]. Patients usually present with hematuria, proteinuria, and hypertension after an infection of the skin (sometimes up to 28 days after impetigo) or pharynx (usually 10–14 days after pharyngitis) with a nephritogenic strain of group A β-hemolytic streptococcus. Mild dehydration (3–5%) is associated with normal pulse rate and quality, flat fontanelle, dry mucous membranes, normal skin turgor, normal capillary refill, and normal to decreased urine output. GSD type II (Pompe disease) is caused by a deficiency of the lysosomal enzyme αglucosidase. The headaches awaken the patient in the early morning hours. Asymmetry of the infant head shape not associated with premature suture closure 2. Alkaline phosphatase may be elevated in biliary disease, although it may also be elevated in children because of rapid growth; in bone, kidney, and intestinal disease; and in trauma. As little as 20 mg/kg of iron is toxic. Myopia, lens dislocation, and retinal detachment 2. 544 2. HgbA2 and Hgb F may also be present in small amounts. Platelet abnormalities may be quantitative (i.e., decreased or increased in number) or qualitative (i.e., intrinsic abnormality in function). This patient’s presentation with fever, toxic appearance, muffled speech, tripod positioning when seated, drooling, and neck hyperextension all suggest epiglottitis as a possible diagnosis. During the first year of life, the recommended daily dose is 400 IUs. To meet the daily recommended dose, formula-fed infants would need to ingest 1 L of formula/day. Fatty acid oxidation defects can have variable effects on different tissues and organ systems, but typically affected children present with hypoketotic hypoglycemia at different ages. This book is protected by copyright. Bluish swelling in the medial canthal area may be apparent and represents fluid sequestered within the distended nasolacrimal sac. The answer is B [VII.C.2, VII.D.1–2, and VII.F.1]. Which of the following is true regarding this patient’s management? History of one of the following moderate- or high-risk medical conditions: nephrotic syndrome, human immunodeficiency virus (HIV), Kawasaki disease with history of coronary aneurysms, and end-stage renal disease c. Failure to thrive 2. Further questioning would probably reveal that this patient loses control of her bladder during the event. Single or multiple bony lesions may be present and may be painful, palpable, and associated with swelling. Although Clostridium difficile can be a cause of bloody diarrhea in children, empiric treatment with antibiotics is not indicated in this child. This patient’s clinical presentation should raise suspicion for acute rheumatic fever. Treatment includes rest, stretching, and analgesics. The initiating event is clotting that leads to consumption of procoagulant factors and resultant hemorrhage. Presence of more copies of SMN2 usually presents with a milder phenotype. If resuscitation is unsuccessful, the child’s body is referred to the medical examiner for autopsy. Microangiopathic hemolytic anemia a. No delay should occur in repeat testing, and it would not be appropriate to wait 1 week to contact the family. The answer is D [V.C]. Recurrent symptoms or chronic sequelae are rare. Studies show pancytopenia, RBC macrocytosis, low reticulocyte count, elevated Hgb F, and bone marrow hypocellularity. 6. Incubation period is 2–6 weeks, with mean symptom onset at 28 days. Total parenteral nutrition with protein and lipids should be started. This disorder occurs equally in males and females, but adolescent females require treatment eight times more frequently. Teratomas may be benign or malignant. Antibiotics are not indicated for croup. Renal Tubular Acidosis (RTA) A. Antibiotics should be given for 4–6 weeks, although the length of treatment varies depending on the organism and patient response. 594 VI. In contrast, adult polycystic kidney disease (question 13) is inherited in an autosomal dominant pattern, and there is considerable variability in its severity, ranging from mild microscopic hematuria to severe hypertension and renal failure. Ankylosing spondylitis. The majority of patients who develop the disease at age 9 years or older go on to develop osteoarthritis in the affected hip as adults. Like the previously described vasculitic diseases (e.g., KD, SLE, HSP), these disorders are characterized by constitutional symptoms and inflammation of blood vessels. Thyroid tests (TSH and T4) should be performed to rule out hypothyroidism. Dietary restriction is the preferred treatment for malabsorption from pancreatic insufficiency. Normal PT, platelet count, and platelet function assay 3. Other agents include mycophenolate, rituximab, azathioprine, methotrexate, and cyclosporine. Movement Disorders A. Accurate height and weight should be plotted on a US National Center for Health Statistics (NCHS) growth chart, along with previous growth points, to assess the child’s growth pattern. The answer is A [IV.C]. Early diagnosis and treatment are ixmportant to avoid intestinal perforation, meconium peritonitis, and volvulus. FIGURE 17.1 Scoliosis. After 15 months of age, the vaccine confers lasting immunity. ADPKD is a common genetic disorder (affecting about 1 in 500 individuals) that usually presents in adulthood (20–40 years of age). Her fever has lasted 6 days, and her parents have noticed eye redness, a truncal rash, and swollen lips. RMSF is a lifethreatening infection and empiric treatment should be given rather than only supportive care. Since licensure, HIB has become a rare cause of such infections. A 3-year-old boy presents with acute onset of fever (temperature up to 103.5°F [39.7°C]), diminished appetite, and drooling. Generalized seizures are caused by the discharge from a group of neurons in both cerebral hemispheres and are associated with an alteration of consciousness. A 5-year-old boy has a 3-day history of headache, “puffiness,” and dark-colored urine. FTT a. Ovarian failure in females 2. Bell-shaped chest 3. The Jones criteria for rheumatic fever are shown in Table 16-6. Steroids are the mainstay of therapy. Apocrine odor is a sign of adrenarche. Physical examination alone may be the basis of diagnosis. The most common type of plagiocephaly is positional plagiocephaly (Figure 1-4), associated with flattening of the occiput and prominence of the ipsilateral frontal area. Clinical findings overlap with those seen in Fanconi anemia, which should be ruled out in anyone presenting with VACTERL. It does not result in chronic joint disease and responds quickly to NSAIDs and aspirin. Aluminum, zinc, or lead does not cause the signs and symptoms seen in this patient. Order a chest radiograph to assess for pneumonia. Eight percent have S trait. General Concepts of Inborn Errors of Metabolism (IEM) VI. The finding of cherry-red lips is a late manifestation of carbon monoxide poisoning. Noonan syndrome can occur in females and has similar physical findings, but affected patients usually have right-sided heart lesions (e.g., pulmonary stenosis). It may be either congenital or acquired. Hepatic injury and necrosis 3. It appears that some cases represent clonal cellular proliferation, whereas others represent immunologic dysregulation. UC can be cured with a total proctocolectomy, but this is reserved for intractable colitis. Adult-strength ferrous sulfate tablets and iron in prenatal vitamins are the most common sources of accidental iron ingestion. Premature adrenarche 1. Nonspecific systemic features are very uncommon and include weight loss, fatigue, fever, and failure to thrive. In von Willebrand disease, factor VIII procoagulant activity is variable, but platelet function is defective because of a decrease or defect in von Willebrand factor, a protein required for platelet adhesion to blood vessel wall. Diagnosis is on the basis of clinical features, liver biopsy, and genetic testing to exclude other causes of cholestasis, such as biliary atresia. Infants with tracheoesophageal fistula should be examined carefully for anomalies associated with VACTERL (see Chapter 5 section IV.I.1.2.). Hyperglycemia, followed later by hypoglycemia 3. Other causes include soy and egg proteins. Characteristic findings involve the ocular, skeletal, and cardiovascular systems. The earlier the intervention, the better the prognosis. 460 III. Culture of synovial fluid is positive in only 60% of cases. It typically involves the lumbar region, particularly L5. Growth failure and short stature 8. Incidence of malrotation is 1 in 500 live births, with a 2:1 male predominance. 87. Incidence varies with the specific virus. The answer is B [I.F.3]. After assessing his vital signs, your nurse pulls you aside and states, “That mother has no control over her child! He is hyperactive, still in diapers, and he stinks!” You walk into the examination room and immediately notice a mousy, musty smell. This malformation is often associated with a lumbosacral myelomeningocele. Macrocytic anemia (large RBCs; high MCV) c. Evaluation of the Child With Fever III. SMA is the second most common hereditary neuromuscular disorder, after Duchenne muscular dystrophy. Nutritional iron deficiency is most common in two age groups. Bethesda (MD): National Heart, Lung, and Blood Institute (US), August 2007:42. On auscultation, a murmur consistent with mitral regurgitation is audible. Soy milk does not contain galactose, which means that an infant who is fed a soy formula will not be symptomatic, which delays the diagnosis. Diagnosis is confirmed by abdominal ultrasound, which can detect stones and a thickened gallbladder wall. Reprinted with permission from Mongan P, Soriano SG, Sloan TB, Gravlee GP. Metabolic derangements (e.g., cystic fibrosis, hypothyroidism, galactosemia) 3. Epiglottitis is now a very uncommon infection as a result of the successful immunization of children against Haemophilus influenzae type b; however, epiglottitis may still occur secondary to infection with streptococcal and staphylococcal species. DIC is a secondary phenomenon that occurs in response to local factors (e.g., large hemangiomas as seen in Kasabach–Merritt syndrome) and systemic factors (e.g., sepsis, hypothermia, malignancy, heat stroke, snakebite, burns). These losses include both measurable forms (sensible losses), such as urinary losses, and less readily measurable but still clinically significant forms (insensible losses), such as losses from the skin, lungs, or gastrointestinal tract. Abdominal ultrasound and CT imaging can be used to visualize abscesses and thickened bowel walls. Niemann–Pick disease 2. Etiology a. Factor VIII disorders include two inherited disorders, hemophilia A and von Willebrand disease, which are described in more detail below. A type V fracture describes a crushing of the physis. 99. After surgery, the infant is in a cast for 2–3 months. Prognosis is worse with larger defects, presence of liver herniation into the thorax, more severe lung hypoplasia, and pulmonary hypertension. Bony disease results in an Erlenmeyer flask appearance of the distal femur on radiographs. This patient’s red-colored urine may have resulted from eating beets the previous day. It is also associated with dactylitis (digital swelling), nail pitting, and onycholysis. Macrovascular complications are usually seen in adulthood and include atherosclerotic disease, hypertension, heart disease, and stroke. A 7-month-old male infant presents with failure to thrive, and he has had two previous episodes of pneumonia. Timing of vaccine: All children aged 11–12 years should receive the two-dose series of HPV, with the second dose administered 6 months after the first. Initial treatment includes nonsteroidal anti-inflammatory drugs (NSAIDs) and rest. Children refuse to bend forward, and young children may refuse to walk. There are four stages of iron toxicity (Table 20-5). Patients usually appear relatively well. Patients with Type II OI are the most severely affected, and rarely survive beyond the first weeks of life. DNA testing is normal. While this patient’s current weight is approximately twice her birth weight, on average, infants triple their birth weight by 12 months of age. burgdorferi into local and distant tissues, and in later stages by a systemic inflammatory response against the spirochete. Potential complications of this disorder include cord compression caused by foramen magnum stenosis, obstructive sleep apnea, and orthopedic problems such as genu varum and back pain caused by lumbar lordosis during late childhood. In childhood dermatomyositis, unlike in adult dermatomyositis, there is no association with malignancy. Only 50% of patients have complete symptom resolution during childhood, and at least 25% of patients go on to have abdominal pain as adults. Hypocalcemia The response options for statements 12–14 are the same. Based on the likely diagnosis, which of the following would you also expect to find on further evaluation? Key point: Sepsis is much more common than metabolic disease. Laboratory evaluation and diagnosis 218 a. Girls have a higher incidence of CPP than boys. By 2–3 years of age, patients develop pallor, weakness, and splenomegaly, as spherocytes are trapped in the spleen and destroyed. Other Rheumatologic Conditions of Childhood Disorders that are rare in children and adolescents are marked with an asterisk (*). Developmental delay, intellectual disability, and attention deficit hyperactivity disorder 3. Wilson, MD, Associate Program Director, Pediatric Neurology, Assistant Professor of Pediatric Neurology and Psychiatry, David Geffen School of Medicine at UCLA, Los Angeles, California Lauren J. (Lee Todd) editor. Congenital diaphragmatic hernia would likely have been apparent on chest radiographs. Note that this can be higher in neonates but is typically 2 weeks is most consistent with Gianotti–Crosti syndrome (GCS). Outbreaks of pertussis with significant mortality continue to occur in the United States. 13 Acknowledgments We would like to recognize the individuals who have made tremendous impacts on our medical education and on our professional growth and development. Pathologic short stature. Brachial plexus (C5–T1) injury most commonly occurs as a result of birth trauma from excessive traction on the neonate’s head, neck, or arm, which stretches the nerves of the brachial plexus. Polycythemia Review Test Answers and Explanations Chapter 5 Genetic Disorders and Inborn Errors of Metabolism I. Sources of lead include ingestion of lead-based paint chips, water carried by 707 outdated lead pipes, improperly glazed or foreign-made ceramic food or water containers, and pica (compulsive eating of nonnutrient substances such as dirt, paint, and clay). The most common form of nephrotic syndrome in children is minimal change disease, which comprises 90% of all cases. Therefore, it would be appropriate to obtain thyroid studies. If the diagnosis of poststreptococcal glomerulonephritis is confirmed, the prognosis for this patient is excellent; complete recovery usually occurs. Hematologic abnormalities include hemolysis and DIC. Transient synovitis is a diagnosis of exclusion, and it is extremely important to rule out septic arthritis, especially if a patient is ill-appearing, given the potentially disabling consequences of missed septic arthritis (Table 17-4). Tables 1-1, 1-2, and 1-3 detail general “rules of thumb” for expected gains in weight, height, and head circumference. 508 3. The risk of recurrence is 5% after contrast reduction and 1% after surgical repair. Meconium is passed within 24 hours after birth in 90% of term infants and within 48 hours in 99% of term infants. Delayed puberty The mother of a 5-year-old boy is very concerned and shows you a note from his kindergarten teacher. β-Thalassemia major (Cooley anemia or homozygous β-thalassemia) may be caused by either total absence of the β-globin chains or deficient β-globin chain production. Intubation and mechanical ventilation with high positive end-expiratory pressures (PEEPs) are indicated for patients with respiratory failure. If the child has widespread edema, scrotal or labial edema, hypotension, or symptomatic pleural effusions, IV infusions of 25% albumin should be given initially to achieve a diuresis and to maintain the intravascular volume. Biliary enzymes a. Risk factors include a history of previous affected pregnancy, inadequate folic acid intake, and maternal diabetes. Renal and Suprarenal Tumors VI. Systemic lupus erythematosus nephritis C. The cause is unknown in up to 10% of cases. Parents should be educated and warned about the importance of compliance with medicines and how febrile episodes, vomiting, and surgical operations may require additional steroid therapy to prevent adrenal shock. Because of his presentation with hematuria at a young age, this patient will likely have persistent microscopic hematuria. The prognosis for a child with this disease is poor, even with treatment. It is the most common hepatitis virus–causing infection. Neonatal myasthenia gravis D. Severe cases are associated with oligohydramnios, pulmonary hypoplasia, and early neonatal death. 615 FIGURE 17.2 Cobb angle measurement noted on radiograph (A) and figure (B). Febrile seizures are benign events that are not generally associated with serious acute or long-term neurologic sequelae. The diagnosis of a febrile seizure is based on history, a normal neurologic examination, and the exclusion of CNS infection. Stools are often dry and hard. Quality of pain. 89. The majority of patients are asymptomatic, although 10% have jaundice at birth, and later may develop splenomegaly and gallstones. The goal is to avoid aimless formula changes, inappropriate early weaning, medications, homeopathic remedies, or costly seats and straps. Infectious causes are suggested by history of contact with others who have conjunctivitis. Lower Extremity V. Carbohydrates make up approximately 50% of a typical diet and are converted by the body to glucose and other monosaccharides. 537 538 VI. Diagnosis is by abdominal radiographs that reveal intestinal distension with minimal air–fluid levels. In addition to the characteristic features of anemia, patients may also have anorexia, a smooth red tongue, and neurologic manifestations (such as ataxia, hyporeflexia, and positive Babinski responses). Most infections are asymptomatic, although severe pulmonary infections can occur in healthy persons. Important short points and various tables that simple things and make it easy to learn pediatrics. 100. On examination, you note diffuse wheezing and moderate subcostal retractions. End plate changes and disk space narrowing are noted on radiographs. Bone marrow transplant is curative and is considered for children with severe manifestations. MMR vaccine 30 a. Atresia is surgically corrected by duodenoduodenostomy. You will be required to select one answer for each statement in the following set. The answer is A [III.B]. Improving symptoms and decreasing ESR/CRP generally indicate response to intravenous antibiotics, at which time oral antibiotics can be used to complete the antibiotic course. The most severely affected infants have a maternal history of oligohydramnios secondary to nonfunctioning or poorly functioning kidneys in utero. Mammalian Bites A. Physical examination 1. It is the primary manifestation of JDM. Diagnosis of mumps is made by culturing the virus from urine or by serologies, but not from parotid gland aspiration. Incidence. Symptoms and signs: severe watery conjunctival discharge, hyperemic conjunctiva, chemosis (conjunctival edema), preauricular lymphadenopathy, and typically a foreign body sensation caused by corneal involvement 2. Male puberty a. Laboratory findings include hypochromia and microcytosis with target cells and anisocytosis (excessive variation of size of RBCs) on smear. Hydrops of the gallbladder C. Heparin may be useful if the underlying defect cannot be corrected. Other SS diseases include sickle cell–thalassemia disease (with clinical features similar to SS disease) and sickle cell–hemoglobin C disease (Hgb SC disease) caused by the inheritance of both Hgb S and Hgb C genes. Enteropathy, which is characterized by progressive onset of diarrhea, vomiting, irritability, and abdominal pain. Sometimes I’d be reading and then see an abbreviation that I had no idea about and I’d have to go back several pages even to find where they mentioned the original meaning of it. Rheumatoid factor–negative polyarticular JIA D. Clinical features of anemia (Table 13-1) D. Chronic osteomyelitis results from a nidus of residual infection, such as a sequestrum (i.e., focus of necrotic bone) or an involucrum (i.e., formation of new bone or fibrosis surrounding the necrotic, infected bone). Instillation of a topical antibiotic prevents bacterial superinfection. Initial management should include fluid resuscitation. Treatment includes eyelid hygiene, in which eyelids are scrubbed twice daily with baby shampoo. Type of vaccine: HIB vaccine is a conjugate vaccine (inactivated) with H. They are bought the most and mainly used for the preparations of USMLE tests. They are the best books and would save a lot of your precious time because they don’t write lengthy paragraphs. Witcoff, MD, Stanford, California Pediatric Pulmonologist, Past Clinical Associate Professor of Pediatrics, Stanford School of Medicine, Derek Wong, MD, FAAP, FACMG, Division of Medical Genetics, Associate Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California 16 Contributors to the Previous Edition Lloyd J. Ocular Examination and Vision Screening A. A 10-year-old boy develops a headache, fever, and rash approximately 2 weeks after camping with his family. Imaging should include abdominal ultrasound with Doppler evaluation of the hepatic vessels. Its peak age of presentation is 2–7 years. Primary NS, which refers to cases that are not a consequence of systemic disease. Sense of restless agitation d. Severity 1. 84. In most states, health care personnel have a legal obligation to report suspected child abuse or neglect to appropriate protective service or law enforcement agencies. (See Chapter 2, Table 2-2 for a description of the Moro reflex.) Eventually, the arm is held in internal rotation with the elbow extended, forearm pronated, and the wrist held in flexion. An example is the triad of arthritis, urethritis, and conjunctivitis (Reiter syndrome), which is most commonly triggered by Chlamydia trachomatis. In young children, the headache itself may be absent, but there is a prolonged, albeit transient, alteration of behavior that manifests as cyclic vomiting, cyclic abdominal pain, or paroxysmal vertigo. His chest radiograph will be normal because he has improved clinically. Neuromuscular junction disorders: congenital and neonatal myasthenia gravis E. In neonates and infants, urine for culture must be collected by suprapubic aspiration of the urinary bladder or by a sterile urethral catheterization. Patients with aqueductal stenosis have the best cognitive outcome. Treatment includes appropriate intravenous antibiotics. Myotonic dystrophy is a CTG trinucleotide repeat disorder with autosomal dominant inheritance, with symptoms becoming more severe with each successive generation (genetic anticipation). Sexually abused children typically present with multiple nonspecific complaints, including abdominal and urogenital symptoms. Corticosteroid therapy is contraindicated. She is irritable and is crying, although she stops crying when held by her parents. Liver Tumors IX. The pain often occurs in intervals followed by periods of calm. Extraintestinal complications are more common in CD than in UC and are noted in Table 10-4. Topical corticosteroids and topical vitamin D analogues are not appropriate for this condition but might be tried for psoriasis of the nails. Prolonged bleeding after simple surgical procedures, circumcision, trauma, or dental extraction d. Chronic Kidney Disease (CKD) and End-Stage Renal Disease (ESRD) XII. Serum albumin and serum transaminases to assess nutritional status and liver disease d. 221 5. Sporadic D. In juvenile myasthenia gravis, treatment involves the following: a. Bilirubin collects in the skin, conjunctiva, and mucous membranes and becomes clinically evident when the total bilirubin level exceeds 3 mg/dL. Ten percent of infants with GER have GERD. Boys present with either feminization (gynecomastia) or with premature onset of pubic hair. Chemical conjunctivitis. 430 IX. Hemoglobin screening is 12.5 g/dL, and lead screening is 90% of cases, is comprised of proximal esophageal atresia with a distal tracheoesophageal fistula. HPV serotypes have been associated with cancers of the female reproductive tract but have not been associated with cancers in male patients. The cause of rheumatic fever is unknown, but the disease appears to be autoimmune in nature and may represent “molecular mimicry,” in which streptococcal antigens resemble autoantigens and trigger an autoimmune response. Patients are at risk for spontaneous pneumothorax. Complications are caused by the transmural nature of CD and include abscesses, fistulas, strictures, and adhesions. In contrast, bruises on exposed areas, such as the shins, knees, elbows, and forehead, are typically from noninflicted trauma. HPV is a rare cause of sexually transmitted disease in the United States. Homocystinuria. The untreated clinical course of Lyme disease is, therefore, divided into two stages, early and late disease. Complications a. Less than 30% of all calories should come from fats. 357 Answers and Explanations 1. PPHN, pneumothorax, gastroesophageal reflux, intestinal obstruction, recurrent herniation, and progressive sensorineural hearing impairment may occur. Peritonitis presents with diminished or absent bowel sounds, abdominal wall rigidity, involuntary guarding, and often rebound tenderness. It is characterized by inflammation of the heart, blood vessels, joints, CNS, and skin. Decreased lipase activity results in steatorrhea (fat in stool) and decreased absorption of fat-soluble vitamins (vitamins A, D, E, and K). Generally no workup is necessary as long as no other secondary sexual characteristics (clitoromegaly, pubic hair, enlarged labia minora, pink vaginal mucosa) or growth spurts are noted. Serum creatine kinase (CK) levels to screen for myopathy. To interpret the pH probe correctly, results should be correlated with the clinical presentation. Before 6 months of age, UTIs are twice as common in boys, but after 6 months of age, UTIs are more common in girls. Cerebellar dysfunction. Which of the following is correct regarding her presentation, evaluation, and management? Incidence is approximately 1:5000 live births. Kaposi sarcoma 743 20. Surgical therapy for CD is only considered for persistent obstruction, bleeding, abscesses, and fistulas that do not respond to medical therapy. Klumpke palsy is less common and is the result of a lower brachial plexus injury caused by upward traction on the arm. Seizure Disorders of Childhood A. Failure to thrive is a common presenting symptom as well. Normal Visual Development and Amblyopia III. Note that mild 33 illnesses, including febrile illnesses, are not contraindications to immunization. Painful limp d. Clinical features include a flaccid arm and an asymmetric Moro reflex. Complete resolution of symptoms usually occurs by 3 weeks. Patients usually present with multiple absence seizures, which are brief staring spells that occur without warning and are not followed by postictal drowsiness. Hepatitis B vaccine (HBV) a. Pregnancy may be prevented with high-dose oral contraceptives (morningafter pills). If elevated orotic acid, suspect ornithine transcarbamylase deficiency CBC = complete blood count; NH_3 = ammonia. Angular deformity or limb length discrepancy may occur if the growth plate is involved in the infection. Children with Gaucher disease present with neurodegeneration, splenomegaly, and bony changes (the most characteristic of which is an Erlenmeyer flask–shaped distal femur). Glycosylated hemoglobin (HbA1c) level, reflecting diabetic control for the past 2–3 months, should be checked every 3 months. Fever is absent. Valvular insufficiency or stenosis is usually delayed (usually more than 3 years after rheumatic fever) and, if severe, may require valve replacement or valvuloplasty. Patients are typically active, thin boys who are small for their age. Hemolytic anemias a. Mumps is a vaccinepreventable disease, and the vaccine is recommended for administration at 12 months of age and again at kindergarten entry, but can be given any time with at least 4 weeks following the first vaccine. Foreign body sensation 4. Consumption of clotting factors and platelets may also occur with liver disease. This patient may be diagnosed with microscopic hematuria if there are ≥10 red blood cells (RBCs) per high-power field on a single urine sample. Death usually occurs by 3 years of age. Many, but not all, children with MCD achieve a sustained remission by adolescence. Watch for hypoglycemia. Croup is the most common cause of acute stridor with cough in toddlers, and parainfluenza virus causes the majority of these infections. Pneumonia from aspiration of fluid containing mouth flora may develop after 24 hours. Quantitative assay for vWf antigen and activity (ristocetin cofactor assay) are diagnostic. Prevention of hepatitis A and B is now possible through vaccination. Chronic blood loss in the stool may lead to anemia, and significant stool protein loss may cause edema and FTT. Temperature of 40.5°C within 48 hours after prior vaccination 2. In addition, the leukemic cells demonstrate hyperploidy (>53 chromosomes). aureus and S. Central hypotonia is dysfunction of upper motor neurons (i.e., cortical pyramidal neurons and their descending corticospinal pathways). If both hips are dislocated, Galeazzi sign will not be positive as the knees will be at the same height. A booster vaccination is given at 16–18 years of age. Infantile spasms are rare in children older than 2 years of age. Other features include lymphadenopathy, hepatosplenomegaly, exophthalmos, anemia, and pulmonary infiltrates. Accessory tests should include a skeletal survey to evaluate for old or healing fractures and head and abdominal CT scans to evaluate for acute injuries. The child is unwilling to use the affected arm, but hand function is normal. ROP is the proliferation of vessels in the immature retina, seen in premature infants exposed to oxygen. There are four states categorized on the basis of the number of α-globin genes deleted (normally there are four α-globin genes per diploid cell). Typical presentation is with ecchymosis and petechiae. Constitutional delay of growth and puberty (CDGP) is defined as short stature with a delayed bone age, late onset of puberty and a minimum growth velocity of 2 inches (or 5 cm) per year. Normal physiology. Supportive care includes bed rest, hydration, electrolyte correction, analgesia, and nasogastric suctioning. Other conditions that may cause chorea include many acquired and congenital conditions, including N-methyl-d-aspartate (NMDA) receptor encephalitis, kernicterus, systemic lupus erythematosus, Huntington disease, and Wilson disease. The principal sites of demyelination are the ventral spinal roots and peripheral myelinated nerves. The patient in this scenario is immunosuppressed and is taking an anti-tumor necrosis factor (TNF) drug, which will impair her T-cell immunity. The pain is usually bifrontal but may be diffuse. Vitamin D supplementation in the breastfed infant is not necessary as long as the mother takes adequate amounts of vitamin D and calcium in her diet. Both inhaled corticosteroids and leukotriene inhibitors are effective management options for the prevention and chronic management of asthma, and these agents can be considered for her long-term controller therapy. The classic presentation includes several weeks of polyuria, polydipsia, nocturia, and occasionally enuresis. Parenteral steroids (commonly referred to as “stress doses”) are given in adrenal crisis until the patient is stabilized (50–100 mg/m^2 per day of hydrocortisone). This is excessive glucocorticoid production caused by benign or malignant adrenal tumors. Tensilon test. Myotonia is the inability to relax contracted muscles. Evaluation for clotting abnormality typically includes these screening tests: a. An apical click is associated with a bicuspid aortic valve. In most cases, at least some degree of developmental delay and intellectual disability will be present. Limb length discrepancy, limp, and osteoarthritis if DDH is not treated B. SS disease is the result of having two genes for Hgb S (homozygous). Excessive urinary protein losses lead to the hypoproteinemia of NS. IBD is increasing in frequency in children, with 25–30% of all cases of CD and 20% of UC cases presenting before 20 years of age. Type 2 DM, along with these comorbidities, are collectively known as the metabolic syndrome. Nagel, MD, FAAP, Angeles, California Clinical Assistant Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Charles E. It is important to highlight that the patient senses real pain, even in the absence of an underlying organic cause. Which of the following is the most commonly identified cause of the 484 6. The answer is E [IV.A.7 and Tables 9-4 and 9-5]. Duration of prophylaxis depends on severity of heart disease. Children under 5 years of age and those with conditions that predispose to complications of influenza are considered to be at high risk. coli–associated diarrhea. Which of the following is the most likely cause of the acute abdominal pain? Oral polio vaccine (OPV) and IPV a. Ultrasound is used to assess for DDH in young infants because the femoral head does not ossify until 4–6 months of age. Malignant potential is based on the amount of immature tissue and the presence or absence of other germ cell tumor cells within the teratoma. In a positive Ortolani maneuver, the hip can be felt slipping into the acetabulum as it is being reduced. Protein intolerance occurs in up to 8% of children. Incubation period is 45–160 days with mean symptom onset of 90 days. Mitochondrial Disorders XI. It is not useful to observe the infant feeding in the emergency department. General concepts 1. Amniotic band syndrome occurs when rupture of the amniotic sac during pregnancy results in small strands of amnion wrapping around areas of the fetus’s body, causing deformation and/or amputation. This group of disorders involves defects or deficiency in the von Willebrand factor (vWf) portion of the factor VIII complex and is the most common hereditary bleeding disorder. Pancytopenia is defined as low white blood cells (WBCs), RBCs, and platelets and implies bone marrow failure. In general, children have a better outcome from drowning because their primitive dive reflex shunts blood to vital organs, such as the heart, brain, and liver. You order an electroencephalogram, which shows a generalized 3-Hz spike-and-wave discharge pattern arising from both hemispheres. Dysostosis multiplex (bony abnormalities that include a thickened skull, malformed vertebrae, and abnormal ribs and clavicle) is found in patients with mucopolysaccharidoses (e.g., Hunter and Hurler syndromes). Low or absent plasma citrulline and high urine ototic acid level d. Kayser–Fleischer rings are dark rings that encircle the iris due to copper deposition in Descemet’s membrane. Calcinosis (calcium deposition in muscle, fascia, and subcutaneous tissue), which occurs in approximately 40% of children c. This patient meets the diagnostic criteria for Kawasaki disease based on the duration of fever and the presence of four of the five diagnostic criteria, including conjunctivitis, oropharyngeal changes, a truncal rash, and swelling of the distal extremities. Moderate to severe symptoms can be treated with anti-inflammatory (e.g., aspirin) or immunomodulatory (e.g., glucocorticoids) medications, but intravenous immune globulin is not recommended for use in JIA. Rhabdomyosarcoma should be considered in any patient presenting with a painless enlarging mass. NAC may be given intravenously as an infusion for more than 21 hours. Adult polycystic kidney disease may also be associated with cerebral aneurysms and early death. Hyponatremia can produce fatigue and neurologic symptoms; however, symptomatic hyponatremia usually presents more acutely than the several month history noted in this case. Progression of scoliosis generally occurs only during growth. Lumbar kyphosis in late childhood B. Laboratory findings include leukocytosis with a polymorphonuclear predominance. Intrinsic RBC defects that cause hemolysis include RBC membrane disorders and RBC enzyme disorders. There is a deficiency or abnormality of the structural RBC membrane protein spectrin, causing the RBC to assume its spherical shape. Rationale for vaccine: Rotavirus infections are the most common cause of severe acute gastroenteritis worldwide. Erythrocyte sedimentation rate (ESR) is typically elevated. The HPV vaccine is universally recommended for females and is optional for males. Vitamin K deficiency a. High levels are associated with visual and auditory changes, vomiting, confusion and later syncope, slurred speech, cyanosis, myocardial ischemia, coma, and death. The physical examination of this adolescent boy, in which the left testicle feels like a “bag of worms” that diminishes in size when the patient is supine, is consistent with a diagnosis of a left varicocele. Hydrocortisone is administered at a dose that sufficiently suppresses ACTH production so that androgen production decreases, but is not excessive enough to interfere with proper growth. Brown, MD, FAAP, Regional Chair of Pediatrics, Palo Alto Division, Palo Alto Medical Foundation, Palo Alto, California, Adjunct Clinical Associate Professor of Pediatrics, Stanford School of Medicine, Stanford, California Lee Todd Miller, MD, FAAP, Associate Dean for Student Affairs, Professor of Pediatrics, Director, Global Health Education Programs, David Geffen School of Medicine at UCLA, Los Angeles, California Tiffany Merrill Becker, MD, California Division of Academic Primary Care Pediatrics, Cedars-Sinai Medical Center, Los Angeles, Peter A. This patient’s physical features are consistent with achondroplasia, the most common skeletal dysplasia. Overgrowth syndromes (e.g., Sotos syndrome), in which all growth parameters are enlarged c. If the hip is dislocated, the affected femur, and therefore knee, appears shorter compared with the normal limb. Physical examination is notable for diffuse weakness, hypotonia, and hyporeflexia (i.e., diminished deep tendon reflexes). Typically, patients develop more classical signs of puberty (voice change, growth spurt, and facial hair) at the later stages of puberty. Myotonic dystrophy a. It is the most common form. Intubation and mechanical ventilation with 100% oxygen should be initiated immediately. MRI of affected bone can better delineate the tumor and its local extension. Title: Pediatrics / Lloyd J. Maintenance water and electrolyte calculations are designed to balance the usual daily losses of water and salts as a result of normal daily metabolic activities. The answers are D, C, and F, respectively [I.F.1–3]. Amblyopia is the most common cause of decreased vision during childhood and occurs in approximately 2–4% of the general population. Adenovirus infection is a cause of hemorrhagic cystitis, which often causes hematuria; however, this would be very unusual in a neonate. The disease though may present at any age. Timing of vaccination: HBV is given as a three-shot series within the first year of life. Topical antibiotics applied to the skin may prevent secondary bacterial infection. Puberty generally begins after the age of 9 years and is not considered delayed until after the age of 14 years. Coller, MD, MPH, FAAP Chief, Division of Pediatric Hospital Medicine Assistant Professor of Pediatrics University of Wisconsin School of Medicine and Public Health Madison, Wisconsin Lee Todd Miller, MD, FAAP Associate Dean for Student Affairs Professor of Pediatrics Director, Global Health Education Programs David Geffen School of Medicine at UCLA Los Angeles, California 2 Contents Cover

image Title page Copyright Dedication Preface Acknowledgments Contributors Contributors to the Previous Edition Chapter 1 Pediatric Health Supervision I. Surgical Conditions of the Newborn XIII. Rheumatoid factor–positive polyarticular JIA E. Respiratory alkalosis with an anion-gap metabolic acidosis is the most common acid–base disturbance. Neonatal hepatitis is idiopathic hepatic inflammation during the neonatal period. These patients present earlier and progress more rapidly. Clinical findings include profound hemolytic anemia beginning in infancy, marked hepatosplenomegaly, and, if untreated, bone marrow hyperplasia in sites that result in a characteristic "thalassemia facies" appearance (frontal bossing, maxillary hyperplasia with prominent cheekbones, and skull deformities). Hypovolemia, blood loss, and shock should be anticipated and treated. Radiographs show the tube in the upper part of the thorax. Depression, alcohol consumption, and obesity may lead to false positives. Daily folic acid is given to prevent folic acid deficiency. Indications for surgery include nerve impingement, persistent pain, or progression of the subluxation beyond 50% of the vertebral width. Normal variant short stature describes a child with short stature and a normal growth velocity. Retinal hemorrhages are highly suggestive of child abuse. History may reveal antenatal problems, such as decreased fetal movements in utero. Russell–Silver syndrome E. Viral infections and occasionally drugs may be causal in some patients. Which of the following statements would support this diagnosis? Adolescents, most commonly males, whose submersion injury is typically associated with alcohol or drug ingestion C. Only Hgb Bart's are formed, and in utero, this causes profound anemia, congestive heart failure (CHF), and death if not identified early enough for intrauterine transfusion to occur. Lloyd J. Rationale for vaccine: MMR immunizes against three viral diseases. Some disorders that present in infancy have less severe forms that can present later in infancy, childhood, adolescence, or even adulthood. Gaucher disease B. i. RBC morphology a. All survivors have intellectual disability (average intelligence quotient [IQ] of 50–65). Rotavirus is highly contagious and nearly every child is at risk of getting infected. Patients can have short, webbed neck with low posterior hairline, and broad chest with widely spaced nipples. At birth, physical examination of patients with duodenal atresia may reveal a scaphoid abdomen with epigastric distension. Clues to protein malabsorption include increased levels of fecal α1antitrypsin, which reflect enteric protein losses, and low serum albumin levels seen in protein-losing enteropathies. Presenting features in the first week of life include jaundice and hepatomegaly in 50% of patients. Patients with neurofibromatosis are at higher risk. Underlying disease is frequently present. FFR becomes a behavioral pattern of stool withholding that may lead to large amounts of retained feces with secondary anorectal distension, encopresis, fecal halitosis, abdominal distension and pain, anorexia, urinary tract problems (from pelvic displacement), and psychosocial problems. Although recommended during the preteen years, HPV vaccine may be administered to sexually active individuals regardless of age. New-onset seizures in a child from a developing country should include evaluation for parasite infection and neuroimaging. Physical examination should focus both on acute injuries and on identifying old lesions that may be secondary to abuse. He has had bloody stools for 4 days, and in the past 2 days he has developed fatigue and pale skin. His lung function will continue to deteriorate with time and further growth. Cardiac involvement is the hallmark and most important complication of rheumatic fever. The causes of conjunctivitis vary with the age of the patient. The major causes of mortality are infection (because of immunosuppression), renal failure, and CNS complications. Before 6 months of age, UTIs are 10 times more common in uncircumcised boys as compared with circumcised boys. shop.lww.com 10 11 Dedication To Bobbie, for her love, support, and laughter and for being a truly wonderful mother to our son; to Danny, the sunshine and pure joy of my life; and to my family and friends for their ongoing love and friendship. Appendectomy is the most common pediatric emergency operation. Some patients are responsive to treatment with vitamin B6. Positive staining is most commonly associated with trauma, but may also be associated with a bacterial corneal ulcer or with herpes simplex keratitis. Meconium plug is delayed passage of meconium (>24– 48 hours after birth) due to obstruction of the left colon and rectum by dense dehydrated meconium. The purpose of ocular examination and vision screening is early detection and treatment of pediatric ocular disease. Intravenous immune globulin is the preferred treatment in children. Hypokalemia d. Although the family history may be negative in recessive disorders, there is a 25% risk of affected siblings in subsequent pregnancies. Eighty percent of sexual abuse occurs in females. Three common types of growth disturbance include failure to thrive (FTT), short stature, and head growth abnormalities. Radiography of the lower extremity reveals an Erlenmeyer flask–shaped distal femur. The classic patient is an Asian adolescent or young adult female, with constitutional symptoms and aneurysmal dilation or thrombosis of the aorta, carotid, or subclavian arteries. The cause is likely a combination of peripheral tissue resistance to insulin and progressive decline in insulin secretion, both of which result in a hyperglycemic state. Cutaneous bleeding (e.g., ecchymoses, petechiae) b. Head circumference at birth is 25% of the normal adult head size, and it increases to 75% of the normal adult head size by 1 year of age. Incidence is 2–3.5 in 1000 live births. His father reports that his son seems to have developed a different personality. Causes of protein malabsorption a. Blasco, MD, FAAP, Director, Neurodevelopmental Program, Child Development and Rehabilitation Center, Portland, Oregon, Associate Professor of Pediatrics, Oregon Health and Science University, Portland, Oregon David Seung Chun, MD, Fellow, Pediatric Cardiology, James Whitcomb Riley Hospital for Children, Indiana University School of Medicine, Indianapolis, Indiana David A. The diagnosis of acute rheumatic fever requires evidence of previous group A β-hemolytic streptococcal infection, in addition to either two major Jones criteria or one major and one minor Jones criteria. Antiphospholipid antibodies (e.g., positive lupus anticoagulant or anticardiolipin antibodies) present in patients with SLE reflect an increased risk of thrombotic events. The GnRH stimulation test can demonstrate premature activation of the hypothalamus. Diagnosis is often on the basis of the history and physical examination. If the patient's urine then turns red or pink (vin rose) (the color of chelated iron), the challenge is considered positive, indicating a clinically significant iron ingestion. Menarche is the onset of the menstrual cycle. Which of the following will be the most appropriate treatment? Jaundice is generally absent, and quite often patients are asymptomatic. 9 8 7 6 5 4 3 2 1 Printed in China Library of Congress Cataloging-in-Publication Data Names: Brown, Lloyd J., editor. burgdorferi in the patient's serum and is the recommended laboratory approach. There is visible or palpable breast tissue only, with no other secondary sexual characteristics. Ryan J. Evaluation and management. Increased risk of colon cancer 2. Lee Miller, one of the coeditors of this book, has influenced my career immeasurably as one of the finest teachers I have known. Bone age determination (anterior–posterior [AP] film of the left hand and wrist to assess the characteristics of the epiphyses or growth plates) is helpful to compare with chronologic age (Table 6-1). Organ system diseases, including gastrointestinal diseases (inflammatory bowel disease, celiac disease), cardiac diseases (cyanotic congenital heart disease), renal diseases (renal failure, renal tubular acidosis), chronic lung diseases (cystic fibrosis, asthma), and endocrinopathies (hypothyroidism, GH deficiency, and cortisol excess; see also section I.F) b. It is mediated by neuraminidase, which functions as a toxin in the presence of a S. Diagnostic studies are only necessary when organic disease is suspected, or if the patient with FFR is failing management. Long QT syndrome is a disorder of myocardial repolarization. Elevated erythrocyte sedimentation rate C. If severe gastrointestinal symptoms are present c. Associated comorbidities may accompany type 2 DM, including hypertension, polycystic ovarian syndrome, and hyperlipidemia. If appropriate, testing for pregnancy and assessment of vaginal fluid for spermatozoa should also be performed. Deep tendon reflexes are absent in the distal lower extremities. Features of well-child care include the following: 1. Males and females appear equally affected by UC, but there is a 2:1 male-to-female ratio in CD. Distal small bowel resection (i.e., ileum) limits vitamin B12 and bile acid absorption. Achondroplasia C. Intestinal atresias are discussed in Chapter 10, section IV.C. 3. Unless the child is toxic-appearing, cultures should be obtained before administering antibiotics. Diabetes Mellitus VI. The parents report that their daughter's activity and appetite are diminished. Liver Abnormalities and Hepatitis Review Test Answers and Explanations Chapter 11 Nephrology and Urology I. The answers are E, H, D, C, F, and A, respectively [VIII.A.4, V.C.2, VIII.A.1, III.E.3, IV.C–E, Table 16-5, and I.C]. Subdural hematoma with herniation 485 D. You order an electroencephalogram, which reveals a highly disorganized pattern of high-amplitude spike and waves in both cerebral hemispheres, consistent with a hypsarrhythmia pattern. Diagnosis is based on serology. Alcohol intoxication 11. Other treatments include valproic acid, topiramate zonisamide, ketogenic diet, pyridoxine, and surgery. Other immunosuppressive agents, including methotrexate, IVIG, hydroxychloroquine, 593 cyclophosphamide, and cyclosporine are often used in combination with steroids to minimize adverse effects of long-term high dose corticosteroid use. Management includes transfusions and splenectomy for severe disease. Head injuries, caused by trauma, asphyxiation, or shaking, are the leading cause of death and morbidity from child abuse. Cow's milk protein causes the majority of these reactions. Direct damage to the gastrointestinal tract leading to hemorrhage 2. A "thumbprint sign" will be found on a lateral radiograph of the neck. Infection results in varying degrees of liver inflammation and swelling. Cholinesterase inhibitors are the mainstay of treatment. Because conjunctivitis is not present, topical antibiotics would not be indicated. No attempt should be made to neutralize the caustic agent, because the combination of acid and alkali will generate an exothermic reaction and worsen any burn. Although the rash of pityriasis rosea can also last several weeks, it is more common in older children presenting with oval pink scaling plaques on the trunk and extremities. Caucasians and first-born male children are affected more commonly. 447 Answers and Explanations 1. The shorter the headache duration, the less likely a serious disorder is responsible. aureus, should be started after blood and joint cultures are obtained. Infants who remain symptomatic past 1 year of age and older children are unlikely to have spontaneous resolution of GERD and usually require long-term medical management or surgery. The neonate is hypotonic with diminished deep tendon reflexes. Boys: No testicular enlargement by 14 years of age. Increased free erythrocyte protoporphyrin may be noted. Adrenocorticotropic hormone (ACTH) intramuscular injections for a 4- to 6week period are effective in 50-70% of affected patients. Unlike the other major criteria, the onset of chorea is usually several months after the other manifestations. This age group is more susceptible to overt disease owing to rapid growth. Therapy includes treatment of the underlying cause and transfusions of fibrinogen, FFP, and platelets as needed. Laboratory analyses a. Oral feedings are usually restricted to minimize pancreatic stimulation. Nonprogressive encephalopathies: cerebral malformations, hypoxic–ischemic encephalopathy, intracranial hemorrhage, and trauma (e.g., spinal cord injury) 3. This autosomal recessive disorder is characterized by pancreatic exocrine insufficiency, failure to thrive (FTT), short stature, neutropenia, and sometimes pancytopenia. Primary hypothyroidism e. Referral to a pediatric endocrinologist for additional workup may well be indicated once hypothyroidism is confirmed. Some patients present as adolescent females who do not develop breast tissue (as opposed to patients with androgen insensitivity—see below). Electromyography reveals delayed nerve conduction. Anterior mediastinal teratomas are generally benign and may present with signs and symptoms of airway obstruction. Incidence is 1–2/1000 children. Management is surgical repair, most often indicated when cosmetic concerns are significant. Management includes resection of the aganglionic segment. (See also Chapter 16, section I.) 2. 98. Differential diagnosis. An oral gastric tube is inserted until it meets resistance. The prognosis for a child with any stage of Lyme disease, if treated, is excellent. You refer her to a pediatric oncologist who performs a bone marrow aspirate, which confirms your suspicion that she has acute lymphocytic leukemia (ALL). Expert Panel Report 3: Guidelines for the Diagnosis and Management of Asthma. Blunt trauma to the eye or orbital rim fractures the orbital floor, which is normally thin and easy to fracture. Nursemaid's elbow is a subluxation of the annular ligament over the radial head. Normal variant short stature refers to children with short stature and normal growth velocity. This patient should undergo renal biopsy to confirm the diagnosis and to establish an appropriate approach to management. Short bowel syndrome 1. As serum iron decreases, iron-binding capacity increases, manifested as increased transferrin and decreased transferrin saturation. The answer is A [IX.A.1]. Pathophysiology involves abnormal calcium transport. The toxin prevents the release of acetylcholine at peripheral cholinergic synapses, initially causing constipation, which is followed by a weak suck and swallow, cranial nerve palsies, and weakness. On examination, the abdomen is mildly tender to palpation in the periumbilical region without rebound tenderness. Instruction on management of local reactions and removal of a stinger may be helpful but are not as important as prescribing epinephrine. Which of the following is a potential complication of this patient's likely disorder? It is typically performed between 6 and 12 months of age. Defects of Carbohydrate Metabolism VIII. However, it uses WAYYYY too many abbreviations. Cyclophosphamide, intravenously pulsed, is useful for children with severe clinical manifestations, including renal and CNS disease. Antibiotics against Staphylococcus aureus are indicated. Individually, each disease is rare but causes significant morbidity and mortality. Coller, Robert M. Fetal hydrops. The answer is C [III.E.3–6]. Females are more likely to be affected. Benign, functional cardiac murmurs that are "musical" in nature are typically systolic and are heard over the precordium. Acids (e.g., toilet bowl cleaner) cause coagulation necrosis that leads to superficial damage to the mouth, esophagus, and stomach. Anemia A. Live vaccines are more likely to induce long-lasting immunity, but carry the risk of vaccine-associated disease in the recipient, or in a secondary host. 189 IX. A home apnea monitor should be ordered to prevent sudden infant death syndrome (SIDS). For patients with clear acute cerebellar ataxia, neuroimaging is generally not necessary. An Erlenmeyer flask–shaped distal femur is a feature of Gaucher disease and not Tay–Sachs disease. 27 FIGURE 1.4 Infant with positional plagiocephaly. He is drinking less than normal, and his urine output is somewhat decreased. GH deficiency is uncommon. 709 710 FIGURE 20.1 Selected physical examination findings and their associated toxins. Patient may also have an odor of sweaty feet. Discitis typically begins with signs and symptoms of an upper respiratory illness or minor trauma, followed by back pain with tenderness over the involved disk. Intussusception may occur at a variety of sites within the intestine, but ileocolic intussusception is the most common location. The patient has at least two unprovoked seizures >24 hours apart b. Management includes copper chelating agents such as penicillamine and avoidance of copper-containing foods. 483 Review Test 1. High-spiking fevers, rash, swollen lips, cervical adenopathy, edema of the distal extremities, and elevations of the ESR and C-reactive protein are all characteristic of the acute phase of Kawasaki disease. Congenital Glaucoma VII. The parents of a 15-year-old boy consult you because they are concerned that their son has been acting strangely for the past 6 months. Which of the following would be the most appropriate recommendation at this time? Pain with passive extension (not flexion) of the digits of the hand is a sensitive indicator of compartment syndrome. Treatment includes lifelong transfusions, chelation therapy to avoid iron overload and often splenectomy. The answer is E [III.C.2]. Respiratory Distress A. Contraindications and precautions to immunization 1. Which of the following statements regarding this patient's likely diagnosis is most accurate? Hemochromatosis (iron accumulation within the heart, liver, lungs, pancreas, and skin) is a major complication and is caused by increased iron absorption from the intestine and from iron in transfused RBCs. Chelation of iron with the intravenous agent deferoxamine and/or the oral agent deferasirox promotes iron excretion and may help prevent or delay hemochromatosis. Hyphema 1. Pathologic short stature describes a child with short stature and a suboptimal growth velocity. 370 b. The classic rash of systemic-onset JIA is described as salmon-colored and maculopapular, located on the trunk and proximal extremities. Adenovirus infection C. Important clinical information about a patient's headache helps to determine the cause. Chest Pain Review Test 5 Answers and Explanations Chapter 9 Pulmonology I. Emesis from physiologic reflux generally resolves by 6–12 months of age. Etiology 496 1. Rationale for vaccine: Poliovirus is an enterovirus with propensity for the central nervous system, which may cause transient or permanent paresis of the extremities and meningoencephalitis. Female patients who are missing a part or all of one of the X chromosomes may present with short stature or lack of puberty. By late adolescence, both sexes are equally affected. The pathophysiology of migraines is complicated and evolving. 432 X. 449 9. Any complaint of back pain that interferes with play should be further evaluated. The most common cause is dehydration, and this should be considered in every patient with a high Hgb or Hct. Infantile botulism a. Laboratory results are the same as those seen in DIC [see section IV.C.3], including prolonged PT and aPTT, increased fibrin degradation products, and thrombocytopenia. Hypothyroidism. RBCs that appear as normal biconcave disks usually originate in the lower urinary tract, unlike dysmorphic RBCs, which are more likely to originate in the glomerulus. Treatment includes surgical resection, if possible, and chemotherapy. Referral to a pediatric urologist for ureteral reimplantation is appropriate. The answer is C [VII.D]. Thrombosis may also occur but is rare during childhood. In addition, symptoms cannot be secondary to other illnesses or medications. The diaphragm develops between the fifth and eighth weeks of gestation. Prompt treatment requires intravenous fluids with 5% dextrose in normal saline to correct hypotension and hyponatremia, and to prevent hypoglycemia. Other causes include infection (e.g., congenital rubella syndrome), ocular abnormalities (e.g., aniridia [absence of the iris]), or genetic syndromes, (e.g., Sturge–Weber syndrome, neurofibromatosis, Marfan syndrome). Dysrhythmias VII. Spinal cord compression D. Examination reveals tachycardia with a normal blood pressure, dry mucous membranes, and good peripheral perfusion with normal skin turgor. Vision problems can mimic the appearance of an unsteady gait. Inflammation is transmural. The answer is D [Figure 11-1]. Other common findings of pericarditis include a pericardial rub and muffled heart sounds. Classification of hypotonia 1. Some of the organization is weird, too. Symptoms may range from transient jaundice and acholic stools to liver failure, cirrhosis, and portal hypertension. Timing of vaccination: PCV-13 is recommended for all children at 2, 4, and 6 months of age, with a booster at 12–15 months of age. The answer is E [, section XVII.A.2]. Headaches in Childhood A. Causes of carbohydrate malabsorption include isolated congenital enzyme deficiency (e.g., lactase deficiency) or mucosal atrophy, which can cause a loss of enzymes or a disruption of transport mechanisms. LGA infants 1. Clinical features: Male infants become symptomatic in first 48 hours of life with poor feeding, hypotonia, and hyperventilation, which can rapidly progress to lethargy, coma, and seizures. Antistreptolysin-O titers are abnormally elevated in 70–80% of patients with rheumatic fever and are evidence of a recent GABHS infection. The answer is B [III.B.2]. Infants and toddlers may inappropriately retain stool as a result of traumatic events (e.g., hard stool, painful diarrhea, diaper rash, physical abuse) leading to a self-fulfilling cycle of increased fecal mass, fecal hardness, and painful defecation. Infants who have undergone intestinal surgery for necrotizing enterocolitis may develop short bowel syndrome. Abdominal radiographs reveal air–fluid levels, and contrast studies reveal the atresia. Total parenteral nutrition may be necessary during flare-ups to ensure adequate growth and healing. The US Centers for Disease Control and Prevention currently recommends a two-step procedure: 1. It involves the C7 and C8 nerve roots. Brain Tumors V. History and clinical presentation are the basis of diagnosis. Treatment includes NSAIDs, rest, and observation. Measurement of direct and total bilirubin is therefore necessary in all patients with suspected jaundice. Etiology (Table 11-2) 1. Adolescent females might also present with alopecia or Raynaud phenomenon when their disease is active. Affected females are sometimes initially misdiagnosed with Turner syndrome (which is associated with left-sided heart defects vs. There are two gene mutations that cause ADPKD; ADPKD1 mutations are more common and cause an earlier onset of disease than ADPKD2 mutations. Metaphyseal fractures ("bucket handle" or corner fractures), which are caused by torsional force on the limb (i.e., pulling and twisting) or by violent 701 shaking 2. Maple syrup urine disease is in the differential diagnosis. Topical antibiotics are effective and include bacitracin, sulfacetamide, polymyxin B and trimethoprim sulfate, gentamicin, tobramycin, ofloxacin, and erythromycin. He would be a candidate for palivizumab prophylaxis during respiratory syncytial virus season. Elevated IgM anti-HAV is present early and can persist for as long as 6 months after infection. Developmental delay and mild to severe intellectual disability. I am also immensely grateful to my colleagues and friends at the Palo Alto Medical Foundation who, for the past 14 years, have supported me and helped me to grow as a clinician, leader, and educator. IV ceftriaxone C. Symptoms generally present at around 2–3 months of age with hypotonia, loss of developmental milestones, seizures, and failure to thrive. These parents should now co-sleep with their infant to monitor for any future events. Hyperammonemia results from inhibition of one of the urea cycle enzymes, and measurement of urine organic acids will diagnose the disorders. growth period. Treatment may include phototherapy for mild to moderate jaundice and exchange transfusion for severe jaundice. Criteria for the classification of seizures include the presence or absence of fever, the extent of brain involvement, whether consciousness is impaired, and the nature of the movements (Figure 12-2). In patients with these problems, parenteral rehydration is more appropriate. A 4-day-old male infant has gross hematuria. A minority of patients die during the acute phase from the complications of colitis, such as toxic megacolon, or from central nervous system complications, such as cerebral infarctions. Testing should only be performed on patients with appropriate risk factors and symptoms. Nausea and vomiting c. TE—tracheoesophageal (TE) fistula 5. Ischemia may lead to avascular necrosis and cartilaginous damage. DTP, which contained whole-cell, killed Bordetella pertussis, had a high rate of side effects and was replaced with DTaP (1996), a vaccine that contains purified components (acellular) of B. Aseptic meningitis is a welldescribed complication of Kawasaki disease, but there is no known increased incidence of aneurysm formation in patients with aseptic meningitis. The duration of the headache is >1 hour. In subsequent pregnancies, antibodies pass from the mother to the fetus causing hemolysis that presents as severe jaundice (which can lead to kernicterus), anemia, hepatosplenomegaly, and hydrops fetalis. Lack of plagiocephaly can help distinguish acquired from congenital torticollis. Babies born to mothers with SLE are at risk for congenital third-degree atrioventricular (AV) block due likely to immune-mediated damage to the conducting system through maternally passed autoantibodies. Diagnosis is by history and physical examination and by exclusion of other causes of ataxia. Essential trace minerals, such as iron, iodine, fluorine, zinc, chromium, selenium, and copper, play important roles in metabolism and as enzyme cofactors. Diffuse muscular pain is present without neurologic deficits. Laboratory findings. Tics are brief, stereotypical behaviors that are initiated by an unconscious (premonitory) urge that can be temporarily suppressed. Asymptomatic to mild DKA. Shingles (reactivation of varicella zoster virus) E. Viral infections including enteroviruses (coxsackie) and rubella have been implicated but not definitely proven. Transient synovitis is the most common cause of a painful limp in toddlers. Genu varum presents with bowed legs rather than intoeing. DIC a. extension of the arms and sudden flexion of the head (jackknife or salaam seizures). Patients are prone to frequent exacerbations despite treatment. Nephrotic syndrome in children is defined as heavy proteinuria (>50 mg/kg/24 hours), hypoalbuminemia, hypercholesterolemia, and edema. Infection may occur, manifesting as warmth, erythema, tenderness, and increased induration. burgdorferi, with the caveat that symptoms caused by systemic inflammation may not resolve immediately. Pregnant patients should not receive live vaccines, including OPV, MMR, and varicella. Hirschsprung disease, or congenital aganglionic bowel disease, is caused by a lack of caudal migration of the ganglion cells from the neural crest. Surgery a. Numbness of the cheek and upper teeth below the orbital fracture may occur as a result of infraorbital nerve injury. Febrile seizures are a common but benign type of seizure associated with fever [see section V.K]. Abetalipoproteinemia, an inherited disorder in which beta-lipoproteins are absent, leading to severe fat and fat-soluble vitamin malabsorption. 611 II. Moderate to severe obesity may be associated with sexual precocity. He now takes only 1 oz of formula every 3 hours, instead of the usual 2–3 oz. For SMA type 1, survival beyond the first year of life is unusual. This distension produces visceral pain referred to the T-10 dermatome or periumbilical region and the release of inflammatory mediators. This can be assessed on examination by logrolling the leg internally. Sixty percent of all poisonings occur in children younger than 6 years. Central nervous system (CNS) lymphoma D. Key point: It is critical to evaluate growth velocity and not just absolute height when evaluating short stature. Improper spine development, including failure of vertebral formation or abnormal fusion, leads to congenital scoliosis. Parenteral antibiotic treatment with gentamicin is indicated for the treatment of suspected Escherichia coli hemorrhagic colitis. Often presents in the first two months of life. Congenital myasthenia is familial and is not transferred by the mother. There are many searches for brs pediatrics ebook online and you can also check its trends in google trends which shows that this book is searched by many students. This condition is more common in girls than in boys. A 6-month-old male infant is evaluated for lethargy and poor feeding. Abdominal radiography shows air in the stomach and proximal duodenum, 373 creating a "double bubble" sign in infants with atresia. Pancreatitis is uncommon in children. Because of the risk of vaccine-related polio, OPV is no longer used in the United States. You also note the presence of rocker bottom feet and delicate, small facial features. However, if a polyingestion is suspected, activated charcoal should be given. This coupled cotransport process of intestinal absorption continues to function normally during secretory diarrhea, whereas other pathways of intestinal absorption of sodium are impaired. Indications. The diagnosis is suggested by the history and physical examination findings and confirmed by the identification of the toxin or bacteria within the stool. No laboratory tests are routinely indicated in the initial evaluation of FTT. For preschool children, first-line treatment is parent- and school-based behavior management. Medical a. A 3-month-old female infant required stimulation by her mother when she became blue and limp after feeding. Impaired vision. Diagnosis may be suspected if prenatal ultrasound demonstrates gastric dilation with polyhydramnios. When the oculocephalic response is negative or cannot be performed because of possible cervical cord injury, caloric testing should be performed. 207 2. The typical patient is an obese adolescent male. You suspect nephrotic syndrome. Macrocytic (megaloblastic) anemias. This patient would not be expected to have a low white blood cell count or prolonged international normalized ratio (INR) based on the described symptoms and signs. Cyanotic Congenital Heart Disease V. Afebrile seizures are either generalized (starting throughout the entire brain) or partial (starting from a single part or "focus" in the brain). Etiology is often unknown. His parents noticed the bleeding today when they changed his diaper. Administering the test takes only a few minutes and is painless, and the expenses for equipment and training are lower than for brainstem auditory-evoked response/auditory brainstem response (BAER/ABR) as below. The mass within the sternocleidomastoid muscle represents bleeding or fibrosis within the muscle body and does not require biopsy by otolaryngology. A 4-year-old boy with transient mild wheezing two to three times per year associated with cold symptoms. Other causes of vitamin B12 deficiency include pernicious anemia, inflammatory bowel disease, and short gut sydrome. coli sepsis. In addition, teens who are depressed usually diminish their extracurricular activities, not increase them. 427 E. Examples include the following: a. The most common causes include glomerular diseases (e.g., FSGS), congenital or inherited kidney diseases (e.g., renal dysplasias or obstructive uropathies), reflux nephropathy, collagen vascular diseases, cystic kidney diseases, interstitial nephritis, and HUS. C—cardiac defects 4. Tdap (has less diphtheria component compared with DTaP) is a booster immunization, offering continued protection from these diseases in the adolescent and adult age group. Causes include severe hypertension, hemolytic uremic syndrome (HUS), artificial heart valves, a giant hemangioma, and disseminated intravascular coagulation (DIC). Vitamins, including iron e. Staining of the cornea with blood 4. Synthetic function is assessed by evaluating protein production (e.g., prealbumin, albumin, and prothrombin time), serum chemistries (e.g., glucose, cholesterol), and toxin clearance (e.g., lactate, ammonia). The clinical features of the three most common disorders occurring in childhood, congenital hypoplastic anemia (Diamond–Blackfan anemia), transient erythroblastopenia of childhood, and parvovirus B19–associated RBC aplasia, are presented in Table 13-5. Unusual facial characteristics include a broad forehead, deep-set and widespaced eyes, a saddle nose with a bulbous tip, pointed chin, and large ears. A—choanal atresia or stenosis 4. Less commonly, acute pancreatitis may be the result of a systemic illness, such as systemic lupus erythematosus or cystic fibrosis. (The Barlow maneuver attempts to dislocate a dislocatable hip.) With the hips at 90° flexion, the thumb is placed on the medial side of the thigh and the middle finger on the greater trochanter, and gentle pressure is applied posteriorly and laterally to dislocate the hip. Most cases involve the left diaphragm in the posterior and lateral area. Bilious emesis and abdominal distension occur within the first few days of life. SS disease occurs in 1 in 800 black newborns in the United States. Another clue to carbohydrate malabsorption is a stool pH below 5.6. 2. Hydrocephalus requires the surgical placement of a ventriculoperitoneal shunt to divert the flow of CSF. Patients with a history of intussusception should not receive rotavirus vaccine. Two-thirds of rhabdomyosarcomas occur in children younger than 10 years. Psychosocial causes (e.g., neglect, child abuse) c. The answer is E [XI.A.2]. OAE accuracy may be affected by debris or fluid within the external or middle ear. In girls, most cases are idiopathic. An initial insult, such as ductal obstruction, abdominal trauma, or viral infection, causes premature activation of pancreatic proenzymes and autodigestion of pancreatic cells. Tension headaches 1. Hyperventilation suggests metabolic acidosis (Kussmaul respirations or rapid, deep breathing may occur), neurogenic pulmonary edema, or midbrain injury. Small bowel disease often leads to malabsorption with resultant iron, zinc, folate, and vitamin B12 deficiencies. Medications may interfere with vitamin K metabolism (e.g., cephalosporins, rifampin, isoniazid, warfarin). Household bleach has less corrosive potential and generally does not require treatment. β-blockers +/− angiotensin receptor blockers are prescribed to help prevent or slow progressive aortic enlargement. Intestinal perforation may result from GI vasculitis. Two additional causes of unconjugated hyperbilirubinemia are worth noting now: 1. About 50% of tumors occur in the distal femur or proximal tibia. Other pathogens include other strains of E. Alport syndrome is characterized by renal manifestations, including hypertension and hematuria, as well as renal failure more consistently in the males; hearing loss; and ocular abnormalities of the lens and retina. Malignant tumors may extend locally or may metastasize to retroperitoneal lymph nodes, lung, or liver. This form of anemia results from mechanical damage to RBCs caused by passage through an injured vascular endothelium. Clinical features include poor growth with delayed bone age, central obesity, moon facies, nuchal fat pad, easy bruisability, purplish (hemorrhagic) striae, hypertension, and glucose intolerance. Diagnosis and treatment is the same as for spondylolysis. Cardiovascular abnormalities include dysrhythmias and myocardial ischemia. Unless the child is toxicappearing, cultures should be obtained before administering antibiotics. Prader–Willi syndrome A 12-year-old boy is brought to the office for a routine health maintenance visit by his parents. Absence seizures are brief staring spells that occur without loss of posture and with only minor motor manifestations (e.g., eye blinking or mouthing movements). Mucopolysaccharidoses (MPSs) are caused by defects in enzymes required to break down glycosaminoglycans, resulting in accumulation of molecules in various organs. The outlook is excellent, and complete recovery is expected. Two hours ago a 2-year-old boy had a 5-minute episode of whole body shaking associated with a temperature of 39.4°C (103°F). Start an oral leukotriene modifier. Lung disease eventually develops in all individuals with classic CF; the lungs are initially colonized with Staphylococcus aureus and subsequently with mucoid Pseudomonas aeruginosa. Subtypes. Disorders of Hemostasis V. Elevated ESR and CRP b. Lowe, MD, Attending Physician, Division of Emergency and Transport Medicine, Children's Hospital Los Angeles, Los Angeles, California, Assistant Professor of Pediatrics, Keck School of Medicine at the University of Southern California, Los Angeles, California Elizabeth Mumper, MD, FAAP, President and CEO, Advocates for Children, Lynchburg, Virginia, Associate Professor of Clinical Pediatrics, University of Virginia School of Medicine, Charlottesville, Virginia Ronald A. I hope to live up to all of the support that they, and others, have given to me. Corneal abrasion is damage to, and loss of, corneal epithelium. The differential diagnosis of disorders of hemostasis is summarized in Figure 13-3. Intramuscular administration of vitamin K after birth prevents hemorrhagic disease of the newborn. 385 c. Acuity assessment a. Headaches may have intracranial or extracranial causes (Figure 12-3). An Ortolani-positive hip is one in which the hip is dislocated but can be reduced. Persons with S trait have Hgb A (50–60%), Hgb S (35–45%), and a small percentage of Hgb F. CNS bleeding may occur occasionally. Sexual abuse 1. Upper Extremity A. Accidental burns have an irregular, splashlike configuration. Developmental delay, with variable intellectual disability d. The most common cause of glucocorticoid excess is iatrogenic, as seen in patients who have been treated with long-term steroids for chronic diseases, such as asthma, inflammatory bowel disease, and juvenile idiopathic arthritis. Patients cannot convert testosterone to DHT, the more potent version of testosterone that binds to androgen receptors and virilizes the external genitalia. Table 5-5 Characteristics of Selected Defects in Amino Acid Metabolism Phenylketonuria (PKU) Inheritance Clinical features Diagnosis Management Prognosis Maple Syrup Urine Disease Tyrosinemia Type I Autosomal recessive Autosomal recessive Autosomal recessive Developmental delay Infantile hypotonia Mousy or musty odor Progressive mental retardation Eczema Decreased pigment (light eyes and hair) Mild PKU may present in early childhood with developmental delay, hyperactivity Progressive vomiting and poor feeding Lethargy, hypotonia, and coma Developmental delay Maple syrup odor in urine Hypoglycemia and severe acidosis during episodes Episodes of peripheral neuropathy Chronic liver disease Odor of rotten fish or cabbage Renal tubular dysfunction ↑ Phenylalanine: Tyrosine ratio in serum ↑ Serum and urine branchedchain amino acids Succinylacetone in urine Phenylalanine-restricted diet Dietary protein restriction Dietary restriction of phenylalanine, tyrosine, NTBC Liver transplant Near-normal intelligence if diet restriction begun < 1 month of age Protein restriction within 2 weeks of life may avert neurologic damage Death by 1 year of age if disease begins in infancy Increased risk of hepatocellular carcinoma and cirrhosis 185 NTBC = 2-2 nitro-4-trifluoromethylbenzoyl 1,3-cyclohexanedione. This severe inflammation of the colon leads to decreased intestinal motility, disruption of the mucosal barrier that allows bacteria to enter, and colonic dilatation. GH treatment has been shown to improve the ultimate height of these patients. Persistent central cyanosis (trunk, lips, tongue) is an emergency and requires immediate diagnosis and treatment. Because of end-organ dysfunction (i.e., gonadal failure), these patients have high FSH and high LH levels with low testosterone or low estradiol levels. In both Duchenne and Becker muscular dystrophy, the onset of weakness is slow and progressive. Additional tests may be warranted, including preductal (measured at the right wrist or hand) and postductal (measured in either lower extremity) oxygen saturation measurements, electrocardiogram, echocardiogram, and blood culture. Diagnosis is based on suggestive history, neurologic examination, and identification of the toxin or bacteria in the stool. Children with a cerebellar gait have an unsteady, wide-based stance with irregular steps, also known as a "drunken gait." See Table 12-4 for the causes of cerebellar ataxia. (The Ortolani maneuver attempts to reduce a dislocated hip.) In the same position described above, the hip is abducted with pressure gently applied upward with the middle finger to slide the head of the femur back into the acetabulum. Patients with 21-OH deficiency have increased 17-OHP levels. Patients with a shortened small intestine may have malabsorption resulting in malnutrition and compromised bowel function. Mycoplasma pneumoniae 4. Birth = 1.7 2. CBC often shows anemia or leukocytosis. F. Signs and symptoms are similar in liver tumors and include presentation with a right upper abdominal mass, loss of appetite, and weight loss. Diarrhea IX. Human bites may be found anywhere on the body, including the genitalia and buttocks of infants. Hyperventilation for 2–3 minutes frequently provokes absence seizures. The major risk factor is pharyngitis caused by certain strains of GABHS. Trisomy 13 is associated with midline defects of the brain and forebrain. Lack of fever or pharyngitis 4. Minerals, including sodium, chloride, potassium, calcium, phosphorus, and magnesium, are also required daily. Polymorphisms in human leukocyte antigens (HLAs) (specifically DR3 and DR4 haplotypes) on chromosome 6 are responsible for most of the genetic risk in large genome-wide association studies. After a single unprovoked seizure, approximately 40% of children will have a second seizure. Tall stature with long extremities, long fingers (arachnodactyly), pectus deformity, scoliosis, pes planus, decreased upper-to-lower segment ratio, increased arm span-to-height ratio 3. Both types of thalassemia result in hemolysis that leads to increased bone marrow activity. The toxin binds to vascular endothelial cells, especially in the renal vasculature, causing platelet thrombi and resultant renal ischemia. NAC is hepatoprotective if given within 8 hours of ingestion. Infectious Diseases. Bilirubin levels are more variable with a lower likelihood of kernicterus. Rarely, patients may present with stroke or other thrombotic events such as renal vein thrombosis, deep vein thrombosis, and sagittal sinus thrombosis. This child's presentation and computed tomography (CT) findings (a parenchymal cyst that may be enhancing) are consistent with neurocystercercosis. Specific peripheral hypotonic disorders are described in more detail in the following discussion. The answer is B [, section XIII.A.3.b]. If serum iron levels > 500 μg/dL, or if > 300 μg/dL and acidosis, hyperglycemia, or leukocytosis are present b. Admission to the hospital and immediate treatment with intravenous immune globulin are necessary. Other signs and symptoms include hepatosplenomegaly, ascites, poor growth, steatorrhea, peripheral edema, and coagulopathy. Girls: No breast tissue by 13 years of age, or no menarche by 16 years of age. Assessment. Pathophysiology and etiology 1. The answer is B [, section IV.A.1, Figure 17-3 and Table 17-5]. The differential diagnosis of red eye in infants and children is extensive (Figure 18-1). However, prognosis is poor for the following victims: 1. The prognosis for patients with Shiga toxin–associated HUS is generally good, although poor prognostic signs include elevated white blood cell count on admission and prolonged oliguria. Preliminary studies show that WBI may be effective for ingestions of iron and other heavy metals and sustained-release medications. The answer is B [V.A.4.b, Table 17-6, and Figure 17-5]. If this patient develops a high fever, she should be empirically treated with antibiotics to cover possible pneumococcal peritonitis. Hypothyroidism may rarely present with CPP, but in this case, there is poor growth and a delayed bone age (unlike all other causes of sexual precocity in which growth is accelerated). The Hgb is high at birth in most newborns and then declines, reaching the physiologic lowest point (nadir) between 2 and 3 months of age in the term infant and between 1 and 2 months of age in the preterm infant. Serum IgA-endomysial testing and serum tissue transglutaminase antibody testing are extremely sensitive and specific screens for celiac disease, except in individuals with IgA deficiency. The distinguishing clinical features of the common causes of pediatric red eye are presented in Table 18-3. This inflammatory condition of children results in myopathy, progressive muscle weakness, and characteristic skin findings. Celiac disease (gluten-sensitive enteropathy) 1. Favorable histology and stage I, II, or III disease result in a 2-year survival rate >95%. If patients are also aldosterone deficient, mineralocorticoid replacement (fluorocortisol) in combination with salt supplements (in young children) may be given at a dosage that normalizes the plasma renin activity (PRA). 32 b. Burns V. Urolithiasis XIV. Chlamydia trachomatis should be considered as a possible etiologic agent. Inflammatory mediators and cytokines damage the pancreas and cause interstitial edema and necrosis. Neuroimaging 1. Hydrocephalus III. Examples of live vaccines include intranasal influenza; varicella; and measles, mumps, and rubella (MMR) vaccines. To evaluate for constitutional delay of growth and puberty, or CDGP [see section I.D.1], ask whether the family history is positive for males with "late growth spurts" in high school or college and the age of maternal menarche. There is no recommendation to vaccinate with a measles, mumps, and rubella (MMR) vaccine before college entry, unless an individual has not yet received two vaccines. Poor prognosis is associated with metastases (20% have metastatic disease at time of diagnosis) and with recurrence. Other effective treatment modalities in cystic fibrosis are mucus-altering agents with aggressive pulmonary toilet, including chest physical therapy to help expectorate thick mucus. Physical examination of the neonate is notable for facial diplegia (bilateral weakness), which results in characteristic "V" shape of the upper lip, 455 hypotonia, areflexia, and arthrogryposis (multiple joint contractures). On physical examination, a periorbital violaceous heliotrope rash is evident. Classification is by location and degree of cell differentiation. Autoantibodies may be detected in asymptomatic patients years before the onset of clinical symptoms. Location and radiation. Which of the following statements is most consistent with the most likely diagnosis? Unfavorable histology accounts for 12% of cases but 90% of deaths. Nonlive or inactivated (IPV), administered subcutaneously or intramuscularly, does not cause vaccine-related polio, but also does not confer secondary immunity. Onset is between 9 and 14 years of age. It will likely disappear by 2–6 months of age. Appropriate fluid management normalizes the Hct. Malnutrition and corticosteroids may weaken the collagen supporting vessels. SB occulta does not require treatment. Dialysis may be required for life-threatening ingestions. Ortolani and Barlow maneuvers are not useful after about 4 months of age because the hip stabilizes in either the dislocated or reduced position. Henoch–Schönlein purpura, an IgA-mediated vasculitis, presents with palpable purpura on the lower extremities and buttocks, renal insufficiency, arthritis, and abdominal pain. The answer is C [II.C–F]. Jaundice 2. Craniosynostosis is usually noted by 6 months of age. Developmental delays Table 5-1 Clinical Features and Complications Associated with Down Syndrome, with Recommendations for Screening Clinical Features Complications 177 Craniofacial features Atlantoaxial instability (1–2%) Anemia Brachycephaly Epicanthal folds Check hemoglobin annually Leukemia (1% of patients but more common than the general population) Upslanting palpebral fissures Brushfield spots (speckled irides) Celiac disease (5%) Protruding tongue Hypotonia Intellectual disability Early onset Alzheimer disease Obstructive sleep apnea (50–75%) Sleep study by 4 years of age Musculoskeletal features Hearing problems (75%) Clinodactyly Hearing screens needed every 1–2 years Thyroid disease Single palmar creases Wide space between first and second toes (sandal gap) Annual TSH screening Gastrointestinal features Cataracts, glaucoma, and refractive errors Duodenal atresia Annual ophthalmologic examinations Hirschsprung disease and omphalocele Pyloric stenosis Cardiac defects (40%) Endocardial cushion defects (most common)— echocardiogram at birth TSH = thyroid-stimulating hormone. An autosomal recessive disorder caused by cystathionine β-synthase deficiency, resulting in increased levels of homocysteine and methionine 1. False-negative guaiac results may occur as a result of large ingested doses of vitamin C. You note scattered petechiae on his wrists and ankles, and he appears unwell. Along with hepatitis B vaccine, HPV vaccine is one of the few cancer-preventive immunizations. All rights reserved. This work is no substitute for individual patient assessment based upon health care professionals' examination of each patient and consideration of, among other things, age, weight, gender, current or prior medical conditions, medication history, laboratory data, and other factors unique to the patient. K. Apneustic breathing (pausing at full inspiration) indicates pons or upper medulla damage. Periumbilical pain is the classic presentation of FAP. Hemolytic Uremic Syndrome (HUS) See Chapter 13, section I.F.2.b.(3) and Chapter 7, Table 7-6. The majority of poisonings occur at home when the child's caregiver is distracted. Early detection and early intervention are critical to the treatment of amblyopia. Liver biopsy is invasive and is reserved for confirmation of diagnosis, for assessment for hepatic injury, or when the evaluation remains inconclusive. gonorrhoeae. The answer is D [XII.E]. The differential diagnosis of conjugated hyperbilirubinemia is presented in Chapter 4, Figure 4-4. Manifestations include cyanosis, respiratory distress (which may be characterized by tachypnea, retractions, expiratory grunting, nasal flaring, and stridor), and decreased air entry or gas exchange. There are two types of vaccines, with recommendations for administration updated annually. Which of the following statements regarding this patient's presentation, evaluation, and management is correct? You will be required to select one answer for each statement in the set. The most commonly associated infectious agent is Campylobacter jejuni, which causes a prodromal gastroenteritis. It most commonly occurs during adolescence. Major disadvantage is that it does not confer as broad coverage against as many pneumococcal strains as PPSV-23. Treatment involves pinning the epiphysis to prevent further slippage. The radial head in children younger than 6 years has a cylindrical shape, allowing it to slip out of the annular ligament, which normally keeps it in place. 11β-OH deficiency (accounts for 5% of cases). This disorder is better termed systemic sclerosis because it is characterized by excessive fibrosis and subsequent dysfunction of multiple organ systems. A simple febrile seizure lasts less than 15 minutes and is generalized. c. Cholangitis is a worrisome complication of the procedure that occurs in as many as 50% of patients, and repeated episodes can stop all bile flow. Here the enzymatic defect is further down the mineralocorticoid synthetic pathway. Intravenous antibiotics should be started. The answer is C [III.A.7]. 3β-hydroxysteroid dehydrogenase deficiency (rare). Specific Viral Infections XIV. Laboratory studies. A partial list of those to whom I am indebted includes Drs. This patient is likely to have an aura with her headache. Complete resolution of symptoms generally occurs in 2–3 weeks. Any pathologic condition that disrupts alignment or retinal stimulation during this period may result in poor vision as a result of amblyopia [see section II.B]. Anticonvulsant therapy may be indicated, but discontinuation of antiepileptic medication after 2 years of being seizure free can be considered. The boy's parents state that their son has had runny nose and cough for the past 24 hours. Upper intestinal endoscopy for diagnosis and management is indicated for active bleeding with hemodynamic changes. Osteogenic sarcoma is the most common malignant bone tumor in children. The findings of scissoring of the lower extremities, clenched hands with overlapping digits, rocker bottom feet, and delicate small facial features are consistent with the diagnosis of trisomy 18, the second most common trisomy syndrome after trisomy 21. Blepharitis 1. Like there is an end of chapter practice question where the answer is to give a systemic course of steroids for an asthma exacerbation, but it's not on the algorithm page for asthma. Topical mast cell–stabilizing drops, such as cromolyn c. Blood culture is positive in 30–50% of cases. Hypercholesterolemia is a consequence of the hypoproteinemia. RBCs that appear to be normal biconcave disks usually originate in the lower urinary tract. A nuclear medicine scan identifies the ectopic gastric mucosa, and surgical resection is required. On physical examination, you note that he has large ears and large testes. The normal production of CSF is between 400 and 500 mL daily. See Chapter 5, section XI.A. F. Associated cerebral aneurysms may occur, with early death. The subsequent repletion phase, or the more gradual correction of fluid and electrolyte deficits, should occur over 24 hours for patients with isonatremic and hyponatremic dehydration, and over 48 hours for patients with hypernatremic dehydration. A detailed history focusing on symptoms and risk factors for nonorganic CAP, and a comprehensive physical examination with special attention to the abdominal and genitourinary systems, are necessary to exclude organic disease. Early enteral feedings are important to ensure adaptive growth of the remaining small bowel, proper hepatic function, and proper oral motor development. Imaging is indicated in selected children with UTI, because children with UTI have 442 an increased incidence of structural abnormalities of the urinary tract (e.g., vesicoureteral reflux). Hemolytic Uremic Syndrome (HUS) VIII. Immunizations A. Table 13-3 describes the clinical features and management of the common SS disease crises. History provides an opportunity to obtain diagnostic information and to form a doctor– patient–family alliance. Hydrocephalus ex vacuo is not true hydrocephalus, but rather a term used to describe ventricular enlargement caused by brain atrophy. Ferry, MD, FACC, Managing Partner, Pediatric Cardiology Medical Associates of Southern California, Encino, California, Attending Pediatric Cardiologist, Children's Hospital Los Angeles, Los Angeles, California Amy E. Osteomyelitis may occur and may mimic a painful bone crisis. α-Thalassemia minor. 2nd Ed. Philadelphia: Lippincott Williams & Wilkins, 2013. Positive Ortolani maneuver. Clinical manifestations include bruising, oozing from skin puncture wounds (e.g., previous blood draw sites), and bleeding into organs. Ocular abnormalities involving the lens and retina occur in 25–40% of patients. Laboratory findings may include mild elevations of bilirubin and serum transaminases. Melena is dark, tarry stools and often indicates an upper GI bleed proximal to the ligament of Treitz. Hurler disease E. Neonatal Jaundice XI. The majority of cases are caused by inadequate iron intake. Microcephaly, short palpebral fissures, smooth philtrum, and thin upper lip 2. Attention should be paid to fluid and electrolyte imbalance. Magnetic resonance imaging (MRI) shows diffuse muscle edema in proximal muscle groups and can be used to avoid more painful procedures, such as electromyography (EMG) and muscle biopsy. Signs of trauma should be noted. coli hemorrhagic colitis is controversial and may actually increase the likelihood that a patient will go on to develop HUS. It is important to remember that almost all growth in females ceases within 6 months of menarche. Causes of postnatal-onset proportionate short stature a. Classification by degree of dehydration a. Chronic Abdominal Pain VII. Clinical features suggesting abnormal hemostasis include the following: a. On the basis of your findings, you suspect Kawasaki disease. Electromyography (EMG). UGI study is very effective at diagnosing atresia and stenosis. Infants may cry and draw their legs toward the chest. If the patient has altered mental status, administer dextrose for hypoglycemia and naloxone for possible opiate overdose. GSD type I (von Gierke disease) is most commonly caused by glucose-6-phosphatase deficiency. Wilson, Jason T. Patients present with a history of poor growth and increasing weight gain, purpuric stretch marks and a dorsal neck fat pad on examination, with delayed bone age. Conservative management for a presumed diagnosis of Henoch–Schönlein purpura E. She is treated with cephalexin and returns 10 days later for an imaging evaluation to rule out a structural abnormality of the urinary tract. The diagnosis may be missed if the mother of the patient is not seen or examined because the typical myotonia (i.e., inability to relax contracted muscles) is most apparent in adults. A group of malformations generally observed as a sporadic occurrence in an otherwise healthy family. This boy likely has Rocky Mountain spotted fever (RMSF). The multifactorial etiology includes environmental, genetic, nutritional, and teratogenic factors. The rash is annular and "targetlike" with central clearing. Laboratory findings 1. Immunizations 8. Associated conditions include duodenal atresia, tracheoesophageal fistula, trisomy 18, and Cornelia de Lange syndrome. Diabetic Ketoacidosis (DKA) IX. Note that the jaw pressure of a dog may exceed 200–450 pounds per square inch. Biologics, including bioengineered anti-TNF monoclonal antibodies, are quite effective for IBD but are expensive and require parenteral administration. Voluntary guarding may progress to involuntary guarding and rebound tenderness as peritoneal irritation increases. Inciting medications, if any, must be stopped immediately. Small-for-gestational-age (SGA) infants and FGR 1. The prevalence of Tourette syndrome is 1 in 1000 live births. A complete blood count is normal, but an electrolyte panel shows metabolic acidosis. Cholecystitis a. See I.E.2.a and I.E.2.b. Key point: All patients with GH deficiency must have neuroimaging, preferably with an MRI with thin cuts through the pituitary. Migraines are characterized by unilateral, or sometimes bilateral, frontal throbbing headaches that last for at least 1 hour. Ipsilateral conjunctival injection, eyelid edema, lacrimation, nasal congestion, rhinorrhea, and forehead sweating. It is characterized by enthesitis. Education is the most important intervention. A ratio of testosterone:DHT of >20:1 with β-hCG stimulation strongly supports the diagnosis. Vitamin and iron supplements may be required. Symmetric FGR affects all growth parameters equally (head circumference, length, and weight), whereas asymmetric FGR affects birth weight but preserves length and head circumference. Asymmetry of the shoulder height, scapular position, and the waistline may be present. Infants and young children tend to present with growth failure and vomiting, and, at times with life-threatening metabolic acidosis. Air remains trapped in the meconium; thus, there is no definite air–fluid interface. Unabsorbed sugars are fermented by colonic bacteria, which produce hydrogen gas, carbon dioxide, and acids. Progression of disease is rapid, with bile duct obliteration and cirrhosis 393 occurring by 4 months of age. Gastric emptying delay may also play a role in GERD by increasing the quantity of gastric refluxate and by increasing gastric distension, which induces more TLESR. Mental retardation, developmental delay, severe handicaps, hyperactivity, or challenging temperament (including colic or frequent tantrums) 3. A deformation sequence that results from compressive forces exerted on a developing fetus subjected to prolonged oligohydramnios. The patient should be assessed for scalp injuries, breath odors (for alcohol intoxication or ketosis caused by diabetic ketoacidosis), and nuchal rigidity (caused by meningitis). Psychosocial and family support H. The answer is C [IX.B, IX.E, and IX.I.1]. Orbital tumors typically present with proptosis, chemosis (i.e., conjunctival edema), eyelid swelling, and cranial nerve palsies. Type of vaccine: live attenuated vaccine c. Increased copper deposition on liver biopsy 3. Diagnosis is based on the history and physical examination, but typically also includes a plain radiograph of the shoulder to evaluate for an associated clavicular fracture. Management 1. The answer is E [V.A]. Sideroblastic anemia is a group of anemias characterized by the presence of ring sideroblasts in the bone marrow. Complications include TPN cholestasis with resultant gallstones and cholestatic liver disease, intestinal bacterial overgrowth, nutritional deficiencies, poor bone mineralization and renal stones from hyperoxaluria, and secretory diarrhea as a result 367 of the osmotic load in the remaining intestine. Omphalocele occurs in approximately 1:6000–8000 live births. A more appropriate categorization of FTT describes whether there is inadequate caloric intake, inadequate caloric absorption, inadequate utilization of nutrients, or excessive metabolic demand. Transmission from Ixodes pacificus, which is the tick most prevalent in the Western United States, is less common. Complications include increased incidence of fetal and neonatal morbidity and death due to placental insufficiency and related severe intrauterine asphyxia, meconium aspiration syndrome (MAS), and polycythemia. Supportive therapy a. Chronic diseases such as hypothyroidism, renal failure, central nervous system (CNS) disease, severe asthma with frequent and prolonged steroid use, sickle cell anemia, celiac disease, and inflammatory bowel disease may manifest as short stature. Perpetrators are often known to the child before the abuse. Disorders of Metal Metabolism Review Test Answers and Explanations Chapter 6 Endocrinology I. Risk factors relating to family history include alcoholism, antisocial or conduct disorders, attention deficit hyperactivity disorder, and family members with functional pain syndromes (e.g., headaches, CAP, musculoskeletal pain). Testing the stool with cultures for enteropathogenic E. Diabetes Mellitus A. After this age, decreased ability to abduct the affected hip, asymmetric thigh or buttock folds, or Galeazzi sign is more useful in the diagnosis of DDH. Blood culture in combination with a culture of the aspirated bone may identify the organism in 70% of cases. Upper and lower airway disease may be induced or worsened by GERD. This usually resolves by 2–6 months of age. Laboratory studies show metabolic acidosis, hyperammonemia, and ketotic hypoglycemia. In contrast to adults, acute cholecystitis is uncommon in healthy children. 480 2. Physical examination reveals an irritable but consolable child with a temperature of 39.9°C (103.8°F). Cholestasis is characterized by retention of bile within the liver with prolonged elevation of conjugated (direct) bilirubin. Diagnosis is on the basis of characteristic clinical features and does not rely on specific laboratory tests. Pyridostigmine bromide is the drug of choice. An 8-month-old female infant with a 5-day history of constipation presents with a weak cry, hyporeflexia, ophthalmoplegia, and loss of the ability to sit without support. Inheritance is autosomal dominant with age-dependent penetrance. Because the β-HCG molecule cross-reacts with LH, it too can bind to LH receptors and enlarge the testes slightly, stimulating Leydig cells and secreting androgens. The resulting stool is watery and acidic and contains unabsorbed sugars. Interference with oxidative phosphorylation c. There is a petechial eruption on the lateral thighs, along with some edema of the hands and bilateral ankles. Liver transplantation may also be required. Air or contrast enemas, often under ultrasound guidance, successfully reduce the intussusception in 80–90% of cases. Absorption requires an adequate intestinal mucosal surface and villous brush border with intact transport mechanisms. Septic arthritis is a bacterial infection of the joint, which can be caused by hematogenous spread (most common), contiguous spread, or direct inoculation. Increased upper-to-lower segment ratio B. Mammalian Bites X. Females with Turner syndrome are at risk for developing gonadoblastomas, a neoplasm that occurs in dysgenetic gonads (question 107). Visceral injuries are the second leading cause of death from child abuse and include rupture and injury of the intestinal tract, liver, and spleen. An autosomal dominant disorder most often caused by a spontaneous mutation b. *Scleroderma 1. Clinical features of MCAD. Most patients suffering from malnutrition have a combination of energy and protein depletion. Apnea of Prematurity X. Vitamin B12 deficiency a. Laboratory evaluation is likely to demonstrate an elevated erythrocyte sedimentation rate and leukopenia. Assessment of fetal lung maturity can be made by determining the presence of surfactant in amniotic fluid obtained by amniocentesis. Bronchoscopy is indicated. Complete blood count (CBC) b. Genetic syndromes 1. Torticollis is defined as the tilting of the head to one side. Prolonged bleeding time and prolonged aPTT may be present, but not always (but they are always present in type III disease). Which of the following characteristics predict an unfavorable prognosis in this patient? No sensory loss occurs, although low-back or leg pain may be present in 50% of patients. Unlike physical abuse, there are typically no overt physical signs of trauma. However, appropriate visual input from each eye is necessary to refine and maintain these binocular neural connections. Purulent discharge, conjunctival erythema, and lid swelling are usually present. Hepatic damage, the major sequelae of toxicity, is directly related to the depletion of glutathione, a cofactor used during the metabolism of acetaminophen by the cytochrome P-450 system. Fine gas bubbles may be seen mixed within meconium, producing a characteristic soap-bubble appearance. The answers are F [IX.B.1], B [III.A.14], and A [III.A.6], respectively. Asymmetric head shape (plagiocephaly, see Chapter 1, section II.B.2.e) may develop in untreated torticollis. Age of onset is between 4 and 8 years of age and is more common in girls. Health care professionals, and not the publisher, are solely responsible for the use of this work including all medical judgments and for any resulting diagnosis and treatments. Pain is described as dull, aching, and rarely throbbing, and it increases in intensity during the day. Retinoblastoma See Chapter 18, section VIII.B. 543 X. Other patients at high risk include young infants, pregnant women, and the elderly. Topical antihistamines d. Disorders of Granulocytes (Figure 15-3) XII. In the emergency department, the physical examination is completely normal. Corticosteroids are often given to calm acute exacerbations and induce remission. Papilledema e. aHUS is much less common than STEC-HUS. Seizures typically occur in the early morning hours when patients are asleep with oral–buccal manifestations (i.e., moaning, grunting, pooling of saliva). Only frank aspiration of barium can be interpreted as GERD. Magnetic resonance imaging (MRI), bone scan, or a single photon-emission computed tomographic (SPECT) scan may be used for diagnosis if the fracture is acute. Babies often present with bradycardia (question 103). The neurologic examination of patients with migraines is usually normal. Physical examination classically demonstrates tenderness to palpation at McBurney point (an area two-thirds of the way from the umbilicus to the 376 anterior superior iliac spine). Which of the following statements regarding the most likely diagnosis is most accurate? He was adopted, and his birth history is unknown. CAP, occurs in 20–40% of children and adolescents. This patient's nephrotic syndrome is most likely a consequence of a primary glomerular disease, such as IgA nephropathy. Children: use of eye charts or cards 3. Angelman syndrome B. Blood culture is positive in 50% of cases. aPTT is prolonged and low factor IX activity is found. The answer is E [II.D]. Infants with open cranial sutures have the following clinical signs: a. The patient's normal platelet count is inconsistent with the diagnosis of immune thrombocytopenic purpura. Empiric intravenous antibiotics to cover Gram-positive organisms, including consideration for methicillin-resistant S. You note that his height is below the third percentile, yet his head circumference is at the 75th percentile. Discitis is defined as infection (Staphylococcus aureus is the most commonly identified organism) or inflammation (idiopathic, traumatic, or rheumatologic) of the intervertebral disk. The cause of incomplete canalization is unknown. This reactive inflammatory disorder of the skin, heart, CNS, and connective tissues is caused by spirochetal infection with Borrelia burgdorferi (in the United States) and transmitted via a tick bite. Cholescintigraphy may be useful. A 2-week-old female infant is being fed both breast milk and infant formula. There are usually no chronic joint sequelae from acute rheumatic fever. Brown, FAAP, Regional Chair of Pediatrics, Palo Alto Division, Palo Alto Medical Foundation, Palo Alto, California, Adjunct Clinical Associate Professor of Pediatrics, Stanford School of Medicine, Stanford, California, Ryan J. D. All of the UCDs are autosomal recessive, except ornithine transcarbamylase (OTC) deficiency, which has Xlinked recessive inheritance. We have created case-based review questions at the end of each chapter to emphasize key takehome points and to help the reader to assess his or her understanding of each chapter's contents. However, it is important to keep in mind that the presenting symptoms of IEM may be similar to those of sepsis, and patients with IEM are vulnerable to sepsis. Biologic Poisonings (Venoms) Review Test Answers and Explanations Index 9 Copyright Acquisitions Editor: Matt Hauber Development Editor: Andrea Vosburgh Editorial Coordinator: Kerry McShane Editorial Assistant: Brooks Phelps Senior Marketing Manager: Michael McMahon Production Project Manager: Bridgett Dougherty Design Coordinator: Elaine Kasmer Manufacturing Coordinator: Margie Orzech Prepress Vendor: TNQ Technologies Second edition Copyright © 2019 Wolters Kluwer Copyright © 2005 Lippincott Williams & Wilkins. Tachycardia and tachypnea may be present. Classic salt-wasting CAH (i.e., both mineralocorticoid and glucocorticoid pathways are affected, resulting in both cortisol and aldosterone deficiency). In addition to the Southwestern United States, endemic areas for coccidioidomycosis include the Mississippi River Valley. Electrolyte intake should be matched to estimated electrolyte losses. The development of total parenteral nutrition (TPN) now allows patients to maintain adequate nutrition and significantly improves survival. (C) Reprinted with permission from Weinstein SL, Flynn JM. A sequence that can be seen as an isolated finding or as part of a specific multiple malformation syndrome. Disorders of Development III. Head MRI may show increased signal intensity in the caudate and putamen on T2-weighted sequences. Time of onset. The intestines are then fixed to the abdominal wall. In general, the acute management of an inborn error of metabolism involves supplying a source of energy that can be used, removing toxic metabolites, and preventing continued exposure to the offending substance. Infants may be SGA. Specific therapies may modify disease progression, some genetic diseases may occur in siblings or offspring, and some diseases may recur in transplanted kidneys. The age of onset and severity of the different UCDs varies. Timing of vaccination: MMR vaccine is recommended at 12–15 months of age with a booster at 4–6 years of age. Well-Child Care—General Concepts II. The two most common types of microcytic, hypochromic anemia during childhood are iron-deficiency anemia and β-thalassemia minor. Constipation is often the initial symptom in infants. Patients are afebrile, have a characteristic staccato-type cough, and may have a history of conjunctivitis (in 50% of cases). Indications for referral to an ophthalmologist include severe eye involvement, conjunctivitis associated with contact lens use, suspected corneal ulcer, or lack of improvement with topical antibiotics. Niemann–Pick disease types A and B are neurodegenerative disorders caused by acid 190 sphingomyelinase deficiency. In boys, consider Klinefelter syndrome (XXY; see Chapter 5, section IV.C.2). Restlessness may indicate colicky pain as seen in an obstructed viscus. Central nervous system (CNS) involvement is suggested by seizures, axial hypotonia, scissoring of the legs on

vertical suspension, dysmorphic features, cortical thumbing/fisting (persistent adduction and infolding of the thumbs), and global developmental delay. Inappropriate transient lower esophageal sphincter relaxation (TLESR) is the predominant cause of GERD during childhood. Hgb electrophoresis is a highly sensitive and specific test that demonstrates Hgb S and Hgb F (fetal hemoglobin) in the newborn with SS disease. Myelomeningocele a. Physical therapy and restriction of vigorous exercise are effective. Immunohistochemistry and cytogenetic evaluation of tissue obtained by biopsy provides a definitive diagnosis. MRI or bone scan confirms the diagnosis. Marasmus is the most common energy depletion state and is characterized by near starvation from protein and nonprotein calorie deficiencies. Symptoms are usually apparent by 6 months of age. Plasma infusions may worsen the symptoms in this type of HUS, and if plasma exchange is considered, albumin should be used instead of plasma. Necrosis of blood vessels may lead to parenchymal hemorrhage. Necrotizing enterocolitis should be considered in any newborn who presents with rectal bleeding, feeding intolerance, or abdominal distension (see Chapter 4, section XII.E). Testotoxicosis is a rare disease in which the testes enlarge bilaterally independent of the HPGA. HBV is found in most body fluids, including blood, tears, saliva, semen, vaginal secretions, urine, feces, and breast milk. Universal screening for hypercholesterolemia, regardless of body mass index (BMI) or risk factors, has been recommended by the National Heart, Lung and, Blood Institute (NHLBI) and endorsed by the American Academy of Pediatrics (AAP). Damage also occurs as a result of the host immune response. Protein-losing enteropathies result in hypoproteinemia as a result of transudation of protein from inflamed intestinal mucosa. CD is a chronic disorder with high morbidity but low mortality. Physical examination should be comprehensive. Pathophysiology. Renal failure may occur. Mortality is < 1%. A low reticulocyte count indicates bone marrow failure or diminished hematopoiesis. There are multiple choice questions and their detailed answers both of correct and wrong choices. Fever will result in increased insensible losses, and maintenance fluids should be increased by about 10% for every degree of temperature above 38°C. Dehydration and electrolyte abnormalities from vomiting may occur. This occurs when the evening dose of insulin is too high, causing hypoglycemia in the early morning hours, resulting in the release of counter-regulatory hormones (epinephrine and glucagon) to counteract this insulin-induced hypoglycemia. This is a ciliopathy that occurs in several forms. When calculating maintenance fluids using the surface area method, the maintenance water requirement for children is 1500 mL/m^2/day. Abdominal ultrasound is the most common method used for diagnosing and monitoring acute pancreatitis. Diagnosis is suggested by findings on radiographs and MRI. Syncope with exercise is a “red flag” and should be fully investigated immediately by a pediatric cardiologist before any of the other interventions may be considered. Developmental delay, intellectual disability, and behavioral problems Neurofibromatosis type 1 (see also Chapter 19, Table 19-3) a. Rare causes of congenital torticollis include cervical spine deformities, for example Klippel–Feil syndrome [see section II.A.3]. DHT is responsible for penile enlargement, scrotal fusion, and the entire masculinization of the external genitalia. Ewing sarcoma may occasionally develop in soft tissue instead of bone. The infant’s diaper has a sickly sweet smell. If the hip is affected, it is usually held in flexion, abduction, and external rotation to relieve the pressure within the joint capsule. 204 2. Rhizomelic (proximal limb) shortening of the limbs, bowed legs, and trident appearance of the hands. Admit the patient to the hospital for overnight observation. The classic electrolyte presentation seen in renal tubular acidosis (RTA) is a hyperchloremic metabolic acidosis with a normal serum anion gap. When slow-decay markers are used, radioactive tracer detected in the lungs confirms aspiration. Primary ocular herpes simplex virus a. A line drawn flanking the superior edge of the femoral neck (Klein line) crosses 10–20% of the epiphysis in a normal hip on AP pelvis films. Table 1-4 Causes of Microcephaly Congenital Early prenatal infection (e.g., HIV infection, TORCH) Maternal exposure to drugs and toxins (e.g., fetal alcohol syndrome) Chromosomal abnormality (e.g., trisomy 13, 18, or 21) Familial microcephaly (autosomal dominant or autosomal recessive inheritance) Maternal phenylketonuria Acquired Late third trimester or perinatal infections Meningitis or meningoencephalitis during first year of life Hypoxic or ischemic cerebral insult Metabolic derangements (e.g., hypothyroidism, inborn errors of metabolism) TORCH = toxoplasmosis, other (syphilis), rubella, cytomegalovirus, herpes simplex virus; HIV = human immunodeficiency virus. Neonates with GH deficiency are typically not SGA. Constant pain suggests strangulation of the gut or torsion. The characteristic physical features of fragile X syndrome include large ears, macrocephaly, and large testes. 600 601 Review Test 1. Endoscopic therapy (e.g., photocoagulation, banding, vessel ligation, injection with sclerosing agents) is indicated for active bleeding or if rebleeding is very likely. In addition to failure to thrive, which of the following is a typical complication for a child with perinatally acquired HIV? Because there is no testicular tissue, there is no secretion of testosterone or of AMH, resulting in the regression of the Wolffian ducts and the development of the Müllerian structures, respectively. Contaminated honey is a common source of the toxin. The hip is most commonly affected in younger children, whereas the knee is more commonly affected in older children. Primary NS accounts for 90% of all childhood cases of NS. Table 5-4 Initial Evaluation for Inborn Errors of Metabolism Test Initial studies Reason for Test Serum glucose Rule out hypoglycemia CBC with differential Some organic acidemias may cause pancytopenia Urinalysis Assess for ketones: Presence of ketones is especially suspicious in newborns, because they do not normally produce ketones well Arterial blood gas and serum electrolytes In older children, absence of ketones with hypoglycemia is suspicious for fatty acid oxidation defect Assess for urine-reducing substances: Positive reducing substances with a negative dipstick for glucose is suggestive of galactosemia Assess for anion gap metabolic acidosis Plasma NH_3 Rule out urea cycle defects or organic acidemias Mild elevations can be nonspecific If metabolic acidosis is present: Serum lactate and pyruvate Rule out lactic acidemias or organic acidemias Urine organic acids Rule out organic acidemias 182 If increased ammonia is present: Plasma amino acids If elevated, then suspect aminoacidopathies Urine organic acids If elevated, suspect organic acidemias. Infants with suspected pyelonephritis should be admitted to the hospital for intravenous antibiotics for 2–3 days, pending the results of the catheterized urine culture and sensitivities, and then complete a 14-day course of antibiotics as an outpatient. Type 1—insulin deficiency 2. Which of the following is the next step in management? A 5-month-old female infant in a day care facility develops a low-grade fever (temperature up to 100.8°F [38.2°C]), rhinorrhea, and cough. This patient most likely has congenital muscular torticollis, a tilting of the head to one side as a result of intrauterine constraint or birth trauma. Grey Turner sign (bluish discoloration of the flanks) or Cullen sign (bluish discoloration of the periumbilical area) may be present in severe cases, caused by blood tracking along fascial planes. Inheritance is autosomal recessive. Recurrent middle ear dysfunction and obstructive sleep apnea 5. Three α-globin genes are deleted. 489 CHAPTER 13 490 Hematology Carol Diamond 491 I. This is an inflammatory condition, not infectious, and so antibiotic therapy is not indicated. She is otherwise well with no chronic medical conditions and is taking no medications. Witcoff, MD, Stanford, California Pediatric Pulmonologist, Past Clinical Associate Professor of Pediatrics, Stanford School of Medicine, Kenneth W. Macronutrients supply energy and essential nutrients needed for growth, development, disease prevention, and activity. Dialysis therapy (peritoneal dialysis, hemodialysis, or continuous renal replacement therapy) is used when conservative management fails to maintain the patient in safe biochemical, nutritional, and fluid balance. In the genetic XY male, testosterone made by fetal Leydig cells stimulates the development of the Wolffian ducts (epididymis, vas deferens, and seminal vesicles), and AMH made by fetal Sertoli cells inhibits the development of the Müllerian structures (fallopian tubes, uterus, and upper one-third of the vagina). A 7-year-old boy with complaints of shortness of breath, nonpruritic rash, and very painful migratory arthritis presents to your clinic for evaluation. Hydroxyurea, a chemotherapeutic agent that increases Hgb F, has been shown to decrease the incidence of vaso-occlusive crises. Soft tissue mass or tightness may be palpable within the sternocleidomastoid muscle, possibly representing a hematoma or fibrosis. MRI provides better detail to visualize the posterior fossa for evaluation of other causes of ataxia. There is a wide range of clinical features, but hepatosplenomegaly and progressive neurologic deterioration are constant features. Ocular examination (I-ARM acronym; see section I.B) c. In the steady state, when a patient has a normal Hgb level, the reticulocytes should constitute 1% of all RBCs. In most anemias, reticulocyte counts should rise. RBC aplasias are a group of congenital or acquired blood disorders characterized by anemia, reticulocytopenia, and a paucity of RBC precursors in the bone marrow. Visual acuity rapidly improves during the first 3–4 months of life, as a clear in-focus retinal image stimulates functional and structural development of the visual centers of the brain. Corneal abrasion 1. However, it is ineffective for some poisons such as iron, lithium, alcohols, ethylene glycol, iodine, potassium, and arsenic. Glucocorticoid excess 1. DDH was formerly termed congenital hip dysplasia; however, this name was abandoned to reflect the spectrum of findings possible, including a normal examination at birth with progression to dislocation later in life. Type 2 Diabetes Mellitus (Type 2 DM) VIII. Calculation of the urine anion gap (urine Na^+ + urine K^+ − urine chloride) is important; a positive urine anion gap is seen in Types I and IV RTA and may be seen in Type II RTA. Penicillin orally for 10 days 2. Acquired forms of RTA may be caused by nephrotoxic drugs (e.g., amphotericin) or systemic diseases (e.g., autoimmune disorders). Because of the difficulties breathing and swallowing in young premature infants, his caloric intake should be minimized. Brief, lasting 5–10 seconds 2. Typically, sodium, potassium, and phosphorus intake are restricted. Pediatric Dental Care VII. 428 VIII. This condition occurs sporadically or may have autosomal dominant inheritance. Hyperphosphatemia C. This patient likely has a renal vein thrombosis, which presents in infancy with the sudden onset of gross hematuria and unilateral or bilateral flank masses. Middle and Lower Respiratory Infections VII. Incidence is 1 in 10,000 live births. The subacute phase is also characterized by decreasing erythrocyte sedimentation rate (ESR) and by marked thrombocytosis. Aspartate aminotransferase (AST) elevation is a sensitive but nonspecific marker of hepatocyte injury. There are four stages of acetaminophen poisoning (Table 20-4). burgdorferi is passed into the bloodstream of the human host while the tick engorges itself with blood. Epidemiology a. This patient’s clinical features are consistent with bronchiolitis, a viral lower respiratory tract infection. *Sjögren syndrome. Lowe, MD, Attending Physician, Division of Emergency and Transport Medicine, Children’s Hospital Los Angeles, Los Angeles, California, Assistant Professor of Pediatrics, Keck School of Medicine at the University of Southern California, Los Angeles, California David J. Blunt trauma compresses the globe, and when the globe subsequently re-expands, the iris vasculature tears, resulting in bleeding. Miller We also owe so much to our present and past pediatric and medicine–pediatric residents with whom we feel very close, and to the thousands of medical students from all across the country, but most especially to those from Stanford University, from the University of Wisconsin School of Medicine and Public Health, and from the David Geffen School of Medicine at UCLA, who have stimulated us, energized us, and taught us so much along the way. Intravenous injection of edrophonium chloride, a rapidly acting cholinesterase inhibitor, produces transient improvement of ptosis. Fetal phenytoin syndrome is associated with mental retardation, nail and digit abnormalities, and cardiac defects. Endophthalmitis (infection within the vitreous and aqueous fluid), a rare but devastating infection that often results in blindness B. Strabismus, nystagmus, and abnormal back or neurologic examination should prompt evaluation for conditions other than congenital muscular torticollis. Laboratory studies a. Pulmonary function studies demonstrate decreased respiratory flow rates and air trapping, consistent with obstructive lung disease early in the disease process. Bleeding (occurs as a result of prolongation of the prothrombin time as a result of diminished hepatic synthetic function) 6. A type IV fracture is in the physis and through both the metaphysis and epiphysis. Weak cry, tongue fasciculations, and difficulty sucking and swallowing 2. This is in contrast to acrocyanosis (fingers, toes) that is typical in healthy infants, is transient, and does not require further evaluation. Brief tonic extension of the extremities and flexion of the trunk (jackknife or salaam seizures) lasting 1–2 seconds each, occurring in clusters of 5–10 seizures spread over 3–5 minutes 2. Prothrombin time (PT) f. Screening laboratories include CBC, electrolytes, liver function panel, stool for occult blood, stool for parasite screening, urinalysis, and erythrocyte sedimentation rate (ESR) or CRP. Infants with botulism have constipation, hypotonia, problems with suck and swallow, and progressive weakness that may lead to paralysis. Plain radiographs may reveal a widened joint space and may exclude other diagnoses in the differential (e.g., fracture) but are often normal. Menstruation begins at 9–15 years of age, with a mean onset of 12.5 years of age. Diagnosis of OTC deficiency 1. If negative, no further testing is indicated. Laboratory findings reveal elevated Hgb and Hct but normal platelet and WBC counts. Fetal Exposure to Drugs of Abuse XII. Boys may have no apparent genital abnormality. Other presenting features of systemic-onset JIA include fever, a characteristic transient skin rash (salmon-colored), hepatosplenomegaly, and lymphadenopathy. Girls present with virilization at birth, and boys present later in life (1–4 years of age) with tall stature and precocious puberty. On examination, you note a cherry-red macula and poor eye contact. Children as young as 2 months old may receive the first dose of a four-dose meningococcal vaccine series if they are at high risk (e.g., anatomic or functional asplenia, complement deficiency, travel to hyperendemic or epidemic regions), with administration of doses recommended at 2, 4, 6, and 12–15 months of age. Hematuria A. Growth III. Guillain–Barré syndrome is a demyelinating polyneuritis characterized by ascending weakness, areflexia, and paresthesias. Glucose is a basic energy source that can be used in any patient, regardless of the inborn error of metabolism, and should be administered intravenously in a patient who is markedly hypoglycemic and in coma or shock. In older children, acetazolamide may be used to decrease 459 ICP as well. Cervical adenopathy E. Premature closure of the sagittal suture results in an elongated skull 23 (termed dolichocephaly or scaphocephaly) and is the most common form of craniosynostosis (Figure 1-3). Additional disorders 1. Horner syndrome (ipsilateral ptosis, miosis, and anhydrosis) may be present if the sympathetic fibers of the first thoracic nerve have also been damaged. See section XII.D.2 for more details. Unlike UC, the inflammation is eccentric and segmental with “skip lesions.” 2. The younger the patient, the shorter the course of symptoms before DKA occurs. Management includes avoidance of excessive stimulation, such as examination of the pharynx with a tongue depressor, because this may induce respiratory distress. It increases viscosity of blood and may worsen crisis.) Abdominal pain and distension Often caused by sickling within mesenteric artery Low threshold for imaging the abdomen Same management as for painful bone crisis Dysarthria and hemiplegia, but may be asymptomatic Occurs in up to 11% of patients (subclinical stroke occurs in up to 20% of patients) Same management as for painful bone crisis Urgent exchange transfusion Patient should be started on chronic transfusion program to prevent recurrence, which occurs in 60–90% Painful, sustained erection Always consider SS disease in any patient presenting with priapism Same management as for painful bone crisis Acute chest syndrome (ACS) Sequestration crisis New pulmonary infiltrate associated with respiratory symptoms (e.g., cough, shortness of breath, chest pain) Hypoxemia May be severe and may cause up to 25% of deaths in patients with SS disease Causes include infection (e.g., viral, Mycoplasma pneumoniae, Chlamydia pneumoniae, Streptococcus pneumoniae), sickling, atelectasis, fat embolism, painful bone crisis involving the ribs, and pulmonary edema from fluid overload Careful hydration and pain management Oxygen Appropriate antibiotics (usually cefuroxime and azithromycin) Incentive spirometry Early use of partial exchange transfusion in a patient who does not improve rapidly Rapid accumulation of blood in spleen (or less commonly, liver) Occurs in patients 500,000 platelets/μL Increased Sickled cells, target cells, Howell–Jolly bodies Erythroid hyperplasia Table 13-5 Characteristics of the Red Blood Cell Aplasias Aplasia Congenital hypoplastic anemia (Diamond– Blackfan anemia) TEC Parvovirus B19–associated pure RBC aplasia* Etiology Clinical Features Laboratory Findings Treatment Unknown Autosomal recessive or autosomal dominant inheritance Anemia within the first year of life Rapid onset One-fourth to one-third have physical findings: craniofacial, renal, cardiac anomalies; short stature; triphalangeal thumbs Signs and symptoms of anemia ↓Hgb ↓Reticulocytes ↑Hgb F ↓or normal platelet count Marrow: ↓RBC precursors; other marrow elements normal RBC transfusion Corticosteroids (up to 70% respond) Bone marrow transplant if no response to corticosteroids Unknown Possible postviral autoimmune reaction Anemia begins >1 year of age Slow in onset Signs and symptoms of anemia ↓Hgb ↓Reticulocytes Normal platelet count Marrow: ↓RBC precursors Spontaneous recovery within several weeks No treatment required Parvovirus B19 infection Anemia generally not symptomatic May have associated URI symptoms and facial rash (“slapped cheeks”) of fifth disease Aplastic crisis in patients with SS disease ↓Hgb ↓Reticulocytes Normal platelet count Spontaneous recovery within 2 weeks RBC transfusions may be required for patients with aplastic crisis associated with SS disease * Note that Epstein–Barr virus, cytomegalovirus, human immunodeficiency virus (HIV), and drugs (e.g., chloramphenicol) may cause an acquired RBC aplasia similar to parvovirus B19. Giving 100% oxygen does not significantly improve the PaO_2 because deoxygenated blood bypasses the pulmonary circulation. Postmortem examination may demonstrate intrathoracic petechiae (the most common autopsy finding in 80% of cases, but one whose cause is unknown), pulmonary congestion or edema, small airway inflammation, and evidence of hypoxia. Obesity and obstructive sleep apnea 3. Other signs and symptoms are listed in Table 14-4, which compares osteogenic sarcoma with Ewing sarcoma, the second most common malignancy of bone in children. Neonates with CF may present with meconium ileus, an impaction of inspissated meconium that causes congenital intestinal obstruction. Key point: Children who grow 2 inches per year (5 cm per year) between 3 years of age and puberty usually do not have an endocrinopathy or underlying pathologic disorder. Salicylates. Acquired aplastic anemia is most often idiopathic. 498 3. Migraine with aura. Poor prognostic factors include male gender, age of onset 6 months, maximum parental education less than high school, lower socioeconomic status, a history of abdominal operations, and having unresolved family issues. This male-predominant, HLA-B27–related syndrome of arthritis affects the joints of the hips and sacroiliac joint. Male sexual differentiation is an active process, whereas female sexual differentiation develops by default when genetic and hormonal influences are absent. The cause of Tourette syndrome is unknown. If the first dose is not started until age 15 or older, three doses of vaccine are required. Immediate burning sensation with intense dysphagia, salivation, retrosternal chest pain, and vomiting 2. Lower intestinal contrast studies may reveal the cecum in the left abdomen or right upper quadrant. The course of disease is generally self-limited, with full recovery during infancy in as many as 70% of patients. Medications can “scavenge” ammonia (e.g., sodium benzoate binds with ammonia and provides an alternative pathway to excrete nitrogen). Electrophoresis demonstrates low or absent Hgb A and elevated Hgb F. Transmission is by the fecal–oral route through contaminated foods and water or by contact with contaminated individuals (e.g., food handlers). Juvenile myasthenia gravis presents in childhood secondary to AChR antibody formation. Etiology of organic CAP (Table 10-2) D. Atopic Dermatitis IV. Descriptions of the more common forms of anemia in childhood follow. Activated charcoal is contraindicated because it interferes with endoscopy. Laboratory evaluation should initially include CBC, electrolytes, and assessments of hepatic function. Patients with IgA nephropathy typically present in the second or third decade of life with recurrent bouts of gross hematuria associated with respiratory infections. You plan to admit her to the hospital for management. Central nervous system (CNS) bleeding is the most dreaded complication and is usually the result of head trauma. This occurs because of a transient recovery of residual β-cell function, resulting in endogenous release of insulin in response to carbohydrate exposure. H. The diagnosis can be made by DNA testing of the dystrophin gene, which shows a deletion in more than 90% of patients. 111. Most patients have mild to moderate bleeding, usually involving mucocutaneous surfaces. Electrolyte abnormalities and dehydration should be corrected before surgery. BUN and creatinine should be measured to assess for renal impairment. His lung disease should not affect his development. Patients have no anemia and are asymptomatic. Hepatitis B immune globulin, given to newborns born to hepatitis B–positive women c. Deletion on chromosome 7 E. In addition, no definitive relationship between ALTE and sudden infant death syndrome (SIDS) has been established. Because this dose is given before the recommended time of 12–15 months, it does not count toward the two-shot series and the child will subsequently need two additional doses at the appropriate times. Treatment for significant toxicity includes dimercaprol, British antilewisite (BAL), or calcium disodium ethylenediaminetetraacetic acid (EDTA). Hearing loss typically begins in childhood and progresses; approximately 50% of adults have some loss of hearing, ranging from mild to severe. Each of these conditions is treatable, and if not detected early, each leads to irreversible brain injury. Because of the risk of pneumococcal infection, if the child is febrile, evaluation should include blood culture, urine culture, and chest radiograph. Unilateral dilated nonreactive pupil suggests uncal herniation. In Esophageal atresia with distal tracheoesophageal fistula (Figure 4-5A), air that has crossed through the distal fistula from the trachea is seen in the stomach and intestine. This may obstruct and infarct the bowel. Ironrich foods (e.g., iron-fortified cereal, meats, legumes) or iron supplementation is therefore recommended beginning at 4–6 months of age to prevent anemia. This patient’s staring spells would be expected to last less than 10 seconds each. This acid damages adjacent intestinal mucosa, causing the classic presentation of painless, acute rectal bleeding in an otherwise healthy child. The hallmark of PPP is suppressed unstimulated gonadotropin levels and/or a flat response on GnRHa stimulation testing, because the HPGA has not been activated. Ehlers–Danlos syndrome is caused by a defect in type V collagen that results in hyperextensible joints, fragile blood vessels that cause easily bruised skin, tissue paper–thin scars, and cardiovascular complications (e.g., mitral valve prolapse and aortic root dilatation that can lead to dissection). Small bowel biopsy is the gold standard for diagnosis and demonstrates short flat villi, deep crypts, and vacuolated epithelium with lymphocytes. Serologic testing for collagen vascular diseases 6. Puberty refers to the activation of hypothalamic–gonadal axis. Nonspecific inflammatory markers a. Although antibiotic treatment of E. The epidemiology, etiology, clinical findings, and treatment are described in Table 13-2. Intravenous immune globulin has no role in the management of Duchenne muscular dystrophy. Ulcers associated with H. Nasogastric decompression, hydration, and correction of electrolyte abnormalities (typically hypochloremic metabolic alkalosis) are necessary. Signs and symptoms are listed in Table 14-4, which compares Ewing sarcoma and osteogenic sarcoma. 202 7. 26 FIGURE 1.3 Types of craniosynostosis. TPN may be needed if growth remains problematic. Other risk factors include low birth weight (10% BSA, full-thickness burns >2% BSA, burns to specific areas of the body (e.g., face, perineum, hands, feet, burns overlying a joint, or circumferential burns), suspected inhalation injury, and suspected nonaccidental trauma (i.e., inflicted burn). Psychosocial risk factors include personality (e.g., timid, nervous, anxious, overachiever), birth order (e.g., first-born and last-born), life stressors, including changes in daily routines (e.g., nanny, change in school), family stressors (e.g., divorce, separation, parental fighting), and methods of discipline that are either too extreme or submissive. Paralysis is symmetric and descending, and at times, diaphragmatic paralysis may also occur. The irritated esophagus may be painful, negatively reinforcing feeding, or the child may desire the buffering action of milk to reduce acid irritation. Abdominal radiographs may show gastric or proximal intestinal distension and obstruction, often with little or no distal bowel gas. The answer is E [I.F.2]. A 1-year-old boy has a 2-day history of irritability, decreased oral intake, decreased urine output, occasional watery diarrhea, and tactile fever. DIC = disseminated intravascular coagulation; HUS = hemolytic uremic syndrome; TEC = transient erythroblastopenia of childhood; G6PD = glucose-6-phosphate dehydrogenase; RBC = red blood cell. Monitoring 1. Given the nature of this patient’s symptoms and his young age, he is most likely to have atypical HUS. MMR and varicella vaccines: Intravenous Immunoglobulin (IVIG) administration within the preceding 11 months, which might interfere with the patient’s immune response to these live vaccines 34 IV. The victim may appear alert initially or may be agitated, combative, or comatose. Acquired aplastic anemia a. 368 III. If you want to study pediatrics then you should download BRS Pediatrics pdf below. The defect is a centrally localized herniation of abdominal organs through the umbilical ring with a true hernia sac (abdominal organs are covered with a peritoneal sac). Seizures within 3 days after prior vaccination 4. Atypical HUS (aHUS) 1. The answer is D [VII.D]. However, increasing evidence suggests that nonperforated appendicitis can be treated nonsurgically with antibiotics alone. Duration of treatment for cystitis is usually 7–10 days, and for pyelonephritis, 14 days. Laboratory studies reveal a white blood cell count of 14,000 cells/mm^3, hemoglobin of 11.8 g/dL, and platelet count of 260,000 platelets/μL. Neutropenia Review Test Answers and Explanations Chapter 14 Oncology I. Screening laboratory tests include serum glucose, serum and urine toxicology screens, and electrolytes. Virus is shed in the stool 2–3 weeks before the onset of symptoms and 1 week after the onset of jaundice. It is associated with PR3 antineutrophil cytoplasmic antibody, also known as the C-ANCA. Laboratory evaluation should include a complete blood count (CBC), Creactive protein (CRP), urinalysis, and metabolic panel. On examination, patients have enlarged calf muscles (pseudohypertrophy) as a result of fatty infiltration of the degenerating muscles, and laboratory studies reveal elevated creatine kinase levels. Ophthalmoplegic migraine. The answers are B and D, respectively [, sections XI.A.4 and XII.D]. If a child has a strong preference for one eye and constant suppression of the nonpreferred eye, amblyopia will develop in the nonpreferred eye. Microscopic hematuria may be an indicator of a serious medical condition such as a tumor or chronic glomerulonephritis or may be of no serious medical consequence. Vaccine is indicated in children with an immunocompromising condition and and for those with functional or anatomic asplenia. Parents deny fever, cold symptoms, or diarrhea. Disorders occur when a specific step in a metabolic pathway is disrupted in some way (defective enzyme, receptor problem, etc.), resulting in the accumulation of toxic metabolites. Diabetic ketoacidosis (DKA) may be the initial presentation in 25% of patients [see section VIII]. Anemia is one of the most common laboratory abnormalities during childhood. It is not usually associated with other anomalies. Toxic megacolon. Anorexia, distension, and blood-tinged stools are common. Any evidence of burns to the lips, nose, or tongue, such as singed nasal hairs or presence of soot in the nasal/oral airways, indicates a risk for progressive airway compromise. The differential diagnosis of unconjugated hyperbilirubinemia is presented in Chapter 4, section X.C and Figure 4-3. After birth, the oxygen saturation in healthy term infants gradually increases over the first 10 minutes of life and parallels the resolution of central cyanosis. Singh, MD, FAAP, Fellow, Pediatric Infectious Diseases, Cedars-Sinai Medical Center, Los Angeles, California Liliana Sloninsky, MD, Associate Pediatric Hematologist-Oncologist, Cedars-Sinai Medical Center, Los Angeles, California, Clinical Assistant Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Augusto Sola, MD, FAAP, Director, Neonatal-Perinatal Medicine, Professor of Pediatrics and Obstetrics and Gynecology, Emory University School of Medicine, Atlanta, Georgia Jerome K. Nephrotic Syndrome (NS) VII. There is a 50% chance that a subsequent sibling will have the same illness. Which of the following is most accurate regarding the most likely diagnosis? Types of SB 1. Bites range in severity from scratches, punctures, and lacerations to severe soft tissue injury. Because there is no aldosterone deficiency, there is usually no electrolyte abnormality. Bracing is not indicated because it only prevents progression of scoliosis during the 637 6. Most patients develop severe hypertension and renal insufficiency, eventually requiring transplantation. Affected infants may be irritable but hungry. The goal of the repletion phase is a more gradual correction of the patient’s water and electrolyte deficits. Down syndrome C. Management focuses on supportive care and reestablishing bile duct continuity. Hirsutism 4. Older children with GH deficiency may have obesity and cherubic facies. His fever will result in increased insensible losses, and maintenance fluids should therefore be increased by 5% for every degree of temperature above 38°C. Blood cultures are not positive in this infection. Well-Child Screening V. Some patients have a chronic relapsing course (recurrent HUS). Renal manifestations initially include hematuria, often gross hematuria, and sometimes hypertension. Guillain–Barré syndrome E. Hematuria may be seen on voiding (gross hematuria) or only on urinalysis (microscopic hematuria). Physical examination should be performed after rapport with the patient has been established. Victims may suffer asphyxia from aspirating liquid or from laryngospasm. Examination reveals that the boy is very 195 tall (>95th percentile) with long, thin arms with fingers of normal size. Chest radiographs are not routinely indicated for asthma exacerbations. Plus. Hep A vaccine a. Metabolic disorders: neonatal adrenoleukodystrophy, primary carnitine deficiency, acid maltase deficiency, urea cycle defects, and Zellweger syndrome 5. Features of cholestatic liver disease are indistinguishable from neonatal hepatitis and biliary atresia. Empiric coverage of both rickettsial and typical bacterial pathogens includes doxycycline and a third generation cephalosporin. However, tics occur in 3% of children. A structural abnormality of the urinary tract, such as vesicoureteral reflux, predisposes a child to developing a UTI. B. Nonessential nutrients can be synthesized from other compounds or may be derived from the diet. coli (EHEC) and hemolytic uremic syndrome (HUS), given her decreased urine output and pallor. Two-thirds of patients have an abnormality on chromosome 20 affecting the JAG 1 gene. Diagnosis of GERD is based on the number of acidification episodes, the total duration of esophageal acidification, and the time required for the pH to normalize in the presence of symptoms. Early neurologic signs include neck pain, loss of motor skills, or loss of bowel/bladder control. Vision problems should always be considered in a child with a gaze abnormality, but the presence of a stiff neck muscle makes it unlikely that a problem with vision is the cause of the torticollis. As marrow activity increases, the marrow spaces enlarge, increasing the size of bones in the face, skull, and other bones if severe and untreated. Mumps virus is the most 765 92. Which of the following statements is most accurate regarding this patient’s likely diagnosis? The answer is D [XIV.A, XIV.E.1, and XIV.F]. Etiologies are typically from ophthalmologic, central nervous system, musculoskeletal, infectious, or traumatic causes, and they may include strabismus and refractive errors, posterior fossa tumors, syringomyelia, atlantoaxial subluxation [see section II.A.2], cervical adenitis, retropharyngeal abscess, cervical discitis, osteomyelitis, trauma, gastroesophageal reflux disease (Sandifer syndrome), and dystonic drug reactions. Maintenance fluid calculations should not be adjusted for increased ongoing losses, such as profuse watery diarrhea. Other findings a. A 4-month-old male infant has generalized weakness, hypotonia, areflexia, and tongue fasciculations. Isometric contraction of the temporalis, masseter, or trapezius muscle often accompanies the headache. Germ cell tumors are more common in patients with sex chromosome abnormalities. Diminished interest in activities, fatigue, and energy loss are other general symptoms. The cause is generally unknown. Given continuous, rapid advances in medical science and health information, independent professional verification of medical diagnoses, indications, appropriate pharmaceutical selections and dosages, and treatment options should be made, and health care professionals should consult a variety of sources. Rates of meningococcal disease have been declining since 1990, which is thought to be a consequence of increased vaccination use. Benign familial macrocephaly, associated with an otherwise normal physical examination and a family history of large heads b. Physical abuse 1. Patients with systemiconset JIA may be either male or female. Migraine headaches are prolonged (often 30–60 minutes, but may last up to 72 hours), unilateral (may be bilateral in children) headaches that are associated with nausea, vomiting, or visual changes and are caused by changes in cerebral blood flow. Congenital lesions of the gut, such as gastroschisis, volvulus, or intestinal atresia, may require surgical resection of the small intestine, resulting in an inadequate absorptive surface area. Increased red blood cell (RBC) galactose-1-phosphate in blood b. It is often performed when the diagnosis is unclear, or to correlate reflux with events such as apnea. Any pathologic condition that causes a blurred visual image. Brown 362 I. 647 IV. Coccidioidomycosis is endemic in the Southwestern United States, northern Mexico, and areas of Central and South America. All patients should meet with a registered dietician. Infection is asymptomatic in the majority of children (>70%). Other Rheumatologic Conditions of Childhood Review Test Answers and Explanations Chapter 17 Orthopedics I. Cornelia de Lange syndrome a. Known sources of bacterial contamination include undercooked beef, unpasteurized milk, produce, and contaminated fruit juices. Antibiotics may be necessary if bacterial superinfection occurs. Although contiguous spread and direct inoculation can cause osteomyelitis, children most commonly acquire infection by hematogenous seeding. Orbital fat and the inferior rectus muscle can become entrapped within the fracture, leading to diplopia as a result of restricted vertical eye movement, to strabismus, and to enophthalmos (backward displacement of the globe into the orbit). Instrument-based objective vision screening with devices that measure the focusing and alignment of eyes in preverbal children Table 18-1 Differential Diagnosis of an Abnormal Red Reflex Disorder Cataract Vitreous hemorrhage Retinoblastoma Anisometropia Strabismus Glaucoma Red Reflex Finding Dark, dull, or white reflex Dark or dull reflex Yellow or white reflex Unequal red reflex Brighter red reflex in deviated eye Corneal light reflex will be uncentered Dull reflex 641 II. Table 14-4 Clinical and Radiographic Features of Osteogenic Sarcoma and Ewing Sarcoma Feature Site Local and systemic findings Radiographic findings Metastases Osteogenic Sarcoma Metaphysis of tubular long bones 50% occur near the knee Most common sites (in order): distal femur, proximal tibia, proximal humerus, proximal femur Pain, swelling, and soft tissue massSystemic symptoms uncommon Periosteal reaction with “sunburst” appearance Lytic; or mixed lytic and destructive changes Occurs in 15% at presentation Lungs (90%) and bone (10%) ESR = erythrocyte sedimentation rate. Both are inactivated. Newborn screening tests for additional metabolic disorders (e.g., congenital adrenal hyperplasia) are performed in some states. The answer is D [V.L.3]. Juvenile polyps are the most common cause of significant lower GI bleeding beyond infancy. Genitourinary tract is the second most common site of involvement (20% of cases). pylori infection. If head CT is obtained to rule out acute life-threatening causes, it is normal in acute cerebellar ataxia. Although surfactant is first noted in the lungs at approximately 23–24 weeks’ gestation, a sufficient quantity of effective surfactant is produced only after 30–32 weeks’ gestation. Chronic hyponatremia should be corrected much more slowly. Proper eye alignment b. Head and neck, including the orbit, are the most common sites of involvement (40% of cases). Laboratory tests should include serum electrolytes, BUN, creatinine, urinalysis, and urinary protein and creatinine levels, as well as a more specific investigation for the cause of kidney injury, such as testing for ANCA or drug levels of nephrotoxic medications based on the clinical history. Increased muscle enzymes (creatine phosphokinase, aspartate aminotransferase, alanine aminotransferase, lactate dehydrogenase, and aldolase) 3. Migraines are the most common cause of headaches in children and adolescents. The toxin prevents the presynaptic release of acetylcholine. Postreduction radiographs are not needed. There is no cure, but oral steroids can improve strength transiently and prolong duration of ambulation when the disease is in the early stages. It is commonly given in conjunction with MMR vaccine. Intravenous antibiotics and urgent NLD probing if infection is present 648 V. Vitamin D and calcium supplementation may be necessary to help treat osteopenia and decrease frequency of fractures. There are no specific eye abnormalities in congenital myotonic dystrophy, muscular dystrophy, or cerebral infarcts. Deficiency of the late components of the classic pathway of complement (C5, C6, and C8) is associated with susceptibility to meningococcal infection. Other medications used to prevent N. The answer is C [, section IX.B.4.a]. Constipation VIII. Erythema marginatum is an evanescent, nonpruritic rash that starts as pink to red macules, which may coalesce and spread centrifugally with central clearing over the trunk and proximal limbs. Chronic Kidney Disease (CKD) and End-Stage Renal Disease (ESRD) A. Hypokalemia B. Appropriate polycythemia may be caused by chronic hypoxemia as a result of cyanotic congenital heart disease (the most common cause of polycythemia in childhood), pulmonary disease, or living at high altitudes. None of the other answer choices are associated with the signs and symptoms present in this patient. NLD probing, in which a small steel wire is passed through the nasolacrimal system through Hasner valve into the nose, will cure NLD obstruction in most cases. Abnormal visual development results from the following: a. There are many clinical notes and case studies. The level should then be plotted on the Done nomogram to assess for potential toxicity. Plasma amino acids 1. Retinal hemorrhages 1. Tumors of the head and neck and the genitourinary tract have the best prognosis, with a cure rate >90% if localized. NS that results from other primary glomerular diseases, including focal segmental glomerulosclerosis (FSGS), MPGN, IgA nephropathy, and PSGN 3. While there is no evidence that neither vaccines nor preservatives such as thimerosal are linked to developmental disabilities, as a precautionary measure the Food and Drug Administration (FDA) and the Centers for Disease Control and Prevention (CDC) eliminated thimerosal from vaccines in 2001. Epidemiology 1. Some infants have an associated polysplenia syndrome with multiple spleens, bilobed lungs, abdominal heterotaxia, and situs ambiguous. Table 5-2 Teratogens and Associated Anomalies Drugs Alcohol Cigarette smoking Cocaine Diethylstilbestrol (DES) Isotretinoin Lithium Phenytoin Thalidomide Valproic acid Warfarin Associated Anomalies Microcephaly; short palpebral fissures; long, smooth philtrum; variable intellectual disability; and congenital heart disease Small for gestational age Premature birth Increased risk of vaginal adenocarcinoma and genitourinary anomalies Small low-set ears, micrognathia, depressed nasal bridge, cardiac defects, thymic hypoplasia, and central nervous system malformations Ebstein anomaly Wide anterior fontanelle, short nose, wide mouth, microcephaly, low hairline, hypoplastic nails, and developmental delay Phocomelia (malformed extremities resulting in flipperlike appendages) Neural tube defects Hypoplastic nose with depressed nasal bridge, stippled epiphyses, and hypoplastic nails 179 V. Neonatology. Peripheral nervous system involvement is suggested by atrophy, muscle fasciculation, and loss of deep tendon reflexes. Physical examination is remarkable for a nontoxic, alert, and afebrile child. The streptococcal strains that cause streptococcal skin infection (i.e., impetigo) do not cause rheumatic fever. pH probe measurement is the gold standard for diagnosis but is not indicated for every child with suspected GERD. Osteoarthritis 5. The answer is B [, section XIV.E-F]. Pancreatic insufficiency 9. Features: Here are some of the unique features of this book: This is the best short book of pediatrics. A 3-year-old Caucasian girl is brought to your office with a 3-week history of bruising, left elbow pain, and fever. A history of motion sickness is common. Which one of the following statements about vitamin D supplementation is correct? Hypercortisolism presents with growth impairment and delayed bone age, but obese patients have normal to fast growth and an advanced bone age. Intravenous glucose should be administered. Hepatomegaly is not typically associated with esophageal atresia. Sodium benzoate should be administered. DDH is more common in girls than in boys. CSF or blood draining from the nose or external auditory canal may indicate a basilar skull fracture. Hepatomegaly 5. Activated partial thromboplastin time (aPTT) e.

 Urinary dipstick detects the presence of hemoglobin or myoglobin in the urine. Frequent, tens to hundreds per day 3. Diagnosis must be rapid to affect outcome. As allergy immunotherapy can significantly reduce the rate of reactions in those sensitive to insect venoms, all patients should be referred to an allergist for consideration of venom immunotherapy. Hirschsprung disease may present with colitis and abdominal distension. Strabismus Review Test Answers and Explanations 8 Chapter 19 Dermatology I. Coprolalia, the utterance of obscene words, is a dramatic symptom that occurs in 90% of patients. Numerous studies have shown a beneficial effect on long-term disease activity and survival. The Hypotonic Infant II. Myotonia is not present in the newborn but develops later, almost always by 5 years of age. This arthritis is associated with ulcerative colitis or Crohn disease. The current recommendations for the treatment of attention deficit/hyperactivity disorder (ADHD) vary based on the age of the patient. Perform imaging (testicular, abdominal, CNS, etc.) studies, depending on the suspected etiology. The answer is E [Figure 11-2 and Table 11-3]. Acid inhibition with antacids, histamine H_2-blockers, and proton-pump inhibitors is effective in reducing the irritation caused by the refluxate. Blood culture will be positive in 50% of cases. Anterior shoulder dislocation is the most common type of shoulder dislocation. These may be recalled using the mnemonic CHIPE (chloral hydrate and calcium, heavy metals, iodine and iron, phenothiazines, enteric-coated tablets). Reassure the parents that no workup or medications are necessary at this point. Ulcerative Colitis (Table 10-4) a. Complete recovery is the rule in children but depends on the severity and extent of the weakness. The two most common categories of normal variant short stature are familial (or genetic) short stature and CDGP. DDAVP is most useful in type I disease and is sometimes effective in type II disease. Bilateral constricted reactive pupils suggest opiate ingestion or pontine injury. Often there is a family history in one parent (i.e., mother had late menarche or father had his growth spurt late in high school or in college). The clinical course may be complicated by calcium deposition in the muscle, fascia, and subcutaneous tissue. Characterized by short stature, congenital heart defect, pectus deformity, and characteristic facies 2. SB occulta and meningocele have excellent prognoses as a result of the absence of neurologic deficits. Hematuria III. Guillain–Barré syndrome (acute inflammatory demyelinating polyneuropathy) typically presents with ascending paralysis without sensory loss. β-Thalassemia major occurs predominantly among patients of Mediterranean background. Ursodeoxycholic acid, a bile acid, is used to enhance bile flow and to reduce bile viscosity. Urine culture remains the "gold standard" for diagnosis. Individuals who are susceptible to celiac disease have a genetic predisposition, often due to HLA genotypes HLA-DQ2 or HLA-DQ8. In contrast, patients with myasthenia commonly present with ptosis. Treatment includes stretching exercises with physical therapy to relieve the muscle contracture. However, imaging is helpful if the examination is equivocal or the patient is at high risk, such as an infant who was breech in utero or children with positive family histories. Causes include blunt trauma (most common), infections (e.g., mumps, enterovirus, Epstein–Barr virus [EBV], human immunodeficiency virus [HIV], hepatitis A virus [HAV] and hepatitis B virus [HBV]), drugs and toxins, congenital anomalies, and obstruction. Child abusers come from all socioeconomic, cultural, and ethnic groups. Medical therapy to control UGI bleeding may be useful. Steroid-dependent and frequently relapsing children may achieve a prolonged remission during treatment with cyclophosphamide, mycophenolate mofetil, tacrolimus, or cyclosporine. It reflects involvement of the basal ganglia and caudate nuclei. Treatment should be initiated as soon as the diagnosis is established because of the risk of respiratory muscle paralysis. Detection of hematuria may be by urinary dipstick or by microscopy. Active bleeding is assessed by obtaining a nasogastric tube aspirate for fresh blood. Coller, MD, MPH, FAAP, Chief, Division of Pediatric Hospital Medicine, Assistant Professor of Pediatrics, University of Wisconsin School of Medicine and Public Health, Madison, Wisconsin, Lee Todd Miller, MD, FAAP, Associate Dean for Student Affairs, Professor of Pediatrics, Director, Global Health Education Programs, David Geffen School of Medicine at UCLA, Los Angeles, California. Facial wounds 1:16. (See Chapter 20, section II.D for a discussion of the management of shock.) b. Decerebrate posturing (extension of arms and legs) is seen in diffuse toxic/metabolic disorders or midbrain compression. This positioning is often described as the "waiter's tip." 2. Sydenham chorea is a self-limited autoimmune disorder and is one of the major Jones criteria for diagnosis of rheumatic fever (see Chapter 16, section VI). Aluminum B. Microvascular complications include diabetic retinopathy, nephropathy, and neuropathy. Management should include intravenous immune globulin. IgA nephropathy is most common in Asia and Australia and in Native Americans. Associated anomalies and complications 1. Vesicoureteral reflux (VUR) is caused by abnormalities of the ureterovesical junction, most commonly a shortened submucosal tunnel in which the ureter inserts through the bladder wall. Mild scoliosis is evident. He has no known history of prior bleeding or easy bruising but does have a 4-day history of upper respiratory tract infection (URI) symptoms and low-grade fevers. Disorders of Hemostasis A. MCAD may resemble Reye syndrome and accounts for 5% of cases of sudden infant death. Practical Approach to Neuroanesthesia. Common Behavioral Concerns Review Test Answers and Explanations Chapter 3 Adolescent Medicine I. Visual disturbances include blurred vision, scotomata, and jagged streaks of light that take on the outline of old forts (fortifications). Patients present with hypotonia and often have feeding and respiratory problems. Defects in Amino Acid Metabolism The characteristics of phenylketonuria (PKU), tyrosinemia type 1, and maple syrup urine disease are summarized in Table 5-5. Systemic causes include anemia, hypoglycemia, depression, and hypertension. Malrotation and volvulus are discussed in Chapter 10, section IV.B. 5. Muscle disorders: congenital myotonic dystrophy, congenital myopathy, and congenital muscular dystrophies 6. Other congenital defects (e.g., malrotation, esophageal atresia, congenital heart disease, renal anomalies) may also be present. Nine to twenty-four months of age: owing to inadequate intake and inadequate iron stores (which are typically depleted by 4–6 months of age). FSH stimulates the ovaries to produce ovarian follicles, which in turn produce estrogen. Hypertension V. HLA is responsible for antigen presentation. Surgical treatment is usually indicated for duodenal ulcers with active arterial bleeding, perforation, or varices. Disorders of blood vessels. Treatment of complications a. Prognosis varies according to the underlying cause. In the first 2 years of life, a downward shift in the height percentile is not uncommon and may reflect familial (or genetic) short stature, or constitutional delay of growth and puberty (CDGP). Characteristic skeletal findings: a. Patients are at risk for infection with encapsulated bacteria (i.e., Haemophilus influenzae type b, Streptococcus pneumoniae, Salmonella, Neisseria meningitidis). Children appear restless. Ascending, symmetric paralysis may progress to respiratory arrest. Ferry, MD, FACC, Managing Partner, Pediatric Cardiology Medical Associates of Southern California, Encino, California, Attending Pediatric Cardiologist, Children's Hospital Los Angeles, Los Angeles, California Carole Hurvitz, MD, FAAP, Director, Pediatric Hematology-Oncology, Cedars-Sinai Medical Center, Los Angeles, California, Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Elaine S. Autoantibodies against islet antigens (GAD65, ICA512, insulin, and ZnT8) are present in ~85% of patients. Acquired causes of hydrocephalus include intraventricular hemorrhage (most common in preterm infants), bacterial meningitis, and CSF-producing brain tumors. Coronary artery aneurysms in Kawasaki disease are more likely to occur during the subacute phase of the disease, which begins 1–2 weeks after the onset of fever. Blood smear to evaluate platelet morphology d. This protein-deficient state is characterized by generalized edema, abdominal distension, changes in skin pigmentation, and thin, sparse hair. Phenobarbital is the drug of choice for this patient's condition. EMG demonstrates decreased nerve conduction velocity or conduction block. This patient's clinical presentation is not consistent with Henoch–Schönlein purpura, which is characterized by abdominal pain, palpable purpura on the buttocks and thighs, and joint symptoms. Management should be initiated as soon as the diagnosis is established because of the risk of respiratory muscle paralysis. Laboratory tests should be reserved for cases where the history or physical examination suggests a specific etiology for which laboratory testing will be helpful or for the child who fails to respond to nutritional interventions. Other laboratory tests (e.g., polymerase chain reaction, cultures from body fluids or tissue) are occasionally used, but their clinical utility is limited, offering no advantage over the recommended serologic method. Feeding problems tend to subside with time. Approach to Unsteady Gait VIII. Long-term prognosis of FAP is poor. 542 IX. Evaluation of FTT requires a careful history and physical examination, a complete dietary history, and observation of the parent–child interaction. Live attenuated (OPV), administered orally a. Normal sexual differentiation (Figure 6-3) 1. coli and Shigella dysenteriae type 1. Diagnostic studies 1. Asthma is graded on a severity scale on the basis of the frequency of asthma symptoms during the day and night, and also on pulmonary function testing. Differential diagnosis of the undervirilized male (i.e., a neonate who usually has a 46, XY karyotype with ambiguous genitalia and one or both testes palpable; Figure 6-4). Success is highest if the procedure is performed by 50–70 days of age. Lerner, MD, FAES, Associate Chief of Clinical Pediatric Neurology, Associate Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Calvin G. Talipes equinovarus ("club foot") is present at birth and has a fixed inversion and equinus position of the foot with minimal flexibility. This patient is most likely to have had a prodromal gastroenteritis with Salmonella typhi. Intestinal mucosal atrophy c. There is no gender predilection. The earlier the onset of the disease in the mother, the more likely she will have affected offspring. The younger the child, the more likely it is that the tumor will be malignant. This patient's presentation with increasing weakness in the lower extremities and calf enlargement is consistent with Duchenne muscular dystrophy, an X-linked progressive degenerative muscle disorder for which there is no cure. Emergency phase a. Timing of vaccination: Hep A vaccine is a two-dose vaccination series recommended for all children starting at 1 year of age. A 6-week-old female infant presents with increased work of breathing and a staccato-type cough for 3 days. Any pathologic condition that blocks retinal stimulation, such as a congenital cataract [see section VIII.A] 5. Corticosteroids are indicated and will improve the patient's clinical course. Developmental screening 3. Vitamin D facilitates the dietary absorption of calcium from the gastrointestinal (GI) tract. This book of pediatrics is especially written for USMLE step 2 preparation. Organic causes of constipation represent fewer than 5% of cases of childhood constipation and are often present early in infancy. Patients with nephrotic syndrome are susceptible to infections with encapsulated organisms, such as pneumococcal infections, and are at risk for developing peritonitis, pneumonia, and overwhelming sepsis. Varicella zoster immune globulin (VZIG) for immunocompromised patients who have been exposed to varicella and are at high risk for severe varicella infection, including newborns born to mothers who develop chicken pox between 5 days before and 2 days after delivery. Abnormal respiratory responses may indicate the location of brain injury or its cause. First-time or occasional febrile seizures are not treated with anticonvulsants. This condition is the most common cause of a renal mass in the newborn, occurring in 1 in 4300 live births, and is most often unilateral. If present, symptoms may include malaise, anorexia, vomiting, fever, diffuse or right upper quadrant abdominal pain, edema, and bruising or bleeding. Achondroplasia a. (See Chapter 1, section IV.J.) b. Incidence is 1 in 70,000 live births. Bracing for immobilization may be necessary if pain persists. Inadequate caloric intake is the most common cause of FTT, followed by excessive caloric requirements. Menstrual Disorders IX. Primary headaches are more common and are idiopathic, not occurring for a specific reason, or the result of an underlying disease. Treatment at specialized centers can minimize complications and improve outcomes. Features include microphthalmia, holoprosencephaly, and cleft lip and palate. 110. The goal of assessment of the comatose patient is to determine the depth of coma, to identify the neurologic signs that indicate the site and cause of the coma, and to monitor the patient's recovery. NEC is one of the most common surgical conditions in preterm neonates with an incidence as high as 8–10% in infants 100 beats/minute, 1 point for slow and irregular respirations, 1 point for having some flexion of the extremities, 1 point for reflex irritability or grimace when a catheter is placed in her nose, and 1 point for her peripheral cyanosis or acrocyanosis, for a total Apgar score of 6 points. Type 1 Diabetes Mellitus (Type 1 DM) VII. This arthritis of the small and large joints is seen in patients with or without psoriatic skin disease. A decrease in appetite, which eventually could lead to significant weight loss, fulfills one of the criteria listed in the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) to make a diagnosis of depression. Intravenous immune globulin (IVIG), given for 2–4 days, is the preferred treatment for children because of its relative safety and ease of use. Which of the following statements regarding this patient's presentation and subsequent workup is correct? RTA refers to a group of congenital or acquired disorders that result from the inability of the kidney to maintain normal acid–base balance because of defects in bicarbonate conservation or because of defects in the excretion of hydrogen ions. The growth curve demonstrates poor growth velocity (less than 2 inches or 5 cm per year). Age younger than 4 years, especially younger than 1 year 2. Platelet function is normal. 25 FIGURE 1.2 Cranial sutures in the neonatal skull. Alanine aminotransferase (ALT) elevation is a more specific marker of liver disease but is also found in muscle tissues. These infants should be distinguished from those with high birth weight (>4000 g). Activated charcoal has a very large adsorptive surface area that binds toxins and minimizes their absorption. Attempts to move the joint result in significant pain. The answer is B [IV.B.3–5, IV.B.7, and Figure 9-3]. Etiology (Table 11-3) C. The diagnosis of Kawasaki disease is unlikely given the absence of fever for a minimum of 5 days and the absence of at least four of the five diagnostic criteria. The purpose of routine well-child care is to address the longitudinal health care needs of children from birth through adolescence. Patients with mild phenylketonuria (PKU) may present in childhood with developmental delay, hyperactivity, and a classic mousy or musty odor. Urgent electroencephalography (EEG) should be considered, even in patients without a history of clinical seizures. Certain clinical features are associated with vitamin and mineral deficiencies (Table 10-1). Intestinal obstruction may be functional or mechanical, and if mechanical, this condition may be acquired or congenital (see Figure 4-6 for a complete differential diagnosis). Macroorchidism develops in adolescence. Complete healing usually occurs within 24–48 hours. See Chapter 3, section I.A.2.c. 5. Neuroimaging (magnetic resonance imaging [MRI] is preferable with thin cuts through the pituitary) is appropriate if GH deficiency or concerns for other CNS pathology exist. Usually presents between 4 months to 4 years of age after a period of fasting or during an illness that caused vomiting. Advanced paternal age (>45 years old) increases the risk of having an affected child. These anemias are characterized by normal size (normal MCV) and shape of the RBCs. 1. Although obtaining a chest x-ray and arterial blood gas and determining body surface area burned are important management steps, they are not as important initially as airway management. Essential nutrients cannot be synthesized by the body and must be derived from the diet. Treatment initially includes the ABCs. 1. The gene has been identified on chromosome 19. He required 3 months of mechanical ventilatory support and oxygen therapy. Liver disease a. Chemical irritation (chemical conjunctivitis) results from drops or ointment that are topically instilled into a newborn's eyes as prophylaxis against N. Etiology (Figure 10-1) 3. Diagnosis is made by observing excessive movement at the C1 and C2 junction on lateral cervical spine radiographs in the neutral positions followed by flexion– extension radiographs. Patients with tyrosinemia type I present with peripheral neuropathy and renal and liver disease, and may produce an odor of rotten fish or cabbage. Types of hydrocephalus 1. The most common etiology of FTT is inadequate intake of calories, which may result from a disturbed parent–child bond. Hemorrhagic disease of the newborn is the result of vitamin K deficiency. Many patients treat themselves by lying in a dark, quiet room. Buy in Hard Copy (free shipping): You can Buy this book in 38$ only with free shipping here. Transient hyperammonemia of the newborn is a self-limited disease that may present in premature infants within the initial 24–48 hours of life. 137 138 V. Acute and recurrent episodes of altered mental status, vomiting, acidosis, organ failure, or ataxia c. Differential diagnosis of hypotonia (Figure 12-1) 1. Physical examination should include evaluation of the head and neck, chest, abdomen, back, and genitourinary system. 28 III. Drowning VI. Patients with CPP have a dramatic increase in LH secretion when compared with baseline levels. f. A 4-year-old boy presents to the emergency department for evaluation after 1 day of abdominal pain and a petechial rash on his bilateral thighs. DTaP a. Children with this diagnosis typically present with distal muscle weakness with an ascending pattern. Universal newborn hearing screening is recommended by 1 month of age, with confirmation of an abnormal screen by 3 months of age. The Pavlik harness holds the head of the femur against the acetabulum to stimulate formation of the normal cup shape of the acetabulum. Most cases of childhood nephrotic syndrome (two-thirds) occur in children younger than 5 years of age. Retinal hemorrhages appear as hemorrhagic dots and blots, or hemorrhage within the preretinal vitreous on a dilated funduscopic examination. Jaundice typically begins cranially and extends caudally as bilirubin increases. Figure 10-2 describes the time course of infection and the presence of detectable antibodies to HAV. A 10-year-old girl presents for evaluation of fatigue, diminished appetite, and weakness. Precocious puberty that is independent of the HPGA (i.e., caused by the peripheral production of male or female sex steroids and not FSH- or LH-mediated). Functional fecal retention (FFR) results from inappropriate constriction of the external anal sphincter, and is the most common form of constipation during childhood. An accurate description of a fracture is important when determining a need for surgical intervention or consultation with an orthopedic specialist. 472 Headaches occur in the absence of any warning symptoms. E. A poison control center may be consulted to assist with management. Topical or systemic medications, such as β-adrenergic and carbonic anhydrase inhibitors, may help to lower intraocular pressure. Proteins 1. NLD probing by ophthalmology may be needed in older infants if the obstruction does not resolve. Philadelphia: Lippincott Williams & Wilkins, 2013. Local causes include sinusitis, perioral abscess, toothache, chronic otitis media, or refractive errors. It is useful when symptoms worsen, when respiratory effort is compromised, or when the patient is unresponsive to other therapies. Excessive exposure may occur from fires, tobacco, faulty home heaters, car exhaust, and industrial pollution. Infants should sleep in their own crib with a firm mattress and no pillow or excessive blankets. Saperstein, MD, Director, Pediatric Dermatology, Cedars-Sinai Medical Center, Los Angeles, California, Clinical Associate Professor of Pediatrics and Medicine, David Geffen School of Medicine at UCLA, Los Angeles, California 17 Frank T. The neurologic examination is abnormal during the headache. Tumors increase in frequency during puberty. Table 17-1 summarizes management approaches based on skeletal maturity and the degree of curvature. b. Observation of parent–child interaction 5. Increased risk for malignant peripheral nerve sheath tumors (PNETs), optic 176 nerve gliomas, and central nervous system (CNS) gliomas 3. Risk factors for nonorganic CAP. Timing of vaccination 1. However, the UGI study is a poor test for diagnosis of GERD, even if the refluxate is seen rising up to the oropharynx. Omphalocele and gastroschisis are the most common defects, and both require surgical treatment. Treatment is supportive. Gastroenterology Nephrology and Urology Neurology Hematology Oncology Allergy and Immunology Rheumatology Orthopedics Ophthalmology Dermatology Emergency Medicine Download Brs pediatrics pdf free: You can download Boards Reviews Series Pediatrics pdf free from external source here. Elevated bilirubin may be a consequence of increased heme load (e.g., hemolysis, polycythemia, hematoma), decreased capacity for excretion (e.g., hepatitis, liver failure), or obstruction to bile flow (e.g., biliary atresia, choledochal cyst). It is often referred to as the "olive." 2. Individual and family counseling may be helpful. immunity to pertussis over time. Developmental surveillance is gathered through age-specific questioning, developmental questionnaires, observations during the visit, screening tests, and a review of academic school performance. Hypoventilation suggests opiate or sedative overdose. Alkalinization of urine with sodium bicarbonate to a urine pH > 7 and largevolume intravenous fluids enhance renal excretion of salicylates. For SMA types 2 and 3, survival until adolescent and adult years, respectively, is common. Henoch–Schönlein purpura nephritis 445 8. Blood loss from the GI tract may occur in several ways, and the presentation may help determine origin of the bleeding. TABLE 16-6 The Jones Criteria for Diagnosis of Rheumatic Fever* Major criteria Migratory polyarthritis Carditis Sydenham chorea Erythema marginatum Subcutaneous nodules Minor criteria Fever Arthralgia Previous rheumatic fever Leukocytosis Elevated erythrocyte sedimentation rate 596 Elevated C-reactive protein Prolonged PR interval on electrocardiogram * The diagnosis requires evidence of previous streptococcal infection and either (1) two major criteria or (2) one major plus two minor criteria. Cri du chat syndrome, caused by a partial deletion on the short arm of chromosome 5, is characterized by slow growth, microcephaly, mental retardation, hypertelorism, and a classic catlike cry. Foreign body aspiration is unlikely because of the absence of a history of choking. Familial (or genetic) short stature is defined as a height at least 2 SDs below the mean but within 2 SD of MPH with a normal bone age, a normal onset of puberty, and a minimum growth velocity of 2 inches (or 5 cm) per year. The drug is designed to increase the amount of functional survival motor neuron protein that is deficient in patients with SMA. Associated malformations are found in 50% of patients with esophageal atresia, including vertebral, anorectal, cardiac, renal, and limb anomalies (VACTERL association) (see Chapter 5, section IV.I.2). Acute Kidney Injury XI. With proper management, patients may be able to wean off parenteral nutrition and survive. It is likely that this infant may later have SIDS. Vitamin D supplementation is recommended for both breastfed and formula-fed young infants. The timing of adrenarche is not linked to activation of the hypothalamic–gonadal axis. The teratoma is suggested by the presence of calcium within the tumor on an abdominal radiograph. Advantages include ease of administration and induction of both host immunity and secondary immunity, because it is excreted in the stool of the recipient and may infect, and thus immunize, close contacts. However, with current treatment, the survival rate is now 85–100% at 5–10 years after initial diagnosis. Hereditary fructose intolerance is an autosomal recessive disorder caused by aldolase B deficiency that affects the ability to digest fructose. Complications of polycythemia include thrombosis (vaso-occlusive crisis, stroke, myocardial infarction) and bleeding. g. Tissue fragility and poor wound healing results in widened atrophic scars. The parents state that their son has become increasingly irritable, and they deny fever, vomiting, or other symptoms. Encephalopathy as a result of infection, drug overdose, or recent head trauma may cause decreased levels of consciousness, which may affect gait. Thickening of the circular pylorus muscle results in gastric outlet obstruction with projectile vomiting. influenzae polysaccharide linked to various protein antigens, including diphtheria or tetanus toxoids, to augment immunogenicity. These patients are at risk for significant deterioration with respiratory syncytial virus infections during their first year of life, and would therefore benefit from palivizumab (RSV monoclonal antibody) prophylaxis. Viral infection early in pregnancy (e.g., cytomegalovirus, rubella) 2. Outlook is very good; the seizures usually resolve by adolescence without cognitive impairment. Anion gap $[Na^+ - (Cl^- + HCO_3^-)]$ should be calculated. Type II: qualitative abnormality in vWf 3. There is also a comprehensive 150-question examination at the end of the text to provide the reader with a self-assessment tool and additional review of the primary care and subspecialty areas covered in this text. Binocular vision requires the integration of sufficiently clear and equal monocular retinal images from both eyes into a single, three-dimensional perception (binocular, sensory fusion). Antibodies to the adrenal gland may be detected (i.e., 21-OH antibodies), and there may be other associated endocrinopathies, including Hashimoto thyroiditis and type 1 diabetes mellitus (DM) (type I polyglandular syndrome), or hypoparathyroidism and chronic mucocutaneous candidiasis (type II polyglandular syndrome). It may occur as an acute hemolytic disease, induced by infection or medications, or as a chronic hemolytic disease. 380 2. It also functions as a carrier protein for factor VIII. Hematochezia is bright red blood passed per rectum, usually indicating a lower GI source or a significant rapid bleed from an upper lesion. Communicating (nonobstructive) hydrocephalus is due to either an increase in production or a decrease in absorption of CSF. Dysphagia f. Guillain–Barré Syndrome (acute inflammatory demyelinating polyneuropathy) 1. Degeneration of anterior horn cells in the spinal cord and motor nuclei in the lower brainstem. Hereditary Renal Diseases A. 599 VIII. Vitamin D supplementation is essential for bone health in the pediatric population, especially to prevent conditions such as rickets in infants and toddlers. *Granulomatosis with polyangiitis (formerly known as Wegener granulomatosis). Breath sounds are decreased, and bowel sounds may be heard in the chest. Treatment includes splenectomy for patients with severe chronic hemolysis. Discolored urine may not be from blood in the urine. Acute Abdominal Pain VI. Unilateral sixth nerve palsy d. Precautions (i.e., caution should be exercised) to immunization include the following: a. Clinical features of absence seizures 1. By 9 weeks' gestation, the SRY gene differentiates the gonads into fetal testes, which subsequently produce testosterone and anti-Müllerian hormone (AMH or previously called Mullerian inhibiting substance). Idiopathic pancreatitis accounts for up to 25% of cases in children, and is the second most common cause of pancreatitis during childhood. Marked itching and watery discharge are present. Seizures typically remit spontaneously during adolescence. If this patient is isonatremic or hypernatremic, her fluid deficits should be replaced over 24 hours, but if she is hyponatremic, her fluid deficits should be replaced over 48 hours. Instead, it's at the end of a written section on a totally different part of the chapter. Equal visual stimulation of each retina with clearly focused images 4. Plasma amino acids are normal. Desmopressin acetate (DDAVP) causes the release of stored factor VIII from the patient's own cells and may be useful in mild hemophilia A. Long-acting $\beta_2$agonists should not be used as quick relievers. Treatment is supportive, with nasogastric feeding and assisted ventilation as needed. Systemic lupus erythematosus and juvenile idiopathic arthritis can cause hip pain, but they are not associated with bilateral SCFE; thus, neither antinuclear antibody nor rheumatoid factor would be helpful. Phenylketonuria B. Occult bleeding from the GI tract is confirmed by positive guaiac testing of stool. Antibiotic treatment of the E. Table 6-1 Using Bone Age in the Differential Diagnosis of Short Stature Bone Age = Chronologic Age Familial short stature Intrauterine growth retardation Turner syndrome Skeletal dysplasia Bone Age < Chronologic Age Constitutional short stature Hypothyroidism Hypercortisolism Growth hormone deficiency Chronic diseases 206 II. The diagnosis is strongly suggested in the presence of epidemiologic risk factors and a classic erythema migrans rash. Vasculitis can also cause lower GI bleeding. It almost always occurs in children younger than 3 years of age. Brown, MD, FAAP Professor and Vice Chair of Education and Integrated Care Palo Alto Medical Foundation Palo Alto, California Adjunct Clinical Associate Professor of Pediatrics Stanford School of Medicine Stanford, California Ryan J. Symptoms start in newborns after ingesting lactose in breast milk or milkbased formulas. Depression and Suicide IV. Laboratory assessment of liver injury and function 1. Family dynamics that include single parenthood, unemployment, poverty, marital conflicts, domestic violence, poor parent–child relationships, and unrealistic expectations of the child 2. Chest physical therapy provides little added benefit. History, which often establishes the cause 1. Symptoms of esophagitis, including midepigastric pain ("heartburn") that is temporarily relieved with food or antacids and is exacerbated by fatty foods, caffeine, and being in the supine position. This is more common in boys and may be associated with neurologic complications such as optic nerve atrophy. Congenital myotonic dystrophy C. Child protective services or social services must be notified and should arrange follow-up and support. Elevated free cortisol in 24-hour urine collection. A 6-month-old girl with bilateral abdominal masses and severe hypertension, whose older sister died as a neonate after being diagnosed with oligohydramnios. If the copyright owner wants to remove this, kindly contact the external source owner to remove link. Hearing loss is a common complication of this condition. Supportive care E. In severe constipation liquid stool may leak around a hard, retained stool mass and is involuntarily released through the distended anorectal canal. The femoral head is not placed back into a normal position with the femoral neck because the force required may cause avascular necrosis. Long-term management includes continuous antimicrobial prophylaxis to prevent recurrent episodes of rheumatic fever. Meckel diverticulum is an outpouching of the bowel in the terminal ileum that occurs in 2% of infants. Early in the disease, patients may present with fever, headache, myalgias, arthralgias, and lymphadenopathy, along with an annular, or "targetlike," skin eruption with central clearing. Infants with urinary tract infections (UTIs) have an increased risk of having underlying structural abnormalities, including vesicoureteral reflux. Alport syndrome 1. At the same time, alignment is assessed by evaluating for the symmetry of light reflecting off both corneas (Hirschberg test). Autosomal dominant B. In juvenile myasthenia gravis, several findings are characteristic. Approximately 5% of school children have microscopic hematuria detected on a single voided urine sample, but only 0.5–2% have persistent microscopic hematuria. Bernstein 609 I. Platelet abnormalities 1. Shiga toxin–associated HUS occurs as a result of intestinal infection with a toxin-producing bacterial strain, most commonly Escherichia coli 0157:H7. Motor tics can be simple (e.g., eye blinking, head or shoulder shaking) or complex (e.g., bouncing, jumping, kicking). Carbon monoxide (CO) is a by-product of incomplete combustion of carbon-containing material. Table 18-3 Distinguishing Clinical Features of Conjunctivitis Clinical Feature Discharge Itching Bacterial Purulent Minimal Viral Watery Minimal Allergic Watery or mucoid Severe Preauricular lymphadenopathy Laboratory findings Absent Common Absent Blepharitis Minimal Minimal (irritation rather than itching) Absent Bacteria and PMNs on Gram stain No bacteria on Gram stain Eosinophils on conjunctval scraping Positive culture for Staphylococcus aureus PMNs = polymorphonuclear neutrophils. Evaluation 366 a. Free Download BRS Pediatrics (Board Review Series) 2nd Edition in PDF format BRS Pediatrics (Board Review Series) 2nd Edition PDF Free Download Download BRS Pediatrics (Board Review Series) 2nd Edition 2019 PDF Free BRS Pediatrics (Board Review Series) 2nd Edition 2019 PDF Free Download Download BRS Pediatrics (Board Review Series) 2nd Edition PDF The BMJ Best Practice's database is designed to provide clinicians with quickly accessible patient consult information. A deletion on chromosome 7 causes Williams syndrome, characterized by elfin facies and a loquacious personality. Patients with Fanconi syndrome may present with proximal (type II) RTA with glucosuria, aminoaciduria, and hyperphosphaturia (therefore low serum phosphorus levels, not high serum phosphorus levels). Which of the following statements regarding the acute management of this patient is correct? Prolactinoma f. Table 10-1 Clinical Features of Selected Vitamin and Mineral Deficiencies Nutrient Vitamin A Vitamin D Vitamin E Vitamin K Vitamin B1 Signs and Symptoms Night blindness, xerophthalmia (dry conjunctiva and cornea) Rickets/osteomalacia, dental caries, hypocalcemia, hypophosphatemia Anemia/hemolysis, neurologic deficits, altered prostaglandin synthesis Coagulopathy/prolonged prothrombin time, abnormal bone matrix synthesis Beriberi (cardiac failure, peripheral neuropathy, hoarseness or aphonia, Wernicke encephalopathy) (thiamine) Vitamin B6 Dermatitis, cheilosis, glossitis, microcytic anemia, peripheral neuritis (pyridoxine) Vitamin B12 Megaloblastic anemia, demyelination, methylmalonic acidemia 363 (cobalamin) Vitamin C Folic acid Niacin Zinc Scurvy (hematologic abnormalities, edema, spongy swelling of gums, poor wound healing, impaired collagen synthesis) Megaloblastic anemia, neutropenia, impaired growth, diarrhea Pellagra (diarrhea, dermatitis, dementia), glossitis, stomatitis Skin lesions, poor wound healing, immune dysfunction, diarrhea, growth failure 364 II. Myasthenia Gravis Review Test Answers and Explanations Chapter 13 Hematology I. Fractures that are inconsistent with the history or with the child's developmental ability may be secondary to abuse (see Chapter 17, section V.G). Meconium ileus is obstruction of the distal ileum caused by inspissated (thickened and dried) meconium, secondary to deficiency of pancreatic enzymes. Acidic refluxate induces bronchopulmonary constriction and can also lead to frank aspiration or microaspiration. Newer occult blood testing uses more specific and sensitive immune assays. Microcytic, hypochromic anemia (small, pale RBCs; low MCV) b. Leukemic cells that are CALLA-negative B. Clinical findings result from mandibular hypoplasia, which leads to a cascade of other features. The hypertrophied pylorus muscle may be palpable just above and to the right of the umbilicus. R—retarded growth and development 5. Although there is some variability among states as to which conditions are screened, all states screen for congenital hypothyroidism, phenylketonuria (PKU), and galactosemia. The interview is shaped by family and patient concerns and by age-specific trigger questions about common problems (e.g., sleep, nutrition, behavior). Affected children are born with a normal head circumference that does not grow after the cerebral insult. A 6-year-old girl presents with a 3-day history of bloody diarrhea and dehydration. Racemic epinephrine is not effective in epiglottitis. Two weeks ago, he had a sore throat and fever. Indications for tuberculosis testing include birth in a high-risk area outside of the United States, travel for >1 week (cumulative) in a high-risk area outside of the United States, history of exposure to someone with active tuberculosis, or close contact with anyone with a positive skin test for tuberculosis (TB). Nephronophthisis. Corticosteroid therapy is the treatment of choice for symptomatic croup. Even with treatment, patients may demonstrate speech defects and learning disabilities. Perkins, MD, PhD, FAAP, Associate Dean for Graduate Medical Education, Professor of Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Leigh Christopher Reardon, MD, FACC, Director, Pediatric Mechanical Circulatory Support and Transitional Cardiac Care, Assistant Professor of Medicine and Pediatrics, David Geffen School of Medicine at UCLA, Los Angeles, California Srinath Sanda, MD, Assistant Adjunct Professor of Pediatrics, Division of Endocrinology, Department of Pediatrics, UCSF School of Medicine, San Francisco, California Monica Sifuentes, MD, Professor of Pediatrics, David Geffen School of Medicine at UCLA, Associate Residency Program 15 Director, Pediatric Clerkship Director, Department of Pediatrics, Harbor-UCLA Medical Center, Torrance, California Grant D. Causes include infection with H. UGI study may demonstrate an elongated, narrow pyloric channel ("string sign"). Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) is the most common disorder. There are reports of associated learning disabilities associated with BECTS. Many metabolic diseases can be diagnosed and treated in the newborn period. Loss of SMN1 protein is partially compensated by SMN2 protein synthesis. Perianal disease often precedes the development of intestinal disease and presents with skin tags, fissures, fistulas, and abscesses. Timing of vaccine: All children aged 11–12 years should receive the quadrivalent vaccine to protect against serotypes A, C, W, and Y. Microcephaly should be apparent. Sore throat, conjunctivitis, and red cracked lips are not associated with constipation, arthritis, or intussusception. The major Jones criteria include erythema marginatum, carditis, migratory polyarthritis, subcutaneous nodules, and Sydenham chorea. Limp, refusal to walk, and pain with movement of the joint may occur. One quarter– or one half–normal saline is not an appropriate intravenous fluid for the emergency phase. Congestive Heart Failure (CHF) II. Frequent fractures may result from muscle weakness and osteopenia secondary to steroid therapy. Some patients note that stress, fatigue, menstruation, dehydration, skipping meals, or exercise induces the headache. Abdominal wall defects. On examination, she has a webbed neck with a normal range of motion, a shield chest with widely spaced nipples, and scoliosis. Treatment includes observation and range-of-motion physical therapy to prevent contractures. Vascular Skin Lesions Review Test Answers and Explanations Chapter 20 Emergency Medicine I. Serum porphobilinogen, while not addressed given the scope of this text, is elevated in acute intermittent porphyria, which may present with weakness, abdominal pain and autonomic instabilty. Head CT scan should be

performed to identify mass lesions or trauma. General Approach to the Child With Possible Infection II. Asymmetric abduction of the hips and asymmetric thigh or buttock folds. Enteral feeds with a soy-based formula should be given. Fragile X syndrome A 4-month-old male infant has been brought to your office for a routine health maintenance evaluation. Infant jaundice (Neonatal jaundice is discussed in Chapter 4, section X.) 1. Constipation may be associated with encopresis. Cyanosis associated with congenital heart disease is due to right-to-left shunting of deoxygenated blood. Upper Respiratory Infections VI. Although hepatocytes are primarily infected, hepatobiliary obstruction results from local swelling. Imaging studies. Skeletal abnormalities, which include short stature in almost all patients, and absence or hypoplasia of the thumb and radius 3. 478 2. Autoimmune factors a. Watch for Somogyi phenomenon. Tonic–clonic seizures are the most common type of generalized seizure. The cause is unknown. A 5-year-old girl with arthritis, abdominal pain, and a petechial eruption on her buttocks. Copious wound irrigation b. Marfan syndrome a. Drowning is the process of experiencing respiratory impairment from submersion/immersion of liquid. The following three disorders have high carrier frequency in the Ashkenazi Jewish population. Although the patient's condition is likely to be an inborn error of metabolism (maple syrup urine disease, given the diaper odor consistent with the odor of maple syrup), sepsis is more common and can present in a similar fashion. Imaging complements physical examination and laboratory findings. Later, restrictive lung disease patterns are present. SMA type 1 is also called infantile-onset SMA or Werdnig–Hoffman disease. Tobacco smoke exposure is a known risk factor for sudden infant death syndrome (SIDS). Cyanosis V. It is also indicated immunocompetent children with chronic heart disease, chronic lung disease, diabetes mellitus, cerebrospinal fluid (CSF) leak or cochlear implants. Absent or markedly decreased GALT activity in RBCs c. A complex febrile seizure is distinguished from a simple febrile seizure if it lasts more than 15 minutes, has focal features, or recurs within 24 hours. Anti-Smith (anti-Sm) antibodies are less prevalent in patients with SLE but, when present, are very specific. More than 200 different mutations or deletions have been identified in the factor VIII gene. Outcome is good for local disease, with a 3- to 5-year survival rate of 80%. Celiac disease often presents between 6 months and 2 years of age when gluten, found in wheat, rye, barley, and oats (if the oats are harvested in fields that also contain wheat), is introduced into the diet. Contrast enema may show the classic "coil spring" sign representing the intussusception. Bernstein, MD, Medical Director, The Cedars-Sinai Orthopaedic Center, Executive Vice Chair, Department of Orthopaedics, Cedars-Sinai Medical Center, Professor of Orthopaedic Surgery, Los Angeles, California Carol Diamond, MD, Director of Pediatric Hematology/Coagulation, Associate Professor of Pediatrics, University of Wisconsin School of Medicine and Public Health, Madison, Wisconsin David A. Between 6 months and 8 years of age, first-time vaccine recipients need two doses of vaccine, spaced at least one month apart. If psoriasis is absent, the diagnosis can also be established if there is a first- degree relative with psoriasis. Normal ratios 1. However, ANA is nonspecific for SLE and is found in many other diseases. Definition. Some of the more common pulmonary causes of respiratory distress in the neonate include respiratory distress syndrome (RDS; also termed hyaline membrane disease and surfactant deficiency syndrome) in preterm infants, transient tachypnea of the newborn (TTN) from delayed clearance of fetal lung fluid in term infants during the first day of life, and MAS with persistent pulmonary hypertension of the newborn (PPHN) in full-term infants. Vitamin K, a fat-soluble vitamin, is essential for the synthesis of both procoagulant and anticoagulant factors, such as factors II, VII, IX, and X and proteins C and S. Migraines can be a lifelong disorder with a waxing and waning course. Respiratory problems are among the most common and significant causes of morbidity and mortality during the neonatal period. Most patients present with hyperglycemia documented by a random blood sugar above 200 mg/dL with polyuria, polydipsia, weight loss, or nocturia. 186 VII. Management is supportive. A 10-year-old girl is being evaluated for "weakness" of 3 days' duration. Physical examination may initially be normal with peritonitis occurring later. Stools may be normal or have a bloody, "currant jelly" appearance because of intestinal ischemia and mucosal sloughing. Nasolacrimal massage may help open the distal obstruction. Causes include decreased folic acid intake (i.e., from a diet lacking uncooked fresh fruits and vegetables or from exclusive feedings with goat's milk as the sole source of milk protein) and decreased intestinal absorption of folic acid (i.e., from diseases affecting the small intestine, such as celiac disease, chronic infectious enteritis, Crohn disease, or medications, such as anticonvulsants and oral contraceptives). Past medical history is significant for repaired coarctation of the aorta at 3 years of age. Digestion is an intraluminal event requiring digestive enzymes and bile acids for micelle formation. The answer is C [, section XIV.C]. Prognosis is better in children than in adults. When you greet the baby's mother, she is anxious and has difficulty releasing her grip after shaking your hand. Bilious vomiting and sudden onset of abdominal pain in an otherwise healthy infant is the classic presentation. Transient synovitis (also known as toxic synovitis) is a common self-limited, irritation of 619 the hip joint. Upper intestinal contrast imaging (UGI study with small bowel follow-through) is the diagnostic tool of choice, and demonstrates the abnormal position of the ligament of Treitz to the right of midline, partial or complete duodenal obstruction, and the jejunum to the right of midline. Classic physical examination findings, although uncommon, include cherry red skin (venous blood carries more oxygen than normal as a result of impaired release of oxygen to tissues) and retinal hemorrhages. Physical examination is normal, revealing no focal neurologic signs. Drugs (e.g., oral contraceptives, cyclosporine, tacrolimus) or pregnancy may cause aHUS. There is a bimodal age distribution in childhood. Pneumonia in immunocompromised patients may be caused by Pneumocystis jiroveci. Infantile botulism is caused by the ingestion of Clostridium botulinum spores and the release of botulinum toxin within the intestine. The answers are B, A, and D, respectively [VIII.D, VIII.E, and VIII.B]. Broad-spectrum antibiotics should be used for the treatment of pulmonary exacerbations. Antipyretic treatment during subsequent febrile illnesses is unlikely to prevent febrile seizures. Functional constipation. Patients have moderate to severe anemia at birth with an elevated Hgb Bart's (which is a type of Hgb made up of four gamma-globins that bind oxygen very strongly and do not 493 release it to tissue). Once the infant is stabilized, management includes surgical reduction of the hernia and closure of the diaphragmatic defect. Classification a. The elbow is held flexed and no swelling is present. Family and individual counseling are usually necessary. Sensory examination is normal. Therefore, initial management should include intravenous antibiotics. Pneumococcal HUS 1. Renal ultrasound often shows enlarged kidneys. FAS is caused by maternal alcohol consumption during pregnancy. Timing of vaccination: One dose should be given at 2 years of age and at least 8 weeks after the final dose of PCV13 (see below). Prolonged vomiting of gastric contents classically results in hypochloremic, hypokalemic, metabolic alkalosis. Patients with positive dipsticks for blood should have microscopic evaluations of fresh urine specimens. The mainstay of treatment is supportive care, including gastrostomy tube feeding to ensure adequate nutrition, overnight noninvasive ventilation to prevent sleep apnea, diligent surveillance and treatment 454 of respiratory infections, and physical therapy to maintain range of motion and prevent contractures. Aspergillus species is one of the major pathogens associated with CGD. Anaphylaxis II. The most concerning cardiac pathologies associated with this presentation include long QT syndrome and hypertrophic cardiomyopathy. More severe disease is seen in older children and adults, although it is rarely associated with fulminant hepatitis. 86. Bagged specimens obtained from an infant are inappropriate for culture as they are very likely to be contaminated. This is greater than the physiologic dose of hydrocortisone of 6–8 mg/m2 per day. TABLE 16-5 Diagnostic Criteria for Systemic Lupus Erythematosus* Mnemonic "SOAP BRAIN MD" Serositis (pleuritis or pericardial inflammation) Oral or nasal mucocutaneous ulcerations 591 Arthritis, nonerosive Photosensitivity Blood cytopenias (leukopenia, hemolytic anemia, or thrombocytopenia) Renal disease (hematuria, proteinuria, hypertension) ANA-positive Immunoserology abnormalities (antibodies to double-stranded DNA, Smith antigen, false-positive RPR or VDRL assays, or antiphospholipid antibodies) Neurologic symptoms (encephalopathy, seizures, or psychosis) Malar rash (butterfly rash) Discoid lupus (red raised patches with scaling, can cause scarring) * Four of eleven criteria provide a sensitivity and specificity of 96%. Meningocele is suggested by physical examination findings and is confirmed by MRI of the spinal cord and spine. Table 18-2 Etiology, Clinical Features, Diagnosis, and Management of Neonatal Conjunctivitis Etiology Chemical Watery discharge Neisseria gonorrhoeae Purulent discharge Eyelid swelling Can lead to corneal ulcer Chlamydia trachomatis Serous or purulent discharge Variable lid swelling Onset and Clinical Features Within first 24 hours Few PMNs 2–4 days of life Conjunctival Studies Negative Gram stain Management No treatment necessary Gram-negative intracellular diplococci Intravenous cefotaxime and topical erythromycin Positive gonococcal culture Treat parents 4–10 days of life Cytoplasmic inclusion bodiesPositive DFA or culture Treat parents 646 Oral erythromycin Herpes simplex virus Positive HSV culture 6 days–2 weeks of lifeUsually unilateralSerous discharge Multinucleated giant cells on Gram stain Intravenous acyclovir and topical trifluorothymidine PMNs = polymorphonuclear neutrophils; DFA = direct fluorescent antibody assay; HSV = herpes simplex virus. Etiology (Table 12-1). The publisher does not provide medical advice or guidance, and this work is merely a reference tool. Varicella often causes uncomplicated illness, but may cause severe disease in very young and in older or immunosuppressed patients. Identification of the abrasion on fluorescein staining of the cornea is the basis of diagnosis. Thalassemia is a group of inherited anemias characterized by defective synthesis of one of the Hgb chains. Major disadvantage is that the vaccine has little immunogenicity in children younger than 2 years. Topical antibiotics are administered if infection is present. His maintenance sodium requirement is approximately 1 mEq/kg/day. 178 FIGURE 5.2 Clinical features of Down syndrome. Febrile seizures can be inherited, and several gene mutations have been found. Identifiers: LCCN 2018014018 | ISBN 9781496309754 (paperback) Subjects: LCSH: Pediatrics—Examinations, questions, etc. The age of onset is bimodal, with a peak at 15–20 years of age and a second peak after 50 years of age. Immunizations b. Exocrine pancreatic insufficiency 1. L. Nonfatal drowning refers to a person who is rescued at any time, and the process of drowning is interrupted. Targeted screening for hypercholesterolemia (consisting of fasting lipid profile performed twice within 2–12 weeks) is recommended for children between 2–8 years of age and 12–16 years of age with one of the following risk factors: a. Testicular enlargement is the first sign of puberty in boys. Genitourinary neurologic deficits 461 F. Determining the cause of the child's chronic kidney disease may have implications for the child and his or her family, especially important in cases of inherited disorders. Serologic testing involves the measurement of antibodies to B. It can occur in the small or large bowel. Hypercalciuria E. 199 CHAPTER 6 200 Endocrinology Srinath Sanda 201 I. Patients with X-linked hydrocephalus frequently have cognitive delays. Moderate dehydration (7–10%) is associated with mild tachycardia with a weak pulse, flat fontanelle, deep set eyes, reduced tears, dry mucous membranes, skin tenting, and capillary refill of about of 2 seconds, in a patient with decreased urine output. Tumors in this location present with a painless growing mass. Acute management includes eradication of streptococcal infection with penicillin and control of inflammation with nonsteroidal anti-inflammatory agents or corticosteroids. Weakness. Anemia is treated with iron and recombinant erythropoietin therapy. Vision screening principles can be remembered using the acronym I-ARM PLUS (Inspection, Acuity assessment, Red reflex testing, Motility assessment). Studies show an elevated reticulocyte count, hyperbilirubinemia, spherocytes on blood smear, increased MCHC (mean corpuscular hemoglobin concentration), and abnormal RBC fragility with osmotic fragility studies. Duration. Pulmonary valve stenosis is the most common cardiac defect. Males are more commonly affected. Burning, crusting, and scales at the eyelash base; thickened and hyperemic eyelid margins; broken or absent eyelashes; and a history of awakening in the morning with eyelashes stuck together are characteristic. Injury is triggered by a cell-mediated immune response to an infectious agent that cross-reacts to antigens on the Schwann cell membrane. See Chapter 1, sections III.C.1 and III.C.8 for discussions of these vaccinations. Normocytic, normochromic anemias. The final appearance of gonadal tissue is dependent on both genetic and hormonal influences. Patients with secretory diarrhea still maintain their ability to absorb fluid and electrolytes through an intact, coupled cotransport mechanism. Growth is normal without advancement of bone age. Note that in Marfan syndrome, the lens more often dislocates superiorly, whereas in homocystinuria, the lens more often dislocates inferiorly. The answer is C [, section XI.C.2 and Table 7-6]. Growth velocity is ideally calculated over at least a 6-month period. Muscle biopsy shows a characteristic atrophy of groups of muscle fibers innervated by the damaged axons. This vignette describes a very worrisome episode of syncope that occurred when the patient was exercising. Thymectomy is performed if there is evidence of thymoma (there is also some evidence for thymectomy even in the absence of thymoma) H. Treatment includes rest and elimination of known triggers. Growth Abnormalities IV. If behavioral strategies are ineffective and moderate to severe impairments are seen in the child's functioning at home and preschool/child care and with peer interaction, then treatment with a stimulant medication is suggested. Physical examination reveals hypertension and a right-sided flank mass. Ehlers–Danlos syndrome Cri du chat syndrome Osteogenesis imperfecta Williams syndrome Angelman syndrome Hurler syndrome Hunter syndrome Tay–Sachs disease Gaucher disease Homocystinuria For each patient, select the most likely genetic condition. Laryngeal tumors typically present with hoarseness. Back pain. The cutaneous hallmark is skin thickening with loss of dermal ridges, resembling "tightened" skin. Maple syrup urine disease D. The answer is B [, sections IV.E.2.b and IV.E.6.c]. Juvenile ankylosing spondylitis C. Polycythemias A. Maternal supplementation with vitamin D will not prevent vitamin D deficiency in infants, as breast milk contains low levels of vitamin D. Motility assessment and eye alignment. Iron is one of the most common and potentially fatal childhood poisonings. Because MMR and varicella vaccines are live attenuated vaccines, fever and rash may occur 1–2 weeks after immunization (i.e., after the incubation period of the virus). Depending on disease severity and extent, treatment ranges from low-dose oral to high-dose intravenously pulsed steroids. Pancreatitis is an acute inflammatory process of the pancreas that may involve peripancreatic tissues and other organ systems. Specific Sensory Impairments IV. No breast tissue or testicular enlargement is noted, and there is no clitoromegaly. Physical examination should be comprehensive and may provide additional clues to the identity of the toxin. Sudden Infant Death Syndrome (SIDS) A. Immediate treatment of cyanosis may be necessary and often includes empiric administration of oxygen and correction of abnormalities of temperature, hematocrit, glucose, and calcium levels. A computed tomography scan is not indicated in the absence of papilledema or focal neurologic deficits. The answer is E [I.D, I.E]. Anti-DNase antibodies may also be used to document GABHS infection. The goal of the first phase of parenteral rehydration (emergency phase) is to restore or maintain the intravascular volume to ensure perfusion of vital organs, and this phase is the same for all patients (regardless of the patient's initial serum sodium level). There is no true hernia sac (no peritoneal sac covering), and the bowel is usually the only viscera that herniates. The answer is C [III.D.2.c]. 81. History of violent temperament 5. 546 Review Test 1. Spontaneous epistaxis that is severe and recurrent without an obvious cause c. Low self-esteem, social isolation, depression, or history of substance abuse 2. A 6-year-old boy with erythematous and hypertrophic papules over the metacarpal and proximal interphalangeal joints and "dark rings" around the eyes. Prognosis is excellent. Autosomal recessive inheritance 2. She should not be given antibiotics, as this may increase the risk of HUS. The primary symptoms are diarrhea, vomiting, constipation, bloating, anorexia, and sometimes FTT. Adolescent Growth and Development II. On physical examination you note a temperature of 98.8°F (37.1°C), a respiratory rate of 60 breaths/minute, and bilateral conjunctival erythema. Puberty begins when there is a reduction in this hypothalamic inhibition, resulting in activation of the HPGA. Skin may be described as soft and/or velvety in texture, with bruising caused by fragile blood vessels. Treatment is based on the underlying cause. Spontaneous movement of the arm confirms successful reduction. Management often includes an enema that may relieve the obstruction, and surgery for persistent obstruction and resection of injured bowel in cases with perforation. Vascular endothelial injury by the Shiga-like toxin is the key to the pathogenesis of injury in STEC-HUS. The toxin binds to endothelial cells, causing endothelial cell injury, most especially in the renal vasculature. Age-specific health supervision according to recommended periodicity schedules 2. If children develop the disease when they are younger than 9 years, complete resolution within 3 years is common. Rubella causes a mild viral syndrome in children, but may cause severe birth defects (commonly hearing loss and heart defects) in offspring of susceptible women who are infected during pregnancy. Hematemesis is the vomiting of fresh or old blood, which may have a "coffee ground" appearance from the denaturing of hemoglobin. Noonan syndrome B. This patient is likely to have elevated antistreptolysin O or anti-DNase B titers. Because of the inactivity of the hypothalamus and pituitary gland, these patients have a low FSH, low LH, and, in turn, low testosterone and low estradiol, with a prepubertal (flat) GnRH stimulation test. Patients with Addison disease can present with unusual hyperpigmentation due to simultaneous stimulation of melanocytes from elevated ACTH levels, resulting from lack of cortisol feedback at the level of the pituitary. History of prior steroid use or autoimmune disorders should raise clinical suspicion. PT and platelet count are normal. Sudden Infant Death Syndrome (SIDS) VIII. Delayed puberty 1. Acanthosis nigricans (velvety and hyperpigmented skin of the neck and axillary folds) is common. Neonates with UTIs are admitted to the hospital for initial IV management, which commonly includes ampicillin and gentamicin. liver, spleen and lungs. Viral conjunctivitis 1. Although steroids have been used in treatment of refractory Kawasaki disease, they have no role in primary therapy. Girls present with virilization (mainly varying degrees of clitoromegaly), and at 1–2 weeks of life both boys and girls present with failure to thrive, vomiting, and electrolyte abnormalities. Major advantage is that the vaccine contains antigens from pneumococcal strains causing almost all cases of bacteremia and meningitis during childhood. In children, oliguria is defined as a urine output < 1 mL/kg/hour. A viral prodrome (upper respiratory infection or diarrheal illness) often precedes transient synovitis. Severe, moderate, and mild forms exist based on the activity level of factor 506 VIII protein. The answer is C [, section VIII.B.1 and Chapter 1, section III.C.6]. Vomiting becomes more forceful, is often described as projectile, and typically occurs immediately after feeding. However, surgical intervention for early, acute pancreatitis is controversial. Basilar artery migraine. This is typically a self-limited illness and does not required specific treatment. Outflow of aqueous humor is reduced because of maldevelopment of the trabecular meshwork. There might be a family history of neonatal death. Microscopic hematuria is defined as ≥5–6 RBCs per high-power field (HPF) detected on two or more consecutive samples. The remaining one-third of patients present earlier, within the first 2 weeks of life, and therefore the presentation can be confused with physiologic jaundice. A 8-year-old girl with daily wheezing and nighttime symptoms two times per week. Marfanoid body habitus (tall, slender), scoliosis, and pes planus b. Placement of a protective shield or patch for 24–48 hours may be recommended in severe cases. Life expectancy is beyond the age of 30 years. Bilirubin is derived from the breakdown of heme. In addition, patients with homocystinuria usually have downward lens subluxation (upward lens subluxation is found in Marfan syndrome) and large, stiff joints (joint laxity is found in Marfan syndrome). Although newborn screening identifies the vast majority of cases, patients who are born in countries without newborn screening can present with untreated PKU. Scurvy is vitamin C deficiency and causes impaired collagen synthesis that results in weakened blood vessels. Polycythemias IV. Tension headaches are more commonly seen in children older than 7 years with a prevalence as high as 25%. Normal puberty (see also Chapter 3, section I.A.2) 1. Increased pressure in the third ventricle from noncommunicating hydrocephalus injures the upward gaze center in the midbrain, causing this downward deviation of the eyes. Developmental dysplasia of the hip (DDH) occurs when the acetabulum is shallow or poorly formed, leading to the subluxation or dislocation of the head of the femur. 434 XI. Treatment is based on the tumor type and size. Pleural effusions and hypotension may also occur. Her white blood cell (WBC) count is 25,000 cells/mm3. Clinical features overlap with those seen with Hurler syndrome, except that there is no corneal clouding ("a hunter needs to see clearly!). Classic clinical features include constitutional symptoms, severe sinusitis, hemoptysis, and glomerulonephritis. Consider some form of skeletal dysplasia (e.g., achondroplasia) in patients who are short with short limbs (see Chapter 5, section III.H). The toxin is released from the spores of Clostridium botulinum, which are found in contaminated honey or soil. McCune–Albright syndrome is characterized by fracture-prone bony changes (polyostotic fibrous dysplasia), skin findings (irregularly bordered hyperpigmented macules, or "coast of Maine" jagged-bordered café-au-lait spots), and endocrinopathies (PPP or hyperthyroidism). Neonatal hepatitis a. Diagnosis 1. However, a large, persistent pseudocyst may require surgical drainage. Normally, the major Hgb in RBCs is hemoglobin A1, a tetramer of two αchains and two β-chains. Patients are unable to metabolize galactose and are therefore unable to tolerate lactose. The hip is then gently abducted, and a "clunk" is felt as the hip relocates. The answer is C [, Tables 1-1 and 1-3]. Medullary sponge kidney. Note however that not all bacteria produce nitrites (e.g., enterococcus), which may lead to a negative nitrite test. Extrahepatic mechanical obstruction (e.g., biliary atresia, bile duct stricture) 392 4. Tibial torsion is distinguished from the other common causes of intoeing —metatarsus adductus and femoral anteversion—based on age and the thigh–foot angle with decreased external rotation. Disorders of Sexual Differentiation (DSD or Formerly Known as Ambiguous Genitalia) A. Which of the following chromosomal abnormalities is the most likely cause of the patient's features? LH is responsible for the positive feedback in the middle of the menstrual cycle, resulting in the release of an egg. Older children with closed cranial sutures have the following symptoms and signs of increased ICP: a. Acute symptoms of hepatitis [see section XI F.1.c] occur in 150 mmol/L) 2. Back strain is muscular soreness from overuse or poor body mechanics. A few days later, she is brought to the emergency department with tachypnea, chest retractions, diffuse expiratory wheezing, and fine inspiratory crackles bilaterally. Analgesics (e.g., acetaminophen, nonsteroidal anti-inflammatory drugs, aspirin) f. The 8-year-old girl's asthma would be characterized as moderate persistent asthma because of the presence of daily symptoms and nighttime wheezing more often than once per week. The ataxia usually follows a viral illness by 2– 3 weeks. UDP-glucuronyl transferase deficiency can present in three distinct states. Meningocele is the herniation of the meninges only through a bony cleft, most commonly in the lumbosacral region. Malabsorption A. Specific tumors 1. This vasculitis is characterized by aneurysms and thrombosis of the small- and medium-sized vessels (e.g., brachial, femoral, or mesenteric arteries). CO is odorless, tasteless, and colorless. No vomiting, visual changes, or paresthesias occur. Autoimmune. Discovery of heavy proteinuria, hypoalbuminemia, and hypocholesterolemia on laboratory testing would confirm the diagnosis. Skeletal involvement occurs in 80% of patients. Peak incidence is 10–12 years of age. Ocular Examination and Vision Screening II. Ninety percent of lumbosacral myelomeningoceles are associated with Chiari type II malformation and hydrocephalus. Infantile spasms a. Infantile polycystic kidney disease (question 12) is an autosomal recessive disorder characterized by greatly enlarged cystic kidneys, severe hypertension, and variable degrees of liver involvement. 414 III. Lactate dehydrogenase (LDH) elevation is nonspecific for liver disease, although it may serve as a marker for hepatocellular necrosis. Defects of Carbohydrate Metabolism A. A cystic mass on transillumination of the scrotum is consistent with the diagnosis of a hydrocele and painless inguinal swelling with a diagnosis of inguinal hernia. Microcephaly, wide anterior fontanelle, hypertelorism, short nose, wide mouth 2. Proximal muscle weakness (mostly of the hip girdle and legs) is insidious in onset, is symmetric, and occurs weeks to months after the eruption of skin findings. Head shape in craniosynostosis is determined by which suture closes prematurely, because bone growth perpendicular to the affected suture is arrested. On urinalysis, RBCs that appear as biconcave disks indicate that they originated in the glomerulus, and this suggests that he has glomerulonephritis. No treatment is required. Aortic root dilatation D. Clinical features include micrognathia, glossoptosis (posterior displacement of the tongue), and cleft palate (often a U-shaped cleft). Surgical removal of the nails is a last resort for chronic nail infection over many years. Common signs and symptoms include epistaxis, menorrhagia, bruising, and bleeding after dental extraction or tonsillectomy. In the absence of the SRY gene, the gonads become ovaries. Infection, necrosis, and perforation may all occur. Low-grade fever, limp, and mild irritability may be present. Intestinal Anatomic Obstructions That Result in Vomiting V. Pupillary size and reactivity may provide clues. 5. Hospitalization may be required if medically indicated or until a safe location for the child has been identified. The term acute abdomen describes the sudden onset of abdominal pain that requires urgent evaluation, diagnosis, and treatment. Biliary atresia a. Poisonings IX. The prevalence increases from 3% in 7-year-olds, to 11% in 11-year-olds, and up to 20% in adolescents. Oral rehydration therapy has been shown to be a safe and inexpensive alternative to intravenous rehydration and effective even in the face of secretory diarrhea, such as would be seen in cholera. Matted eyelashes c. Evaluation (Figure 11-1). Because iron stores disappear first, an early finding of iron-deficiency anemia is low serum ferritin. Kawasaki Disease (KD) III. Disorders of Metal Metabolism A. Ocular Trauma A. Obtaining a nonfasting non-HDL cholesterol level is recommended for all children between 9 and 11 years of age, and again between 17 and 21 years of age. Pediatric Physical Therapy. Jaundice occurs very rarely. Her current weight is 8 kg, and her head circumference has increased by 12 cm since birth. Key point: If a patient is developmentally delayed and has a marfanoid habitus, perform screening tests for homocystinuria. Type 1 DM patients are at increased risk for autoimmune thyroid conditions and celiac disease. This 12-month-old patient is demonstrating inadequate weight gain. As blood settles, a blood-aqueous fluid level may be seen. Treatment may include transfusions that unfortunately may provide only transient benefit. Membranoproliferative nephritis, membranous nephropathy, and nephritis as a result of systemic lupus erythematosus during childhood are all less common causes of glomerulonephritis in children. The basic physiologic defect is a loss of the normal charge- and size-selective glomerular barrier to the filtration of plasma proteins. Unlike all other types and subtypes of juvenile idiopathic arthritis (JIA), juvenile ankylosing spondyloarthritis is male predominant and almost always presents in children older than 8 years. If possible, the specific cause should be addressed (e.g., removal of a nephrotoxic drug). Maternal and Child Nursing. Nerve conduction studies are performed to identify myopathies (especially congenital) neuromuscular junction disorders and some neuropathies. I've got the 2nd edition, and I love it. Causes most commonly include nontypeable Haemophilus 644 influenzae, Streptococcus pneumoniae, Moraxella catarrhalis, and Staphylococcus aureus. Characterized by blue sclera and frequent fractures after minimal or no trauma. Both fresh and salt water drowning result in denaturing of surfactant, alveolar instability and collapse, and pulmonary edema. Flexibility in the rounded area should be assessed by having the patient extend his or her 613 back while prone. Oral mineralocorticoids and dextrose-containing fluids should be given as well. Similar to the chapter review tests, the comprehensive examination includes detailed explanations for both the correct and incorrect choices. 354 Review Test 1. Aortic dissection E. Speech is also affected and can be jerky or indistinct. Patients are predisposed to thrombosis secondary to hypercoagulability. Inheritance is usually X-linked dominant, although autosomal dominant and autosomal recessive variants exist. Should be considered in the differential diagnosis of a profoundly hypotonic infant with failure to thrive, cardiomegaly, and macroglossia (large tongue). α-Fetoprotein (AFP), the main serum protein in fetal life, is elevated in amniotic fluid and maternal serum in open neural tube defects, and when measured in maternal serum at 16–18 weeks' gestation, detects 80% of spinal defects. Resuscitation should be attempted on all patients because of the difficulty in ascertaining the period of time the infant has been apneic and pulseless. Patients require either five positive IgG bands or two positive IgM bands. Physical examination findings in both central and peripheral hypotonia may include weak cry, poor suck, drooling, decreased spontaneous movements, delay of motor milestones, frog-leg positioning (hips are externally rotated and flexed), poor posture on horizontal and vertical suspension (infant slips through examiner's hands when lifted up under the axillae), and head lag when pulled to the sitting position. CNS abnormalities that may cause CPP include hydrocephalus, CNS infections, cerebral palsy, benign hypothalamic hamartomas, malignant tumors such as astrocytomas and gliomas, and severe head trauma. Empiric therapy should be initiated with dicloxacillin. IUGR = intrauterine growth restriction. Urinary continence and loss of posture are also not seen in absence seizures. Juvenile Dermatomyositis (JDM) VI. Watch for "honeymoon" period. Increasing head circumference that crosses percentile lines, or head circumference >97% for age 2. Of note, ABO disease can occur in the first pregnancy, unlike Rh hemolytic disease. In North America, the most common pathogen is Escherichia coli 0157:H7. Burns often have distinguishable patterns. Etiology. cerebellar dysfunction, 311 vs. Associated problems include wheelchair dependency, bladder or bowel incontinence, seizures, precocious puberty, pressure sores, and fractures. Neither homocystinuria nor cystinuria has a peculiar odor as a feature; however, patients with homocystinuria may have developmental delay as a result of strokes from their hypercoagulable state. Detection 1. Patients may present with ambiguous genitalia, such as varying degrees of hypospadias, or as adolescents with incomplete pubertal progression. FSH in boys stimulates the Sertoli cells in the seminiferous tubules of the testes to produce sperm. Abdominal pain and large, foul-smelling stools are also common. Adolescent medicine. The answer is C [V.L.4]. Documentation of low serum folic acid is diagnostic. Volvulus is a surgical emergency requiring immediate exploration, untwisting of the gut, resection of any nonviable segments of intestine, and fixation of the gut to prevent recurrence. Miller 14 Contributors Lloyd J. FSH, LH, and sex steroid levels are elevated. Fever is sometimes present. Clinical findings are variable and include abdominal pain, flank masses, urinary tract infection (UTI), gross or microscopic hematuria, hypertension, or 429 renal insufficiency. A 2-year-old toddler presents with a 3-day history of increasing inspiratory stridor, cough, and increased work of breathing. Chromosome analysis in girls to evaluate for Turner syndrome d. Adverse effects of this cytotoxic agent include infertility and gonadal failure, secondary malignancies, and hemorrhagic cystitis. Shwachman–Diamond syndrome. Fetal Evaluation and Prenatal Diagnosis IV. Obesity and Eating Disorders VI. It is our great hope that medical students on their core pediatric clerkships, senior students on pediatric subinternships and pediatric subspecialty rotations, and students or physicians studying for the USMLE Step 2 examination will gain a strong foundation in pediatrics, on which to build their subsequent learning in both primary care and subspecialty pediatrics. Safety of the child should be the highest priority in determining placement. Immunotherapy 1. Lack of fixation of the small bowel results in peritoneal bands (Ladd bands) that can compress the duodenum, causing mechanical obstruction. L—limb/radial defects CHARGE Syndrome a. Conjunctivitis and Red Eye Conjunctivitis is a nonspecific finding that refers to conjunctival inflammation. Laboratory evaluation should be tempered and based on the clinical symptoms and physical examination. One-fourth of cases occur in patients with Down syndrome. Empiric broad-spectrum IV antibiotic coverage should be initiated. Perinatal asphyxia C. Space-occupying lesions (e.g., cysts, tumors) 4. Using measurements collected over a shorter period of time may result in inaccurate growth velocity calculations. Rationale for vaccine: Varicella is the virus responsible for chicken pox and zoster. The femoral head is vulnerable to ischemic injury. 23. Prognosis after 6 months of diligent muscle stretching is complete resolution in 90%, with higher success rates when therapy is started earlier in life. Table 12-2 Causes of Acute Seizures During Childhood Head trauma Brain tumor Toxins Infections Vascular Metabolic disturbances Systemic diseases Cerebral contusion, subdural hematoma Astrocytoma Amphetamines, cocaine Meningitis, encephalitis, brain abscess, neurocysticercosis Cerebral infarction, intracranial hemorrhage Hypocalcemia, hypoglycemia, hypomagnesemia, hypo- or hypernatremia, pyridoxine deficiency Hypertension, hypoxic–ischemic injury, inherited metabolic disorder, liver disease, renal failure, neurocutaneous disorders (e.g., tuberous sclerosis, neurofibromatosis and Sturge-Weber syndrome) FIGURE 12.2 Classification of seizures. Fats are broken down into fatty acids and glycerol. The most commonly used agents are erythromycin or metoclopramide. They result from deficiencies in one of the enzymes involved in the urea cycle. Young children (25%, CO-Hb levels > 10% during pregnancy, history or presence of neurologic symptoms, or presence of metabolic acidosis or ECG changes. An human leukocyte antigen B27 (HLA-B27)–positive 14-year-old boy with abdominal pain and chronic diarrhea. A lateral radiograph is helpful to rule out kyphosis, spondylolisthesis, and congenital anomalies of the spine. Liver transplantation is indicated for liver failure and late clinical presentations. Outcome varies depending on the cause of the hydrocephalus and the success of the intervention. Specific management is based on the identified cause. The answer is B [, Table 8-1]. Diagnosis is on the basis of clinical features and on the following studies: 1. 612 b. She meets criteria with at least 5 days of fever, conjunctivitis, mucous membrane changes, rash, and a unilateral lymph node. Of the 20 amino acids, 9 are essential. Even treated patients may have long-term effects: 1. Short Stature II. Large numbers of RBCs (especially in the presence of dysuria) may indicate acute hemorrhagic cystitis, which may result from bacterial infections, viral infections (e.g., adenovirus), or chemotherapeutic agents (e.g., cyclophosphamide). 82. 184 e. Head ultrasound or computed tomography (CT) scan is performed to rule out hydrocephalus, if suggested by physical examination. Less common causes of acquired adrenal insufficiency are acute adrenal hemorrhage in the neonate and septicemia (especially associated with meningococcemia, known as Waterhouse–Friderichsen syndrome). If this process is disturbed, the result is an abdominal wall defect associated with a decrease in intraabdominal volume. An electroencephalogram is useful when the diagnosis of epilepsy is being considered but not in recurrent febrile seizures. Approach to the Comatose Patient A. These anemias are characterized by large RBCs with MCV > 95. More profound bleeding occurs in type III disease. Short stature is defined as height that is 2 standard deviations (SDs) below the mean height of the population (i.e., below the third percentile), or 2 SDs below the midparental height (MPH). Serum iron level should be obtained 2–6 hours after ingestion. Salicylate poisoning has decreased as acetaminophen's usage has increased; however, salicylates remain as an ingredient in many over-the-counter compounds, such as Pepto-Bismol, Ben-Gay, and oil of wintergreen. False-negative results on dipstick for blood may occur with ascorbic acid (vitamin C) ingestion. The most common form of acute glomerulonephritis in school-age children is poststreptococcal glomerulonephritis. Clinical episodes are often termed crises because they occur suddenly. Prematurity and barotrauma from prolonged mechanical ventilation are significant risk factors for the development of bronchopulmonary dysplasia or chronic lung disease of infancy. Worldwide, each year approximately 2 million children are hospitalized and 450,000 children die from disease caused by rotavirus. Syndromes caused by teratogens(Table 5-2) 1. But it is also one of the best asset for other tests too. I also cannot thank enough some of my finest teachers and role models during my training, Dr. Leigh Grossman, Dr. Dick Kesler, and Dr. Frank Saulsbury of the University of Virginia School of Medicine, whose impact has been immense. Age of onset. BMI > 85th percentile (if 12–16 years old), or smoking b. Supportive care is the most effective management. A physeal fracture that extends into the metaphysis is a Salter–Harris II fracture. A clear retinal image should be ensured by correcting any refractive errors with eyeglasses or by surgically removing any visually significant lens opacities. USMLE step 2 types of pediatrics questions are given at the end of every chapter. Acetaminophen. The answer is D [, section II.E.6]. Ferritin can be a useful assessment of iron stores; however, because ferritin is also an acute-phase reactant, it may be increased in infection, disease states, and stress, therefore appearing normal. 174 4. Type I (classic type): mild quantitative deficiencies of vWf and factor VIII protein. Becker muscular dystrophy A 2-year-old boy is brought to the office by his parents, who note that their son has had weakness in his legs for the past several months that appears to be getting worse. Sacrococcygeal teratoma is the most common teratoma during the first year of life. Other findings include difficult to treat thrush, Pneumocystis jirovecii pneumonia (PCP), and loss of developmental milestones. Viewed from above, the skull has a parallelogram shape with the back of the head, ear, and forehead of one side displaced anteriorly. Hirschsprung disease is five times more frequent in male infants, and in 80% of cases there is a family history. 222 VII. Constipation from gastrointestinal smooth muscle dysfunction 4. Myasthenia gravis, which is more common in girls, presents with weakness that increases during the day and normal deep tendon reflexes. Abdominal ultrasound is the most accurate diagnostic tool in the hands of an 374 experienced ultrasonographer. Imaging studies may include abdominal radiography to identify obstruction, mass, or free air; ultrasound for evaluation of the viscera and fluid; and computer tomography (CT) scan (often with oral, intravenous, and/or rectal contrast) to visualize abdominal anatomy and pathology. Other complications include respiratory insufficiency, acute respiratory distress syndrome (ARDS), renal failure, shock, and GI bleeding. Acute cerebellar ataxia of childhood 1. Approximately 10% of patients with Kawasaki disease may develop hydrops of the gallbladder, which presents with acute right upper quadrant pain. Studies show pancytopenia, low reticulocyte count, and hypocellular bone marrow. Infants typically present with ascending paralysis. Other severe manifestations include cardiac dilatation and heart failure. Laboratory diagnosis is important in establishing a definitive diagnosis because erythema migrans is not always present, and the clinical features and routine laboratory test results may be nonspecific. Henoch–Schönlein Purpura (HSP) II. Wounds on the face or large wounds should be sutured if less than 12 hours old. Cervical Lymphadenitis VIII. The vomiting occurs three to four times per day, and her parents report she has been very fussy. In contrast, nonaccidental burns typically have a clear line of demarcation (e.g., "stocking" or "glovelike" pattern, suggesting submersion injury). Consideration should be given for further workup, including possible electroencephalogram to rule out seizure disorder and pH probe study to rule out gastroesophageal reflux. The answer is C [, sections I and V.D]. Cryoprecipitate, which contains intact vWf, may be used for serious bleeding, for extensive surgeries, or for type III disease. Type I second-degree AV block (also known as Wenckebach) is characterized by progressive prolongation of the PR interval leading to failed AV conduction (question 105). It rarely affects the pulmonic or tricuspid valves. The most common presentation is meningitis. Antinuclear antibodies (ANA) in patients with systemiconset JIA are negative, in contrast to most patients with early-onset oligoarticular JIA and 50% of patients with polyarticular JIA in whom ANA is positive. Causes of an increased anion gap (>16) may be recalled using the mnemonic AMUDPILES (alcohol, methanol, uremia, diabetic ketoacidosis, paraldehyde, iron and isoniazid, lactic acidosis, ethylene glycol, salicylates). A CBC, ESR, bone age, TSH and T4, testosterone or estradiol, FSH, LH, and prolactin levels are necessary. Causes include brain tumors, prior CNS irradiation, CNS vascular malformations, autoimmune diseases, head trauma, and congenital midline defects. Constipation is defined as a reduction in defecation that causes adverse symptoms, which may include difficult defecation, painful defecation, abdominal discomfort, and stool retention. Tumors in this location typically present with hematuria, urinary tract obstruction, vaginal bleeding, and/or an abdominal mass. It is important to distinguish between prenatal onset and postnatal onset. Prognosis varies depending on the severity of initial symptoms before diagnosis and compliance with treatment. Intrahepatic mechanical obstruction (e.g., paucity of intrahepatic bile ducts, Alagille syndrome) 5. Liver Tumors Liver tumors include hepatoblastoma and hepatocellular carcinoma. Short bowel syndrome was formerly a fatal disorder. Seventy-five percent of the testicular volume is the seminiferous tubules. 618 b. Secondary prevention (e.g., care coordination for children with special health care needs) B. General concepts. Increased methionine in plasma b. Routine screening of adolescents who are asymptomatic for Trichomonas vaginalis, bacterial vaginosis, herpes simplex, and human pailloma virus (HPV) is generally not recommended. The answer is C [III.C.1]. The patient's paternal grandfather died of a stroke at 32 years of age, and the patient's father has hypertension. In contrast, weakness is the decreased or less than normal force generated by active contraction of muscles. The scenario of constitutional symptoms in the face of a classic rash suggestive of erythema migrans, shortly after a camping trip, 605 6. Disorders of testosterone synthesis. An overview of different categories of IEM, including examples of conditions within each category, is summarized in Figure 5-3. Timing of vaccination: recommended at 2, 4, and 6 months of age HIB vaccine a. Hypotension, tachycardia, hypoxia, and capillary leak may be seen in severe, acute hemorrhagic pancreatitis. Pancytopenia and Aplastic Anemia III. A 5-year-old girl presents with failure to thrive. This honeymoon period may last from months to 1– 2 years. Neonatal myasthenia is a transient weakness in the newborn period secondary to transplacental transfer of maternal AChR antibodies from a mother affected with myasthenia gravis. Abortive treatment includes nonsteroidal anti-inflammatory drugs (NSAIDs) and triptans, selective 5-HT agonists, available in injectable, intranasal, and oral forms. The diet should consist of meals with no added salt. Hypoalbuminemia and elevated serum cholesterol are present. The recommended initial treatment for moderate persistent asthma is either a medium-dose inhaled corticosteroid OR a low-dose inhaled corticosteroid with a long-acting inhaled β2-agonist for the controller, AND an inhaled shortacting β2-agonist as needed for the reliever. Long-term complications include growth impairment, learning problems, hearing loss, orthopedic deformities, and chronic lung disease. Obesity, retinitis pigmentosa, hypogonadism, and polysyndactyly 4. In this case, further examination should be done to confirm HUS. Gowers sign may be positive (i.e., difficulty standing from the seated position, such that one has to use his or her arms to "climb" up the thighs for support). Diagnosis is on the basis of AP, lateral, and oblique views of the lumbar spine. Risk factors include female sex, firstborn, breech presentation, and family history of DDH. Pain or left-sided curves are signs of underlying disorders, and should prompt investigation for nonidiopathic causes. Physical features include hair that is sparse, lightly pigmented, and/or kinky. 93. In neonatal myasthenia, hypotonia, weakness, feeding problems, and weak cry are the most common findings. In neonatal myasthenia, symptoms are mild and generally resolve within 1–3 weeks. Fatty Acid Oxidation Disorders Fatty acid oxidation is a major source of energy and ketone generation. Children not fully vaccinated by 2 years of age should be vaccinated at subsequent visits. Severe dehydration (≥10%) is associated with tachycardia and feeble pulse, sunken fontanelle, sunken eyes, no tears, parched and cracked lips and buccal mucosa, 409 clammy skin, and capillary refill >3 seconds, in a patient with lethargy and anuria. Patients with Turner syndrome classically have a webbed neck with a low posterior hairline, a shield chest with widely spaced nipples, and transient swelling of the hands and feet during the newborn period. Jaundice, hypoglycemia, hepatomegaly, liver failure, renal failure, and bleeding c. Daily oral penicillin prophylaxis is started in the first few months of life to decrease the risk of S. Ovarian teratomas are the most common ovarian tumor and are generally benign. β-HCG—secreting tumors are unique to boys. It may also be defined as a hematocrit (Hct) > 60% or as an Hgb or Hct more than 2 standard deviations above normal values for age. Review of systems should include questions about cold intolerance and constipation (hypothyroidism), abdominal pain, diarrhea or bloody stools (inflammatory bowel disease), and headaches and vomiting (brain tumor). This classification is no longer recommended because these categories coexist in many cases. RF is often elevated in SLE, but like ANA, is nonspecific. Pneumococcal HUS is triggered by infection with pneumococcus and accounts for about 5% of the cases of HUS. Etiology is unknown. CDGP (i.e., immature hypothalamus or "late bloomers") is much more common in boys than in girls. Asymmetric responses suggest hemispheric injury. False positives may result if the urine is very concentrated (specific gravity > 1.025) or alkaline (pH ≥ 7.0), or if the patient has received certain medications (e.g., penicillin, aspirin, IV contrast imaging agents, oral hypoglycemic agents). Advantages: no need for an injection. The patient's restrictive vegan diet places her at risk for vitamin B12 deficiency. Achondroplasia is the most common cause of disproportionate short stature. Single-photon emission computed tomography (SPECT) may demonstrate increased perfusion to the thalamus and striatum. Intestinal ulceration and infarction of the bowel cause bleeding (see Chapter 11, section VII). Concise descriptions, clinical correlations, numerous figures, and nearly 100 tables detail the essentials of pediatrics, accompanied by case-based review questions at the end of each chapter and a comprehensive end-of-book exam to build test-taking confidence and help your students excel in clerkship.What People say about BRS Pediatrics (Board Review Series) 2nd Edition:1- The book (BRS Pediatrics second edition) is dense with information, but the bullet-point format and tables help keep it organized. The incidence of neural tube defects varies with geographic location. Associated abnormalities may include congenital torticollis, genitourinary anomalies, congenital heart disease, hearing loss, and congenital failure of scapula to descend to its proper location. This disorder should be suspected in all infants with hypotonia. GI sequelae include FTT, esophageal strictures, and Barrett esophagus (conversion of the normal stratified squamous epithelium of the esophagus into columnar epithelium, a condition which may be associated with esophageal adenocarcinoma). Poisonings A. The chance of developing chronic renal insufficiency as a result of VUR is 50%. Seizures may spread to the face and arm and then secondarily generalize into tonic–clonic seizures. Absence of a region on paternally derived chromosome 15 The response options for statements 12–14 are the same. Congenital Glaucoma A. Highly contagious c. Hypsarrhythmia is a characteristic EEG pattern consisting of a high amplitude, disorganized background with multifocal spike-wave discharges. This patient most likely has congenital myotonic dystrophy. Orthopedic problems (e.g., rib anomalies, deformities of the lower extremities, and lower extremity fractures from loss of sensation) 5. Status epilepticus is defined as 5 or more minutes of continuous seizure activity, or repetitive seizures without recovery of consciousness. SB occulta is an incomplete closure of vertebrae without herniation of tissue through the cleft. Sensation in the lower extremities is intact. Cholestatic jaundice is defined as retention of bile within the liver and occurs clinically when the direct component of bilirubin is >2 mg/dL or ≥15% of the total bilirubin. Hepatocellular carcinoma may occur in both young children and in adolescents. Aortic root dilatation, mitral valve prolapse, and valvular regurgitation. Materials appearing in this book prepared by individuals as part of their official duties as U.S. government employees are not covered by the above-mentioned copyright. Young men may be vaccinated up to 21 years of age, and women may be vaccinated up to 26 years of age. 456 FIGURE 12.1 Differential diagnosis of hypotonia. Clinical Assessment of Pulmonary Disease III. Plain film radiography has a limited role in diagnosis but may be useful if a foreign body or perforation is suspected. Organism. Every patient with SS disease is managed with urgent assessment and appropriate cultures (blood and urine), chest radiograph to rule out pneumonia, and parenteral antibiotics until bacterial infection can be safely excluded. Neurologic insult (hypoxic central nervous system [CNS] injury) is directly related to the length and severity of the hypoxia. Management includes withdrawal and avoidance of suspected dietary protein. Radiography of the extremities reveals dysostosis multiplex. Membranous nephropathy B. Duodenal stenosis may present with emesis, weight loss, and FTT. Patients may have sudden 487 6. There would be no significant difference in his risk of UTI had he been circumcised. Constipation A. Cholinesterase inhibitors can be used to aid with feeding and respiratory support. One-third are malignant. Fifty percent of all pediatric liver transplants are performed for liver failure caused by biliary atresia. Bone Tumors A. Table 13-3 Clinical Features and Management of Crises Occurring in Sickle Cell Disease Crisis Clinical Features Vaso-occlusive crisis Painful bone Most common crisis crisis Ischemia/infarction of bone or marrow Deep, gnawing, or throbbing pain lasting 3–7 days Subtype: acute dactylitis—painful swelling of digits of the hands and feet Acute abdominal crisis Stroke Priapism Management Pain control Intravenous fluids at 1–1.5 × maintenance Incentive spirometry to decrease the risk of acute chest syndrome Severe, unremitting pain may respond to partial exchange transfusion. Classic features include a cherry-red macula, enlarging head circumference, neurodegeneration with severe developmental delay, progressive blindness, and seizures. Slurred speech and nystagmus are often present, and hypotonia and tremors are less common. More than 13 mutated genes have been described that lead to nephronophthisis. Glomerulonephritis VI. Inappropriate TLESR allows excessive gastric refluxate to enter the esophagus and even the oropharynx. In addition to the characteristic signs and symptoms of anemia, patients may have failure to thrive, chronic diarrhea, and irritability. During infancy, the risk of developing a UTI is the same as that of an infant girl. Treatment includes prevention of trauma and replacement of factor VIII. Inherited disorders of collagen synthesis (e.g., Ehlers–Danlos syndrome) may result in capillary fragility. Correction of acidosis, hypoxemia, and hypercarbia are paramount. Genetic evaluation may be useful if a genetic syndrome is suspected. Congenital aqueductal stenosis is the most common cause of noncommunicating hydrocephalus. After gut resection, carbohydrate and fat malabsorption with steatorrhea are common. Juvenile Idiopathic Arthritis (JIA) IV. Congenital glaucoma not only results in optic nerve injury but also expands the size of the eye, because the eye wall is much more elastic during infancy. Management involves avoidance of fructose-containing foods and beverages. Iron-deficiency anemia is the most common blood disease during infancy and childhood. Early disease (90% of cases. The inheritance pattern is most commonly autosomal dominant with variable expression but autosomal recessive inheritance has also been described. Treatment includes dietary folic acid and identification and treatment of the underlying cause. Books of BRS series are one of the best short books. 492 b. Normal visual development is dependent on both of the following: a. Migraine equivalent. Recurrent common bacterial infections C. AP radiographs of the pelvis may be used to assess for DDH if the infant is older than 6 months. 387 Yes FTT, delayed sexual development, oral aphthous ulcers, erythema nodosum, arthritis, renal stones Strictures, fistulas, abscesses Somewhat increased X. Production may be appropriate or inappropriate. A neonatal history of prolonged neonatal jaundice, hypoglycemia, microphallus, cryptorchidism, and midline defects (e.g., cleft palate) may be present. It presents within the first 6 months of life. General Concepts of Inborn Errors of Metabolism (IEM) A. Salicylates uncouple oxidative phosphorylation, producing lactic acidosis and enhancing ketosis. FAP a. Known risk factors include intrauterine constraint or crowding, and metabolic abnormalities including hyperthyroidism and hypercalcemia. pylori is high during childhood. On examination, you note that the boy's height and weight are at the 50th percentile, but his head appears very small. The cause is accumulation of fluid as a result of NLD obstruction. Sexually Transmitted Infections (STIs) VIII. Physical examination reveals an afebrile infant with normal vital signs. It often carries a worse prognosis than STEC-HUS because of necrotizing pneumonia. Hemarthroses are unusual. Epidemiology. Diagnosis is on the basis of AP and frog-leg lateral radiographs of the pelvis, which show increased density in the affected femoral head, or a crescentic subchondral fracture in the femoral head, termed the "crescent sign." d. Your nurse takes the initial history and reports that he was adopted from another country. Normally, a cortisol value > 18–20 μg/dL after ACTH stimulation is considered an adequate response. Skin involvement occurs in 50% of patients. Pathogenesis and etiology a. | Miller, Lee T. Angelman syndrome is associated with severe mental retardation and a small head, although a puppetlike gait and inappropriate bouts of laughter are also characteristic. Prolonged-type AIHA is characterized by a protracted course and high mortality. Education and close follow-up every 3 months. First-degree relatives of a patient with type 1 DM have 10-fold higher rate of developing type 1 DM. Abdominal peristaltic waves may be visible after feeding. The index for suspicion of abuse should be high, especially in situations in which injuries found on examination are unaccounted for or are inconsistent with the caregiver's history or the child's developmental abilities. In boys, physical examination shows testicular enlargement, pubic hair, and rapid growth. The answer is B [III.D and Table 16-4]. Order an electroencephalogram. The mother produces antibodies to either the A or B blood group antigen that then pass to the fetus, causing hemolysis with resultant jaundice. It is most common in males and is associated with obesity. Acute cerebellar ataxia of childhood presents with ataxia (unsteady gait or truncal unsteadiness) rather than weakness. There is no increased association with other extraintestinal anomalies; however, there is an increased incidence of other bowel abnormalities, including malrotation and volvulus, and stenosis and atresia that are attributed to inadequate intestinal blood flow. Signs and symptoms depend on the site of involvement. Positioning in an upright or sitting position or raising the head of the bed after feedings or when asleep b. The typical toddler's diet consists of large quantities of iron-poor cow's milk. Unilateral conjunctivitis may be associated with a foreign body, corneal ulcer, or herpes simplex keratitis. The diagnosis of Guillain–Barré syndrome should be considered in any child with ascending symmetric weakness or paralysis, absence of deep tendon reflexes, and a normal sensory examination. Guarding and peritoneal signs may also be present. Benzathine penicillin intramuscular injection (one dose) or b. Almost all head growth occurs prenatally and during the first 2 years of life. These seizures are characterized by increased thoracic and abdominal muscle tone, followed by clonic movements of the arms and legs, eyes rolling upward, incontinence, decreased consciousness, and a postictal state of variable duration. Begin treatment with phenobarbital. The answer is B [VI.A]. Type II second- degree AV block is an abrupt failure of AV conduction without progressive prolongation of the PR interval. Third spacing and pooling of blood in the vasculature leading to hypotension 4. The answer is B [, section IX.C.4]. Myelomeningocele requires urgent surgical repair within 24 hours of birth, or in some cases in utero, to reduce the morbidity and mortality from infection and to prevent further trauma to the exposed neural tissue. ≥10,000 colonies in samples obtained by sterile urethral catheterization c. Bilateral involvement in one-third of patients 5. In boys, sexual precocity tends to be pathologic, and all patients need evaluation with an MRI of the head. Relevant Gram-negative organisms include Neisseria gonorrhoeae in neonates and adolescents and Kingella kingae in children younger than 3 years. Hemolytic uremic syndrome is a vasculitis characterized by microangiopathic hemolytic anemia, thrombocytopenia, and acute renal failure. Ocular Trauma VI. 12. Nonlive vaccines are not infectious and tend to induce immunity for shorter periods, thus requiring booster immunizations. Inspection includes evaluation for pupil and eyelid symmetry, head turn or head tilt (i.e., torticollis), and conjunctival injection. Timing of vaccination: Vaccine is recommended at 12–18 months of age with a booster at 4–6 years of age. History is critical in differentiating inflicted from noninflicted trauma. Other findings may include flattened occiput, occipital hair loss, arthrogryposis (muscle contractures), congenital hip dislocation, and pectus excavatum. The vaccine is ideally administered before the onset of sexual activity to develop protective immunity but may be administered to males through age 21 years and to females through age 26 years. Because many inherited renal diseases present in childhood, a careful family history is critical in all children with renal disease. Undigested sugars are osmotically active and draw water into the intestinal lumen, causing increased stool volume, increased peristaltic activity, and decreased transit time. Patients with thrombosis and antiphospholipid antibodies should be anticoagulated with low–molecular weight heparin or warfarin. A 12-month-old female infant who was born at term with a birth weight of 3.5 kg presents now for a well-child visit. STEC-HUS occurs as a result of an intestinal infection with a toxin-producing bacterial strain. Genetic mechanism 1. Assisted ventilation may help individuals to live longer. Leukemias III. The majority of children with VUR eventually outgrow the reflux, although a minority develop severe renal impairment owing to reflux nephropathy from severe VUR. Uncircumcised male infants under six months of age are at a greater risk for UTI than circumcised infants. Physical examination reveals epigastric tenderness, abdominal distension, and decreased bowel sounds. Mortality approaches 5%, almost exclusively in children who are steroid-resistant and almost always from overwhelming infection or thrombosis. Children younger than 1 year are usually protected by maternal antibody and have low risk of disease. In young children, the ratio of boys to girls is 2:1. Additional problems include intellectual disability, cataracts, cardiac arrhythmias, and infertility. Meconium ileus is a common neonatal manifestation of cystic fibrosis. Patients with HUS have a microangiopathic hemolytic anemia, renal impairment, and thrombocytopenia, which may result in visible petechiae on the skin. Supportive care is the most important management. The answer is D [III.E.3]. Vector. A large hyphema may obscure the pupil and iris, thereby impairing vision. Live attenuated influenza vaccine (LAIV), administered intranasally a. Human Papilloma Virus (HPV) vaccines a. Patients with hypogonadotropic hypogonadism frequently have CDGP [see section I.D.1.b for a description of growth pattern]. Rationale for vaccine: 90% of affected infants will develop chronic infection. Conjunctivitis associated with contact lens use may be secondary to allergy to the contact lens solution, to a corneal abrasion, or to a vision-threatening bacterial corneal ulcer. Types of germ cell tumors include seminoma (in males), dysgerminoma (in females), teratoma, yolk sac tumor, embryonal cell carcinoma, and choriocarcinoma. The effectiveness of home apnea monitor in preventing sudden infant death syndrome has not been established. Clinical features include a claw hand because of loss of intrinsic function and a decreased ability to extend the elbow and flex the wrist. 193 Review Test 1. Initial resuscitation includes the ABCs, cervical spine immobilization (because of the possibility of coexistent head trauma), and removal of wet clothing to reduce heat loss. These disorders include deficiency of factor VIII and von Willebrand disease (both of which are factor VIII–related disorders) and deficiency of factor IX. Peak incidence is at 6–7 years of age. Spina Bifida A. 217 D. The patient has a 10% chance of developing severe chronic arthritis. The autosomal recessive inheritance of CF means that each subsequent sibling has a 25% chance of having the disease. Onset of symptoms occurs 12–48 hours after ingestion of spores. VACTERL (VATER) association a. Severe respiratory insufficiency from pulmonary hypoplasia and pulmonary hypertension is common in patients with CDH, due to mechanical compression of the lung and defective pulmonary vascular development. Hydrocephalus A. Diagnosis is made by performance of the dihydrorhodamine (DHR) flow cytometry test (question 142). Facial weakness and hyporeflexia are common. Adult glaucoma is characterized by increased intraocular pressure that damages the optic nerve but does not change the size of the eye. Isolated gonadotropin deficiency associated with anosmia (inability to smell) 2. More severe injury results from compounds that have a pH 12. Her murmur as described is typical for a benign Still's (left ventricular outflow tract) murmur. Absence epilepsy of childhood a. Meconium-stained fluid can cause meconium aspiration syndrome and is more likely to present with respiratory distress than feeding intolerance. Chronic granulomatous disease (CGD) is characterized by defective neutrophil oxidative metabolism that results in impaired intracellular killing of catalase-positive bacteria and some fungal pathogens. A complete blood count (CBC) should be obtained to examine for thrombocytopenia, as well as a peripheral smear to look for microangiopathic hemolytic anemia. Premature thelarche 1. Arthritis, the hallmark of late disease, occurs months after the initial clinical presentation. Other types of diabetes—Cystic fibrosis–related diabetes, medication induced diabetes, and monogenic diabetes (maturity-onset diabetes of the young [MODY]) 220 VI. ICP = intracranial pressure. However, it is indicated for patients who have impaired creatinine clearance or for those who do not respond to initial management with corticosteroids. The infected tick must be attached for more than 36– 48 hours before the risk of B. Delayed dental eruption may suggest CDGP or GH deficiency. RDS affects approximately 0.5% of all neonates and is the most frequent cause of respiratory distress in preterm infants. Serious DKA is uncommon because children with type 2 DM retain some residual insulin secretion. Which of the following is the most likely diagnosis? Varices as a result of portal hypertension or vascular malformations are less common causes. Glaucoma 3. Niesen, MD, FRCPC, Division of Pediatric Neurology, Cedars-Sinai Medical Center, Los Angeles, California, Assistant Professor of Pediatrics and Neurology, David Geffen School of Medicine at UCLA, Los Angeles, California Kathy L. Laboratory features consistent with the diagnosis include transient low serum complement levels. Urinary dipstick is the most frequently used method of screening for proteinuria and detects variable levels of albuminuria. Juvenile myasthenia gravis affects girls two to six times more frequently than boys. The clinical course is generally more severe with multiorgan damage. Ideally, the series is initiated during the preteen years, to facilitate development of an immune response before the initiation of sexual activity. Noonan syndrome a. Clinical features. Leg length discrepancy can mimic scoliosis. However, a supracondylar fracture is a typical fracture in a child of this age, and this injury fits with the purported mechanism. With time, prolonged contact of the esophageal mucosa with gastric contents results in inflammation. Although universal tuberculosis screening was required for school entry in the past, targeted screening based on risk factors is now recommended. It is a diagnosis of exclusion, and it is the most common cause of cholestasis in the newborn. Infectious agents include Neisseria gonorrhoeae, Chlamydia trachomatis, and herpes simplex virus. Lee Miller, Kate Perkins, Paul Chung, Alice Kuo, Tom Klitzner, and Ellen Wald. Tuberous sclerosis is the most commonly identified cause of this disorder. Pathologic fractures may occur. Diagnosis is by rectal biopsy revealing absence or paucity of ganglion cells. DTaP is safer but has a waning 29 3. Roseola presents as a pink papular rash that lasts only a few days and is preceded by a high fever, and varicella is associated with vesicular and crusted skin lesions as well as fever. If the lesion is not surgically resectable, radiation therapy is used for local control. The visual changes may include blurred vision, small areas of decreased vision (scotomata), streaks of light, or hemianopsia. The postulated cause is thought to be molecular mimicry, where molecular similarity between antigens from the infectious entity and the patient (e.g., cerebellar structures) triggers an autoimmune response. Specific Fungal Infections XV. These patients present with salt-wasting crises, glucocorticoid deficiency, and ambiguous genitalia as a result of an early block in all three adrenal cortex steroid pathways. Obstruction or damage to the biliary system causes retention of bile enzymes that damage the cells lining the biliary tree and hepatocytes. History should focus on timing of jaundice onset, associated symptoms (e.g., poor growth, bleeding, dark urine), feeding regimen (e.g., breast or bottle, frequency of feedings), and stool quality and color. Treatment includes surgical excision of both the tumor and the coccyx to prevent recurrence. Or if you really like it and want to buy brs pediatrics in hard copy, you can buy it from amazon with free shipping from our link below. Clinical features of Hgb SC disease are less severe than SS disease. It occurs in 70% of patients and most commonly involves the elbows, knees, ankles, and wrists. Most infections during infancy and childhood are asymptomatic. Colonoscopy with polypectomy is the definitive treatment. Brown Now featuring more than 400 board-style questions with complete answers and explanations, BSR Pediatrics, Second Edition prepares medical students for success in pediatric rotations, shelf exams, and the UMSLE Step 2 with a comprehensive overview of general and subspecialty pediatrics presented in the popular Board Review Series outline format. There may be an effusion in the hip, and if fluid is present with clinical concern for the possibility of septic arthritis, it should be aspirated and analyzed to rule out septic arthritis. In BMD, patients become wheelchair dependent in their twenties. The Nissen fundoplication wraps the fundus of the stomach around the distal 3.5 cm of the esophagus, resulting in reduction of TLESR in as many as 90% of children. For nonmetastatic disease, about 70% of patients are cured, and if metastatic at diagnosis, about 50% are cured. Colonoscopy with biopsies of the colon and terminal ileum confirms the diagnosis. If pain persists, analgesia, traction, casting, or crutches may be used. Lerner

452 I. Laboratory results reveal no acidosis, hypoglycemia, or hyperammonemia, but there are significant amounts of ketones in the urine. Bracing prevents progression of the curvature; therefore, bracing is not likely helpful after skeletal growth is completed. Physical examination is usually normal, and patients are often asymptomatic. Chronic Abdominal Pain A. Patients with active bleeding or hemodynamic changes should have blood sent for type and crossmatch. Laboratory studies should include hemoglobin and platelet counts, coagulation studies, serum transaminases, and a blood urea nitrogen (BUN) level (suggests a GI bleed if elevated). Eliciting parental and patient concerns 2. Antibiotics should be administered immediately. The answer is E [, section II.A.5]. No contact sports, tumbling, or trampolines are allowed if there is any sign of instability. Emesis is benign, but often dramatic and traumatic for parents. Chemotherapy includes high-dose methotrexate, cisplatin, and doxorubicin. He was born at term by normal spontaneous vaginal delivery and seemed normal at birth, but has not been meeting his developmental milestones. Hyperoxia test in infants with lung disease. FTT and more significant liver disease occur later in infancy in 33% of patients. Examples of mitochondrial DNA disorders include Kearns–Sayre syndrome (ophthalmoplegia, pigmentary degeneration of the retina, hearing loss, heart block, and neurologic degeneration) and MELAS (mitochondrial encephalopathy, lactic acidosis, and strokelike episodes). Esophageal perforation with mediastinitis may follow alkali ingestion. An 8-year-old girl is noted by her teacher to have brief staring spells throughout the day. JDM is the most common idiopathic inflammatory myopathy of childhood (85% of all myopathies). Primary AIHA is generally idiopathic in which no underlying disease is identified. Other brainstem reflexes should be assessed to determine the extent of injury to the brainstem. Learning disabilities are seen in half of patients. In these patients, NS is diagnosed during an evaluation for asymptomatic proteinuria, and they are less likely to have MCD. Malnutrition b. Hepatic dysfunction. Myasthenia Gravis A. Sydenham chorea, if severe, may be treated with haloperidol. Normal stature or slightly shorter than the rest of the family 3. Clinical characteristics are not generally present until protective Hgb F declines (by 6 months of age). Low factor VIII protein activity in the presence of normal von Willebrand factor assay d. LGA infants also have an increased association with congenital malformations and increased risk of neonatal mortality compared with age-matched appropriate-for-gestational age (AGA) infants. Eye anomalies include posterior embryotoxon. This patient has suffered a major burn injury with inhalation of hot gases. Mineral oil is useful but should not be used in aspiration-prone infants and children. In the United States, the prevalence is approximately 0.5 in 1000; however, in China and India, it is as high as 10 per 1000. Diagnosis of SCFE outside the typical age range, or in patients who are thin or have short stature, should prompt consideration of endocrine, renal, or genetic disorders. Serologic testing for this condition is unreliable, and the diagnosis must be confirmed by culture of body fluids or tissue. Most children, when they have completed their growth, are within ±2 SDs, or 4 inches, of their MPH. Treatment includes reassurance and pain control (e.g., acetaminophen, ibuprofen). Absorption then occurs in the terminal ileum. Brisk deep tendon refluxes (DTRs), but usually with a downward plantar response E. Intestinal Anatomic Obstructions That Result in Vomiting A. The cause is adenovirus, types 3 and 7. 769 114. Pulmonary function studies show decreased lung volume consistent with restrictive lung disease. Perforated appendicitis requires drainage of purulent material, irrigation of the peritoneal cavity, and parenteral antibiotics. 359 360 CHAPTER 10 361 Gastroenterology Dorsey Bass, Lloyd J. Hunter syndrome (MPS type II) is an X-linked recessive disorder caused by a deficiency of iduronate-2-sulfatase. Insulin a. NLD occurs in 1–5% of children. 592 V. Expansion of the repeats occurs much more frequently in mothers. Female sexual differentiation. 83. Severe hypertension is common. Onset 18 months of age. Patients may have developmental delay and/or intellectual disability. Physical therapy is often needed. Crohn disease, tumors, and radiation enteritis may also lead to short bowel syndrome. Fetal hydantoin syndrome a. Fatty Acid Oxidation Disorders IX. Mutations can either be inherited or sporadic. Commonly used oral antibiotics include trimethoprim–sulfamethoxazole or cephalexin. Table 12-1 Causes of Impaired Consciousness and Coma in Childhood and Adolescence Infectious Meningitis Encephalitis Focal infection (abscess, cerebritis) Inflammatory Vasculitis Demyelinating disorders (acute disseminated encephalomyelitis) Traumatic Concussion Abusive head trauma Diffuse axonal injury Cerebral hematoma or contusion Vascular disease Cerebral infarction Cerebral hemorrhage Neoplasm Hypoxia Shock Cardiac failure Nonfatal drowning Carbon monoxide poisoning Metabolic disorders Fluid or electrolyte imbalance Hypoglycemia/hyperglycemia Hyponatremia Diabetic ketoacidosis Organic acidemias Amino acidemias Hepatic encephalopathy Urea cycle disorders Disorders of fatty acid metabolism Reye syndrome 464 Hyperammonemia Hypothyroidism or hyperthyroidism Nutritional Thiamine deficiency Pyridoxine deficiency Folate and vitamin B12 deficiency Toxins/poisons Alcohol Prescription medications Atropine, scopolamine, benzodiazepines, barbiturates, lithium, opiates, tricyclic antidepressants Over-the-counter medications Heavy metal poisoning Lead, mercury, arsenic Illicit drugs* * Amphetamine, cocaine, and hallucinogens (lysergic acid diethylamide [LSD], mescaline, phencyclidine hydrochloride [PCP]) cause agitation, confusion, delirium, and hallucinations but not coma. The fracture is nonangulated and cuts all the way across the bone (transverse) in the mid-thigh (diaphyseal fracture). 766 97. Normal extraocular movements and normal sensory examination f. Local complications include abscess formation, necrosis, and pancreatic pseudocyst. Bruises 1. Intussusception causes bowel wall edema and hemorrhage, and may lead to bowel ischemia and infarction. Elevated IgG anti-HAV also occurs early in infection and confers lifelong immunity. Anemia is a reduction in the red blood cell (RBC) number or in the hemoglobin (Hgb) concentration to a level more than 2 standard deviations below the mean. Infection is most commonly caused by Salmonella species acquired through the gastrointestinal (GI) tract, although Staphylococcus aureus may also cause osteomyelitis. FFR is behavioral. Normal stool patterns. Sydenham chorea occurs later than the other rheumatic fever manifestations, often beginning subtly, months after GABHS pharyngitis. Administration of metronidazole empirically for the treatment of Clostridium difficile E. Inflammatory Bowel Disease (IBD) A. Imaging studies may include a renal or pelvic ultrasound and a nuclear renal scan to evaluate renal function. Laboratory studies include an elevated WBC count, ESR, and CRP. Infants of Diabetic Mothers (IDMs) XV. Duchenne muscular dystrophy B. Pathophysiology involves an altered ion-channel regulator (CFTR) protein, resulting in abnormal sodium and chloride transport in epithelial cells, which can cause a hyponatremic, hypochloremic metabolic alkalosis. The diagnosis of dermatomyositis should be suspected in a child 606 with proximal muscle weakness, a violaceous heliotrope rash around the eyes, and erythematous, hypertrophic papules over the knuckles (Gottron papules). Ultrasound is used to measure the pylorus muscle length and thickness and is the diagnostic method of choice. Therefore you should download this book below in pdf format. (Key point: Male infants presenting with saltwasting crisis can be confused for pyloric stenosis. The mass is filled with CSF but does not contain spinal cord tissue and can be transilluminated. Craniofacial findings are most prominent in infancy and include low-set, posteriorly rotated ears, wide-spaced eyes (hypertelorism), eyelid ptosis, and 175 2. A fluctuant midline mass is present overlying the spine. Evaluation of the Newborn II. Pulmonary disease improves with time and lung growth, although chest radiographs may remain abnormal for years. Sydenham chorea occurs in approximately 25% of patients with rheumatic fever. Prophylactic treatments include calcium-channel blockers and valproic acid. Examination of a peripheral smear for evidence of microangiopathic hemolytic anemia D. Management of OTC deficiency includes a low-protein diet and modalities to decrease ammonia levels. AKI is defined as an abrupt decrease in the ability to excrete nitrogenous wastes. Unlike in adults, herniation in adolescents is caused by repetitive activity and rarely by trauma. diagnosis? Infectious Disorders of the Respiratory Tract IV. The most common organic acidemias are caused by abnormal amino acid catabolism of branched-chain amino acids, and are characterized by urinary excretion of nonamino organic acids. Pneumonia is unlikely given the absence of fever, and therefore a noninfectious cause of the patient's symptoms should be sought. Neurofibromatosis (see Chapter 19, Table 19-3) e. Complications include paralysis or even death if the instability is not detected before injury. Types of RTA (Table 11-2) E. On gripping the examiner's fingers, patients are unable to maintain the grip (milkmaid's grip). For most patients, this will require doses of 10– 20 mg/m2 per day of hydrocortisone. Inflammatory Disorders IV. Treatment includes valproic acid or carbamazepine. Annual screening tests for a sexually active girl who is not consistently using a barrier method with each sexual encounter should include nucleic acid amplification tests (NAATs) of urine or a vaginal swab for Chlamydia trachomatis and Neisseria gonorrhoeae, serologic test for syphilis, and human immunodeficiency virus (HIV) testing. Primary classifications include the following: a. On examination, you confirm the inspiratory stridor and also note intercostal retractions and tachypnea with a respiratory rate of 44 breaths/minute. Polio has been eradicated from the Western hemisphere and South Pacific, but remains in isolated pockets throughout the world. Initial laboratory studies should include serum potassium, phosphorus and uric acid, urine pH, and urinalysis to evaluate for proteinuria and glucosuria. There is no obvious precipitating cause, although many migraine sufferers have triggers such as red wine, cheese, preserved meats, and chocolate. Turbulence within the carotid artery would typically produce a bruit that would be a sign of obstruction 764 85. Homocystinuria screening should also be performed in patients who test negative for Marfan syndrome. Nail bed capillary changes d. 604 Answers and Explanations 1. The answer is A [, sections IV.C.1.a and VIII.D.6]. The excess precursor metabolite is 11deoxycorticosterone. His maintenance fluid calculations need to be adjusted for increased ongoing losses should he develop protracted vomiting or profuse watery diarrhea. Evaluation of constipation. Congenital glaucoma results in corneal edema, corneal clouding, and amblyopia. Mucus in the medial canthal area d. Characterized by skin hyperextensibility, abnormal wound healing, and joint hypermobility. Limp is a common symptom during childhood. Conjunctivitis occurring during the first month of life 2. Achondroplasia (see Chapter 5, section IV.H.1) f. The constellation of findings is consistent with Kawasaki disease. Abnormal motor responses to stimuli can indicate the location of brain damage. pyogenes are the most common organisms. Treatment includes abortive therapy with oxygen or triptans. Moderate disease, seen in 30% of cases, presents with nocturnal stooling, cramping, and tenesmus (a continual or recurrent inclination to evacuate the bowels). Bacterial conjunctivitis 1. Definitive diagnosis is by tissue biopsy. Development is often delayed in children with chronic lung disease. Brown and Lee T. No radiograph is needed if the episode was witnessed. Langerhans Cell Histiocytosis (LCH) Review Test Answers and Explanations Chapter 15 Allergy and Immunology I. An emergent urology referral is indicated if the examination was consistent with a diagnosis of testicular torsion. Which of the following is correct regarding her likely diagnosis? Human papilloma virus (HPV) is one of the most common sexually transmitted disease in the United States. Genetic testing confirming homozygous mutations 3. Of note, pruritus in these patients can be debilitating. Viral hepatitis 1. Premature closure of one or more of the cranial sutures (Figure 12). Which of the following is correct regarding his likely diagnosis? d. Autoimmune disorders (e.g., hypogonadism in autoimmune oophoritis, which may also be associated with Hashimoto thyroiditis or Addison disease) 5. Corticosteroids and chemotherapeutics are considered standard of care. Transmission is by perinatal vertical exposure from an infected mother to her fetus; by the parenteral route through exposure to infected blood products, tattooing needles, and intravenous drug use; or by exposure to infected body secretions. The answer is A [Table 5-5]. Vertigo, tinnitus, ataxia, or dysarthria may precede the onset of this headache. Radiographic studies a. Prader–Willi syndrome D. The risk of epilepsy is low (2–10%). The correct answer is to recheck the thyroid-stimulating hormone (TSH) and thyroxine (T4) levels. Symptoms are usually noted by 3 months of age. Back strain is the most common cause of back pain in children. Bilirubin levels are moderately elevated. Neurofibromatosis type 1 is not typically associated with infantile spasms. Past medical history is also significant for meconium ileus during the neonatal period. A 3-year-old girl has a 2-day history of fever, irritability, and emesis. Reactive arthritis can present with arthritis, conjunctivitis, and urethritis. The PaO2 usually increases considerably when 100% oxygen is given, often reaching levels greater than 150 mm Hg. Cyanosis due to lung disease is typically due to ventilation– perfusion mismatch that is improved by giving 100% oxygen. Electromyography may also be helpful in diagnosis and may show brief, small-amplitude muscle potentials with an incremental response during high-frequency stimulation. Physical examination may show the following findings: a. Ataxia can be the result of cerebellar or proprioceptive dysfunction (sensory loss ataxia). Premature closure of multiple sutures is rare, and is associated with severe neurologic compromise. Low hairline, short neck, hypoplastic nails 3. Internal ducts. Standardized growth curves, produced by the World Health Organization (WHO) and Centers for Disease Control and Prevention (CDC), reflect average values for age for 95% of children and are used to plot weight, height, body mass index, and head circumference over time. The inheritance is not clear, but it appears to be a sporadic occurrence. LP shows albuminocytologic dissociation (i.e., increased CSF protein in the absence of an elevated cell count), which is usually evident 1 week after symptom onset. Micronutrients a. Note: Patients with aortic root dilatation are at increased risk for aortic dissection. This involves angling the head at 30° and irrigating each auditory canal 463 with 10–30 mL of ice water. Diagnosis is made by physical examination of the head. Arthritis is classically migratory, asymmetric, and exquisitely painful and often the earliest manifestation of acute rheumatic fever. This vasculitis is characterized by necrotizing granulomas in multiple organs, most commonly the respiratory tract and kidneys. See Figure 11-1 for clues regarding the causes of discolored urine. Increased homocysteine in plasma 2. Group B streptococcus is an organism unique to the neonatal period. Magnetic resonance imaging (MRI) may be indicated to evaluate for structural abnormalities, neuronal migrational abnormalities (i.e., 453 polymicrogyria, lissencephaly), basal ganglia signal abnormalities (seen in metabolic conditions), and deep white matter changes. Glutaric acidemia type I. External genitalia. Fourteen-year-old girl with joint laxity, easily bruisable skin, and a defect in type V collagen 196 Answers and Explanations 1. Management may include range of motion exercises for associated torticollis, repositioning the head during sleep, and increased time in the prone position when awake ("tummy time"). Girls: presence of breast development or pubic hair before 8 years of age b. Management includes frequent feedings with complex carbohydrates and cornstarch to avoid hypoglycemia. Anti–double-stranded DNA (anti-dsDNA) antibodies are much more specific (found only in SLE), and their levels can be used as markers for active disease, especially nephritis. HUS is a condition characterized by acute kidney injury (AKI) in the presence of microangiopathic hemolytic anemia and thrombocytopenia. Treatment includes daily subcutaneous injections of recombinant GH until a bone age determination demonstrates that the patient has reached nearly maximal growth potential (by about 13–14 years of age in girls and 15–16 years of age in boys). The answer is E [, Table 1-5]. Epilepsy is diagnosed if any of the following three scenarios occur: a. All patients should have parenteral glucagon available in case of seizure or coma secondary to low blood sugar. The answer is B [IV.E]. May have yellow or grayish brittle teeth (dentinogenesis imperfecta) which are at increased risk for breakage. Structural and Urologic Abnormalities XIII. But you can download from external source as we don't have excess to it and we can't remove this book from there. It occurs most commonly between the first and second cervical vertebrae and less commonly between the occiput and the first cervical vertebrae. Disease detection through surveillance and screening: a. TPN-associated disease c. Greatly enlarged cystic kidneys c. Further questioning reveals two prior episodes of "brown-colored urine" concurrent with upper respiratory tract infections during the last 3 years. Hypogonadotropic hypogonadism. Clinical features include an increased startle reflex, hypotonia, loss of developmental milestones, hearing loss, cherry-red spot on the macula, and macrocephaly. Low IGF-1 and IGF-BP3 levels, as well as a poor response on GH stimulation testing (with l-DOPA [L-3,4-dihydroxyphenylalanine], arginine, glucagon, or clonidine) d. Cellular damage also leads to the release of intracellular enzymes characteristic of the originating cell. The platelet count may also be elevated. Corticosteroids are often used in patients with severe cardiac involvement, such as congestive heart failure and severe valvular dysfunction. Obstructive airway edema (especially with acid ingestion) 3. In most cases, no treatment is indicated. Obstruction typically occurs distally at Hasner valve. On average, she uses her albuterol inhaler three times per week, but for the past 10 days, she has been wheezing 356 both day and night and is using the inhaler three to four times per day. Three different subtypes of 21-OH 216 deficiency affect the clinical presentation. Patients with polyarticular JIA (whether rheumatoid factor–positive or –negative) are also usually female and have involvement of multiple large and small joints but not typically the sacroiliac joints. Secondary polycythemia is caused by increased erythropoietin production. Management is predominantly supportive but depends on the specific disorder. Systemic pathology: sepsis, meningitis, and both renal and hepatic encephalopathy 2. Infants with congenital heart disease may require treatment with prostaglandin (PGE1) to maintain patency of the ductus arteriosus until surgical repair can be performed. In girls, consider Turner syndrome or gonadal dysgenesis (see Chapter 5, section IV.C.1). Activated charcoal 2. Symptomatic medications, such as antispasmodics, sedatives, or analgesics, are ineffective. Fats are insoluble in water and must be incorporated into bile salt micelles to be absorbed. Thromboembolism can occur in any vessel, increasing the risk of stroke and systemic thrombosis, as well as developmental delay. Biguanides (i.e., metformin) are the preferred drugs in children and are typically used as monotherapy if HbA1c is 12 years SABA Low- Low-dose ICS + LABA OR Medium-dose High-dose High-dose as dose Medium-dose ICS ICS + LABA ICS + LABA ICS + LABA + oral needed ICS corticosteroids Adapted from National Asthma Education and Prevention Program, Third Expert Panel on the Diagnosis and Management of Asthma. This syndrome is defined by a classic triad of findings, including sicca syndrome (dry mouth and eyes), high titers of autoantibodies (usually ANA, RF, SSA, SSB), and connective tissue disease. Henoch– Schönlein purpura is a systemic IgA-mediated vasculitis that involves the skin, joints, gastrointestinal tract, and kidneys. Refer her to an allergist for allergen immunotherapy. Serum antigliadin IgG antibody testing is useful for patients who are IgA deficient. C—colobomas of the iris or retina. Infants are often termed "happy spitters" because they are without other refluxassociated symptoms. Most children present with edema, which can range from mild periorbital edema to scrotal or labial edema to widespread edema. Abnormal corneal and gag reflexes indicate significant brainstem injury. Prolonged aPTT (in mild form, aPTT may be normal) 2. Alcohols (e.g., ethanol) and hydrocarbons (e.g., gasoline, paint thinner, furniture polish) h. This patient is likely to have prolonged postictal periods. Most children have disease involving the terminal ileum. Patients with ataxia telangiectasia will develop malignancy, the majority of which are lymphomas (question 109). Inactivated influenza vaccine (IIV), administered intramuscularly a. Table 10-4 Differentiation of Ulcerative Colitis and Crohn Disease Characteristic Location Ulcerative Colitis Colon Rectal bleeding Rectal disease Perianal disease (skin tags, fistulas, fissures, abscesses) Lesions Frequent Almost always Rare Crohn Disease Any segment of gastrointestinal tract, especially terminal ileum Occasional Occasional Common Contiguous lesions Skip lesions 386 Transmural involvement Extraintestinal manifestations Complications Risk of colon cancer No Uveitis, arthropathy, pyoderma gangrenosum, sclerosing cholangitis Toxic megacolon Significantly increased FTT = failure to thrive. Physical examination may reveal rales, rhonchi, and wheezes. Measure the patient's upper-to-lower (U/L) body segment ratio. Which of the following is the most likely explanation for his hematuria? Gilbert syndrome, in which 50% of enzyme activity is absent. Dietary proteins are broken down into amino acids or oligopeptides by pepsinogen and pancreatic proteases in the proximal small intestine. Sodium benzoate is a scavenger agent that would be useful to facilitate ammonia excretion, but this patient does not have hyperammonemia. A fracture that is displaced will very likely require surgical reduction and pinning, rather than casting alone. Concise descriptions, clinical correlations, numerous figures, and nearly 100 tables detail the essentials of pediatrics, accompanied by case-based review questions at the end of each chapter and a comprehensive end-of-book exam to build test-taking confidence and help your students excel in clerkship. Surgical intervention, and placement of a ventriculoperitoneal shunt, is only required if there is concomitant obstructive hydrocephalus, which is not present in this case. This patient's urinary dipstick for blood may be falsely positive if he has recently ingested ascorbic acid (vitamin C). Small bowel transplantation has been performed but is reserved for patients with coexisting life-threatening TPN-associated liver disease. Environmental exposures (e.g., in utero exposure to tobacco and alcohol) b. Malabsorption is the inadequate absorption of nutrients and is most often characterized by diarrhea, abdominal distension, and impaired growth. Intermittent asthma only needs a short-acting inhaled β2agonist for symptom treatment without need for a long-term controller. Categories of jaundice a. If diagnosed with poststreptococcal glomerulonephritis, this patient would be expected to have mild to moderate impairment of renal function and normal serum complement levels. For children with an abnormal result on the screen, a fasting lipid profile should be obtained. Antibiotics are often prescribed to empirically treat STIs. 703 VII. The boy has developed ataxia, tremors, and seizures. Presenting features include the acute onset of pallor, jaundice, hemoglobinuria, and splenomegaly. Other risk factors include sleeping in the prone position and overheating. Timing of the vaccination: Every child aged 6 months and older should receive the influenza vaccination annually. 140 VI. Ulcers may also develop in children with H. The answer is B [, section III.C.11]. Genital ambiguity is variable. Information about the environment should include location of victim when discovered, and medications, plants, vitamins, herbs, and chemicals in the home. Acholic or light stools 3. The EEG shows the characteristic generalized 3-Hz spike-and-wave discharges. Ceruloplasmin 3. Crohn Disease (Table 10-4) a. It is nonpruritic and evanescent (comes and goes) and tends to be more prominent during febrile episodes. 474 VII. Some cases of aqueductal stenosis are inherited as an X-linked trait, and these patients may have thumb abnormalities and other CNS anomalies such as spina bifida (SB). Other pertinent findings include bilateral nonpurulent conjunctivitis; red, cracked lips; swollen indurated fingers with erythematous palms; and an erythematous macular rash on the trunk. Lipids 1. Infants may require assisted ventilation and gastrostomy tube feedings. This patient's age of presentation is atypical; the peak age of presentation of nephrotic syndrome is between the ages 5 and 15 years. On examination, you note that the calf muscles are enlarged. 113. The murmur described in the vignette is a venous hum that results in a vibratory hum throughout the cardiac cycle as blood returns from the head through the internal jugular vein. Anemia is lifelong. Clinical significance. Osteomyelitis is an infection of the bone. Management may include corticosteroid therapy. Neurologic findings include changes in intellect and behavior, dystonia, dysarthria, basal ganglia symptoms (Parkinson-like), seizures and dementia. FIGURE 10.1 Etiology of acute abdominal pain during childhood. Year: 2019 Page Number: 540 File Type: PDF File Size: 15.49 MB Authors/ Editors: Lloyd J. On examination, you note that the neonate's hands are clenched with overlapping digits and her lower extremities are extended and crossed. Signs and symptoms include bruising, petechiae, pallor, and fatigue, or serious infection as a result of neutropenia. Congenital causes of hydrocephalus a. Fluids, Electrolytes, and Dehydration II. Classification of seizures. Evaluation includes measurement of parental head circumferences, and a careful physical examination that includes observation for split cranial sutures, bulging anterior fontanelle, irritability, or vomiting, all of which may suggest elevated intracranial pressure. Consider poisoning in patients presenting with nonspecific signs and symptoms, such as seizures, severe vomiting and diarrhea, dysrhythmias, altered mental status or abnormal behaviors, shock, trauma, or unexplained metabolic acidosis. Scoliometer is used to quantify the degree of curvature on the forward bend test. Germ cell tumors may be located in the gonadal region (i.e., testis, ovary) or in extragonadal regions (i.e., anterior mediastinum, sacrococcygeal area, pineal gland or suprasellar region of the brain, retroperitoneum, neck). Hearing screening 1. May be administered either via multiple daily injections (MDI) or insulin pumps. Table 4-4 Clinical Problems of Small-for-Gestational-Age Infants 135 Perinatal asphyxia Hypothermia Hypoglycemia Polycythemia Thrombocytopenia Hypocalcemia Meconium aspiration syndrome Intrauterine fetal death Hypermagnesemia (if the mother is treated with magnesium for hypertension or preterm labor) 136 IV. This leads to platelet thrombi formation and renal ischemia from the microthrombi. Screening assessments include a complete history and physical examination, growth measurements, blood pressure measurements, strabismus and vision screening, hearing screening, tuberculosis risk assessment, and laboratory screening. Avascular necrosis and collapse or deformity of the femoral head 2. Well-Child Screening A. Dehydration, hyponatremia, and hypokalemia may all occur if resorptive capacity is limited. Defects in the urea cycle result in accumulation of ammonia, which is toxic, especially to the nervous system. Boys present with premature adrenarche, rapid growth, and premature acne. ORT is based on the principle that the intestinal absorption of sodium and other electrolytes is enhanced by the active absorption of glucose (coupled cotransport mechanism). Cyanosis A. Factor VIII deficiency—hemophilia A a. To be absorbed, dietary vitamin B12 must first combine with a glycoprotein (intrinsic factor) secreted by the gastric parietal cells. Behavioral abnormalities including autism and attention deficit/hyperactivity disorder 2. Diagnosis is based on the presence of arthritis and psoriasis. The 4-month-old infant has spinal muscular atrophy type 1 (Werdnig–Hoffman disease), an autosomal recessive disorder that presents with hypotonia, weakness, problems with suck and swallow, and tongue fasciculations. Management would therefore include immediate endotracheal intubation. 395 b. Hemophilia A represents a defect in factor VIII procoagulant activity (antihemophilic factor; factor VIII protein). The emergency phase is the same for all patients, regardless of the patient's initial serum sodium level. Weakness and hypotonia may be present, but deep tendon reflexes are preserved. Abuse should always be considered in any childhood injury. Alport syndrome is a form of progressive hereditary nephritis that is secondary to defects in the side chains of type IV collagen within the glomerular basement membrane. Myelomeningocele is more common than meningocele. Note that most adrenal tumors are virilizing, but on occasion, they may also feminize. Lysosomal Storage Diseases X. The diverticulum contains ectopic gastric mucosa that produces acid. Boys: presence of testicular changes, penile enlargement, or pubic or axillary hair before 9 years of age 2. A patent ductus arteriosus (PDA) would be a continuous machinelike murmur, which should not be affected by position. In others, continuous nasogastric or nasojejunal feeds may be used successfully. Brown, MD, FAAP, Regional Chair of Pediatrics, Palo Alto Division, Palo Alto Medical Foundation, Palo Alto, California, Adjunct Clinical Associate Professor of Pediatrics, Stanford School of Medicine, Stanford, California Ryan J. Laboratory confirmation of GI bleeding. Often presents by 3–4 months of age with hypoglycemia, lactic acidosis, hepatomegaly, hyperuricemia, hyperlipidemia, short stature, and cherubic facies. DTRs are preserved. Diffuse lymphadenopathy is present, and the liver is palpable 4 cm below the right costal margin. A description should include whether there is a break in the skin (open versus closed), the spatial relation of the fractured ends (displaced versus angulated), the type of fracture, and the location of the fracture. Feeding difficulty is observed immediately after birth. Each individual disorder may have several subtypes that can vary in level of severity. Terms used include the suffix -cytic, referring to size, and the suffix -chromic, referring to color. This disorder may also cause vomiting, and it is described in Chapter 4, section XII.D.5. 375 V. High concentrations of oxygen play a major role in the development of ROP. Turner syndrome (see also Chapter 5, section IV.C.1). 5NT is more specific than GGTP for biliary tract damage. Which of the following statements regarding this patient's condition is correct? Common causes include hemolytic anemias (premature destruction of RBCs), some RBC aplasias, and sickle cell (SS) anemia. Clinical features of physiologic reflux (GER) 1. Although pneumonia as a result of Staphylococcus aureus may occur in an adolescent, the patient would be more acutely ill. Occult blood loss with resultant iron deficiency may be secondary to polyps, Meckel diverticulum, inflammatory bowel disease (IBD), peptic ulcer disease, celiac disease, and the early ingestion of whole cow's milk before 1 year of age. Lumbar puncture (LP) to rule out meningoencephalitis should be considered if the CT scan is negative. Treatment also includes bed rest and analgesics. These tumors are 209 found in the chest, pineal gland, gonad, or liver (hepatoblastoma). The patient then has high blood glucose in the morning. One percent of children have a single unprovoked seizure before 16 years of age. ≥50,000–100,000 colonies of a single organism in urine collected by clean-catch technique 4. The term protein–energy malnutrition is used to describe this state. Cranial nerve involvement. Williams syndrome E. Girls present with premature adrenarche, clitoromegaly, acne, rapid growth, hirsutism, and infertility. Metatarsus adductus is typically identified in children younger than 1 year, and femoral anteversion is typically identified in children older than 3 years. Individuals are usually 5–10 years of age. An infected tick must be attached to the skin for at least 36–48 hours before there is a significant risk of developing this condition. Age of onset is usually during peak linear growth in adolescence. The acute form responds well to steroids. LGA infants have increased risk of birth trauma (such as shoulder dystocia, brachial plexus injury, and clavicular fracture), hypoglycemia, polycythemia, and perinatal asphyxia. Anesthesiology and otolaryngology should be consulted to visualize his airway and intubate him in a controlled environment (e.g., operating room). Onset is between 8 and 13 years of age. Pupils may also be dilated during and immediately after a seizure or after topical ophthalmic application of a dilating agent, but the vestibular nerve is usually unaffected in these situations (the oculocephalic maneuver would be positive). Salicylates directly stimulate respiratory centers. The mother of a 6-year-old boy brings her son to the office for a second opinion regarding her child's developmental delay. Inspissated bile syndrome is associated with hemolysis (e.g., ABO incompatibility) or with a very large hematoma. Developmental delay e. Thelarche is the onset of breast development as a result of the release of estrogen, and adrenarche is the onset of pubic or axillary hair development as a result of the release of adrenal androgens. Renal biopsy is rarely indicated in the young child with typical NS, unless the creatinine clearance is impaired or initial management with corticosteroids is ineffective. Liver inflammation associated with viral hepatitis is caused by either hepatotropic viruses discussed below (HAV, HBV, hepatitis C virus [HCV], hepatitis delta virus [HDV], and hepatitis E virus [HEV]) or by other viruses that cause liver inflammation as part of a more widespread disease process (e.g., EBV, varicella-zoster virus, HIV, herpes simplex virus). Diagnosis is suggested by radiographic findings; however, similar findings are found in osteomyelitis, lymphoma, osteogenic sarcoma, and Langerhans cell histiocytosis (LCH). Cyanosis is the bluish discoloration of the skin and mucous membranes that is directly related to the absolute concentration of deoxygenated hemoglobin (more than 3 g/dL of deoxygenated, reduced Hgb in the capillary blood). The answer is D [I.A, I.C.1–2, and I.E] Henoch–Schönlein purpura is an IgA-mediated vasculitis that involves the skin, joints, gastrointestinal tract, and kidneys. Clinical features are more variable than in hereditary spherocytosis. Incidence has increased as a result of recommendations that infants sleep on their backs to decrease the risk of SIDS. Limited range of motion with passive lateral neck flexion or chin rotation may be present. Curve is visible when standing. Studies show severe anemia, spherocytes on blood smear, prominent reticulocytosis, and leukocytosis. Pain may be referred to the knee and to the groin. This patient's physical characteristics, along with learning problems and attention deficit/hyperactivity disorder, are consistent with fetal alcohol syndrome. The answer is B [II.C and II.D.4]. The answer is E [VII.C.3]. Facial weakness occurs in 40–50% of patients and is often bilateral. Screening tests to detect asymptomatic diseases (e.g., vision, hearing, newborn metabolic screening, anemia, and lead screening) c. The focus of each well-child visit is to identify undetected problems or the risks for such problems. It results in persistent 152 contraction of a distal segment of colon, causing obstruction with proximal dilatation. Other autoimmune disorders, including juvenile rheumatoid arthritis, diabetes mellitus, and thyroid disease, may coexist. Types of diabetes 1. Amniotocele is swelling of the nasolacrimal sac. Influenza vaccines a. Table 12-4 Differential Diagnosis of Cerebellar Ataxia Brain tumors Trauma Toxins Vascular Infections Pilocytic astrocytoma (occurring in the cerebellum) Medulloblastoma (occurring in posterior fossa) Neuroblastoma (opsoclonus myoclonus ataxia syndrome) Cerebellar contusion Subdural hematoma Ethanol Antiepileptic medications Cerebellar infarction or hemorrhage Meningitis 476 Inflammatory Demyelination Migraine syndromes Encephalitis Acute cerebellar ataxia of childhood Acute disseminated encephalomyelitis (ADEM) Multiple sclerosis Basilar migraines and familial hemiplegic migraines Benign paroxysmal vertigo (may have history of migraine) 477 VIII. The triad is low-set hairline, short neck, and limited neck motion. DDH is asymptomatic in infants. Current research is evaluating gene replacement therapy to convert DMD to the more favorable BMD. NSAIDs are useful in the treatment of minor inflammatory symptoms, such as myalgias and arthralgias. Emphasis is placed on identification and correction of triggers, creation of a regular toilet schedule, removal of negative reinforcements, and creation of a positive environment. Trisomy 13 B. If the contrast enema fails to reduce the intussusception, or if the child has signs of peritonitis or pneumoperitoneum, operative reduction is indicated. Subsequent neurologic complications are not common but may include aseptic meningitis, facial cranial nerve palsy, and encephalitis. A variety of different prenatal, perinatal, and postnatal insults to the central nervous system may result in infantile spasms. Treatment includes ophthalmologic consultation, pain control, prevention of vomiting (which may cause a sudden increase in intraocular pressure) and bed rest for at least 5 days. Thyroid imaging may be useful once the diagnosis is confirmed. Hgb values reach adult levels after puberty. Reduced plasma oncotic pressure induces increased hepatic production of plasma proteins, including lipoproteins. Throbbing or pounding pain suggests migraine headaches, whereas a sensation of squeezing or pressure is more common in tension-type headaches. C. The oligohydramnios may have resulted from bilateral renal agenesis, other urinary tract defects, or a chronic leak of amniotic fluid. A clean "bagged" urine sample is adequate for culture in this febrile infant with no obvious source of infection. Recurrent hemarthroses e. Oral rehydration salt (ORS) solutions are balanced mixtures of glucose and electrolytes for use in treating and preventing dehydration, potassium depletion, and base deficits caused by diarrhea. Acne Vulgaris IX. AP and frog-leg lateral radiographs of the pelvis confirm the slipped epiphysis. Acquired clotting factor disorders 1. Steroids are contraindicated. The source of the botulinum toxin is infected foods, such as contaminated honey, or spores unearthed from the ground. Binocular cortical connections are present at birth. Acute cerebellar ataxia is an unsteady gait secondary to a presumed autoimmune or postinfectious cause. Hepatocellular enzymes a. In some patients, there is a genetic predisposition. Tdap is recommended at age 11–12. The pH of the esophagus is continuously monitored for at least 18 hours. However, recovery may take weeks or even months. The face, hands, and arms are mainly affected, and the movements appear continuous, quick, and random. Nutrition A. Renal biopsy (in some cases) D. Upper limb malformations 5. 411 FIGURE 11-1 An approach to red urine. Poor prognostic signs for renal recovery include a high white blood cell (WBC) count on admission and prolonged oliguria. Increasingly common in the pediatric age group, especially after the age of 10, based on epidemiologic studies. Because there are only two β-globin genes in each cell, there are only two states: 1. These all may damage bone marrow stem cells directly or may induce autoimmune destruction. Table 17-1 Management of Adolescent Idiopathic Scoliosis Scoliosis Curvature 10°–25° Skeletally Immature Skeletally Mature Before or During Growth Growth Completed Follow-up scoliosis films 6 months after diagnosis No treatment required ≥7° progression—refer to orthopedic surgeon 25°–40° >40° Bracing indicated No treatment required Surgery may be indicated Surgery may be indicated if scoliosis > 50° Table 17-2 Common Causes of Back Pain in Children and Distinguishing Features Key features Etiology Back Strain Most common cause of back pain Spondylolysis Repetitive hyperextension of the spine Spondylolisthesis Complication of spondylolysis Overuse or poor body mechanics Stress fracture in pars interarticularis Anterior subluxation of the vertebral body after 616 Discitis Staphyloccocus aureus is most common Infection or inflammation of Diagnosis History and physicalNormal examination with possible tenderness of paraspinal muscles Localized painWorse with hyperextensionAP, lateral, oblique radiographs of lumbar spine confirm diagnosis spondylolysis Same as spondylolysis intervertebral disk Tenderness over involved disc Refusal to bend forward Elevated ESRBone scan/MRI confirm diagnosis Management Rest, stretching, and analgesics Rest, analgesicsBracing to immobilize if pain is severe Same as spondylolysisSurgery if signs of nerve impingement or subluxation >50% of vertebral width Antistaphylococcal antibiotics Rest, analgesics AP = anteroposterior; ESR = erythrocyte sedimentation rate; MRI = magnetic resonance imaging. Patients present with fever, abdominal distension, and septic shock and are at risk for perforation and hemorrhage. Bilateral dilated nonreactive pupils suggest topical application of a dilating agent, a postictal state, or irreversible brainstem injury. Activated charcoal is effective and may be readministered every 4 hours in severe poisonings. Urine culture grows >100,000 colonies/mL of Escherichia coli. Cough and cold preparations d. Kasai portoenterostomy (Roux-en-Y intestinal loop attached directly to the porta hepatis) is the treatment of choice to establish bile flow; however, its success diminishes rapidly with increasing patient age at presentation. Etiology 507 1. PPP or heterosexual gonadotropin-independent puberty 1. 7th Ed. Philadelphia: Lippincott Williams & Wilkins, 2013. Many of these signs are nonspecific responses of the newborn to serious illness. Infantile botulism 2. Patients with hypernatremic dehydration generally have their fluid and electrolyte deficits replaced more slowly, usually over 48 hours, to minimize the risk of cerebral edema that may accompany rapid fluid correction. Age of onset is typically 4–7 months. Diagnosis is made by history and the presence of a normal neurologic examination. ABO hemolytic disease occurs when the mother is blood group O and her fetus is blood group A, B, or AB. A seizure is a transient occurrence of signs and/or symptoms due to abnormal excessive or synchronous neuronal activity in the brain. Alport syndrome (question 14) is inherited in multiple patterns, but the X-linked form is by far the most common. Chronic abdominal pain (CAP) is defined as abdominal pain that occurs each month for at least three consecutive months. The lumbar region is most commonly affected. Progressive hearing loss in adulthood I. Seizures. Allergen immunotherapy may also be beneficial but would not be the initial step in management. Myasthenia gravis D. Contrast enema (barium or Gastrografin) can also be used to both establish the diagnosis and to reduce the intussusception. Increasing head circumference and signs or symptoms of increased ICP mandate an urgent head ultrasound in infants or head CT scan in older children. Microcytic, hypochromic anemias. UC usually begins in the rectum and extends proximally in a contiguous fashion. Disorders of Puberty 4 III. Antistreptolysin-O titers would be expected to be abnormally high in 25% of patients with this condition. In severely hypothermic patients, resuscitation should continue until the patient is rewarmed to 32°C (89.6°F). Parenteral rehydration should occur in two phases: 1. Connective tissue disorders 1. Essential fatty acids play an important role in infant brain development. Table 11-3 Etiologies of Acute Kidney Injury (AKI) Categories of AKI Causes of Renal Failure Specific Examples Prerenal Caused by a reversible ↓ in renal perfusion that leads to a ↓ in GFR ↑ Urine SG ≥ 1.030 Urine osmolality > 500 Urine Na+ < 20 * FeNa < 1% in older children, 20 Hematuria hypoperfusionDrugs (semisynthetic penicillins) Eosinophilia, eosinophiluria Postrenal Tumor Ureterocele Urethral trauma Neurogenic bladder Posterior urethral valves in males Vascular neonates Obstruction of urine flow from either a solitary kidney, from both kidneys, or from the urethra Stones Dilation of renal collecting system on renal ultrasound ↓ Perfusion of the kidneys Renal artery embolus (especially in the presence of an umbilical artery catheter) ↓ Renal blood flow on nuclear renal scan Renal vein thrombosis, presenting with sudden-onset gross hematuria and a unilateral or bilateral flank mass, with ↑ incidence in infants of diabetic mothers * HUS = hemolytic uremic syndrome; PSGN = poststreptococcal glomerulonephritis; FeNa = fractional excretion of sodium; SG = specific gravity; GFR = glomerular filtration rate; Creat = creatinine; BUN = blood urea nitrogen. Sydenham chorea occurs secondary to antibodies that cross-react with membrane antigens on both group A β-hemolytic streptococcus and basal ganglia cells. Activity should be restricted during antibiotic therapy to prevent a fracture, especially if the diagnosis of osteomyelitis was initially delayed. Signs and symptoms include a solid, firm, painless testicular mass or generalized testicular swelling. The diagnosis of poststreptococcal glomerulonephritis is suspected pending further evaluation. Two categories of disorders may result in delayed puberty. Shiga-like toxin–associated HUS (STEC-HUS) 1. Secondary headaches are caused by an underlying disease process such as increased intracranial pressure (e.g., hydrocephalus), meningeal irritation (e.g., meningitis, subarachnoid hemorrhage), or inflammatory processes. Classification is made on the basis of the mean corpuscular volume (MCV) and the morphologic appearance of the RBC (i.e., size, color, and shape). Many other infectious agents have been associated with Guillain–Barré syndrome, such as Mycoplasma pneumoniae, cytomegalovirus, EBV, herpes zoster virus, influenza, varicella, and coxsackievirus. Key point: Patients with poor growth velocity with normal screening laboratory results, but low IGF-1 and delayed bone age, should have an additional workup for GH deficiency (i.e., GH stimulation testing). 80–90% of cases are sporadic and 10–20% are familial or part of a genetic syndrome (e.g., Crouzon and Apert syndromes). SABA = short-acting beta-2 agonist; ICS = inhaled corticosteroid; LABA = long-acting inhaled beta-2 agonist; LTRA = leukotriene receptor antagonist. Inflammation is diffuse, limited to the mucosa, and localized to the colon. Diagnostic workup varies with the type of CAH (see Figure 6-5): a. Standing posterior–anterior (PA) films of the spine confirm the curvature and are used to calculate the Cobb angle (Figure 17-2). Glucose should be checked immediately in any comatose patient. Treatment includes rest and analgesics for pain. Microcephaly is usually associated with developmental delay and intellectual impairment. In older children, formal acuity testing is the best screening test. Impairment of T-lymphocyte function increases risk for severe primary infection, disseminated disease, or relapse of disease. 540 6. Management includes enzyme replacement therapy. Multicystic renal dysplasia. Probably why I passed my pediatrics rotation in medical school. Migraines associated with focal neurologic signs 1. Hip pain may be acute or insidious in onset. Most protein intolerance is transitory and resolves by 1–2 years of age. Patients with Chiari type II malformation may have low normal intelligence and language disorders. Mitral regurgitation is the most common early valvular manifestation. Previously healthy infants or children may present with sudden onset of crampy or colicky abdominal pain. Serum amylase levels rise within hours of the onset of pain and remain elevated for 4–5 days. Generally, all patients recover. Neurologic examination is notable for decreased muscle tone and a weak suck. Hemorrhagic disease of the newborn is characterized by serious bleeding in the early and late forms, but classic disease generally presents only with cutaneous bleeding, hematemesis, and bleeding from the circumcision site or umbilical cord. Intestinal obstruction may present with high-pitched bowel sounds, abdominal distension, tenderness, and at times, visible peristalsis. Symptoms may last from several months to 2 years. Rebleeding 3–5 days after initial injury can occur due to repeat trauma or an underlying bleeding diathesis, or as the clot retracts. Table Of Contents: Here is the list of Chapters of boards review series pediatrics: Pediatric health supervision. Six-year-old boy with coarsened facial features, stiff joints, and a cloudy cornea 2. In addition, rare but important causes of macrocytic anemia include bone marrow failure syndromes [e.g., Fanconi anemia, see section II.B.1]. In the United States, most human infections occur during feeding by an infected deer tick, Ixodes scapularis. Absence epilepsy of childhood is inherited in an autosomal dominant pattern with age-dependent penetrance. Wilson disease may cause acute and chronic hepatitis and liver failure. The disease is more common in Caucasians and Asians. Type III: absence of vWf; the most severe type c. The answer is B [IV.C.6]. There is no known association between Guillain–Barré syndrome and prior group A β-hemolytic streptococcal infection. Hydrocephalus (see Chapter 12, section II) g. WBI should be considered for life-threatening ingestion. There are characteristic morphologic features and immunohistochemical markings. Timing of vaccination: In the United States, only IPV is recommended and is given at 2 and 4 months of age, with boosters at 6–18 months and 4–6 years of age. Social history is critical because children who live in neglected or hostile environments may exhibit short stature because of psychosocial deprivation. Rheumatic Fever A. Additional causes include Beckwith– Wiedemann syndrome, Prader–Willi syndrome (see Chapter 5, section IV.E.2), and congenital hyperinsulinism (nesidioblastosis, with diffuse proliferation and dysfunction of pancreatic islet cells). However, a lead point is identified in only 5% of cases and is more common in older children. After 10–14 days of infection, a plain radiograph begins to reveal elevation of the periosteum, suggesting osteomyelitis.

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