Pediatrics Assessment Exam
Answers and Explanations

ANSWER KEY

1. D
2. B
3. E
4. E
5. D
6. C
7. C
8. D
9. B
10. D
11. E
12. C
13. D
14. E
15. B
16. B
17. C
18. B
19. A
20. D
21. E
22. F
23. D
24. B
25. B
26. E
27. D
28. B
29. C
30. C
31. D
32. D
33. D
34. A
35. A
36. E
37. C
38. D
39. A
40. B
41. E
42. C
43. D
44. C
45. E
46. C
47. A
48. D
49. C
50. B

1. **The correct answer is D.** Nearly one third of children may have an innocent murmur during routine examination. Such murmurs are not associated with hemodynamic abnormalities or cardiac defects. They arise as a result of normal turbulence of blood in the chambers of the heart or large vessels during childhood. Innocent murmurs are characterized by their character (short systolic), low intensity (grade I or II), and absence of any other abnormal physical signs. No further investigation is required in such children, and parents can be reassured regarding the benign nature of the finding.

   A chest x-ray (**choice A**) is useful to evaluate heart size when cardiac failure is suspected. It also can show abnormal cardiac shape in certain congenital cardiac lesions. It is not required in the case presented in the vignette.

   Children with anemia have hyperdynamic circulation and can demonstrate cardiac flow murmurs. In an otherwise healthy child, however, the presence of murmur alone does not require obtaining a hematocrit (**choice B**). Instead, hematocrit should be performed at 1 year of age and before school entry as part of the routine health maintenance.

   Cardiac disease can be suspected from history of tiredness, breathlessness, or growth failure. In addition, there are features in physical examination that can help to identify children with heart murmur who have a cardiac disease. These include a pansystolic murmur, very loud murmurs (grade III or greater), presence of click, or an abnormal second heart sound. In the absence of any of these features, an electrocardiogram (**choice C**) or echocardiogram (**choice E**) is not required.

2. **The correct answer is B.** This patient has erythema migrans, a manifestation of Lyme disease. Erythema migrans develops 3 to 32 days after the bite of a deer tick (*Ixodes scapularis*) infected by *Borrelia burgdorferi*. Prompt removal of the attached tick within the first 8 to 12 hours minimizes the chance of transmission of any of these diseases, but appropriate followup blood work should be done to evaluate if anti-*Borrelia* antibody titers are increasing and treatment with antibiotics is necessary. Commonly used antibiotics include amoxicillin, doxycycline, cefuroxime, erythromycin, and azithromycin. In the United States, the most common vector of Lyme disease is *Ixodes scapularis*.

   Intramuscular triamcinolone (**choice A**) is not indicated in the treatment of erythema migrans, which is a bacterial infection.

   Oral doxycycline (**choice C**) is one of the antibiotics commonly used for the treatment of erythema migrans. The age of the patient is a contraindication for the use of a tetracycline antibiotic, however, because it may cause permanent staining of teeth if used in children younger than 8 years of age.

   Topical hydrocortisone (**choice D**) is of no benefit in the treatment of erythema migrans and should not be used, as it may mask the presence of infection through its local anti-inflammatory and vasoconstrictive properties.

   Topical ketoconazole (**choice E**) is an antifungal medication often used to treat fungal infections of the skin. Because the potassium hydroxide preparation of skin scrapings performed in this patient was negative, the described disease is not a fungal infection.

3. **The correct answer is E.** Asthma is a commonly encountered problem in the emergency room. Patients can present with varying degrees of severity and require interventions based upon the initial assessment. In the child presented in this vignette, all the interventions mentioned in the choices are appropriate. However, the presence of severe distress and low oxygen saturation mandate immediate provision of inhaled oxygen to relieve hypoxia. This should be done even before a complete history can be obtained.

   Nebulized albuterol (**choice A**) is the appropriate first drug in patients with an acute attack of asthma. If relief is incomplete, the dose should be repeated immediately. The patient should be closely monitored for deterioration while inhaled therapy is being administered.

   Chest x-ray (**choice B**) is not indicated in most children with previously diagnosed asthma who present with worsening symptoms. Chest x-ray is appropriate if the acute attack is unusually severe, if pneumonia is suspected, or if breath sounds are significantly decreased on one side.

   Some children presenting with status asthmaticus may be dehydrated due to tachypnea and decreased intake of fluids. Thus, it is appropriate to start intravenous fluids (**choice C**) in such patients once the respiratory status is stabilized.

   Steroids (**choice D**) are extremely important and should be instituted early in the management of status asthmaticus. If the child has been vomiting or is in severe distress, then parenteral methylprednisolone can be used.

4. **The correct answer is E.** This patient has cat-scratch disease, a self-limited infection caused by *Bartonella henselae*. Symptoms can vary from mild to severe and may include malaise, anorexia, fever, and headache. Three quarters of cases occur between September and March. Most patients give a history of exposure to a cat, although other small animals also may be the reservoir.

Within 3 to 10 days, a small papule, pustule, or vesicle appears at the site of injury. A week later, edema and tenderness develop in regional lymph nodes. Untreated, the disease resolves spontaneously in 2 to 5 months except in severely immunocompromised individuals such as those with AIDS. Diagnosis is established through clinical presentation, increasing titers of antibodies, or histopathologic evaluation of tissue stained with the Warthin-Starry technique. Treatment is supportive, except in immunosuppressed individuals who require antibiotics.

A nasopharyngeal swab for culture (**choice A**) would not help to establish the diagnosis of cat-scratch disease because the microorganisms are not present in nasopharyngeal secretions. Infection is the result of direct inoculation into the skin.

Needle biopsy of the lymph nodes (**choice B**) is not indicated, because the diagnosis can be established based on clinical presentation and increasing serum antibody titers to *Borrelia henselae*.

Although the diagnosis of cat-scratch disease most commonly is made based on history of contact with an animal and clinical presentation, it should be confirmed either by increasing serum antibody titers or specially stained tissue sections from biopsy of a skin lesion. It is therefore incorrect that no further studies are indicated (**choice C**).

Radiography of the head and neck (**choice D**) would be noncontributory in establishing the diagnosis, as it would show only soft tissue swelling at the site of the enlarged lymph nodes.

5. **The correct answer is D.** This patient has the typical clinical presentation of erythema infectiosum (or Fifth disease) caused by parvovirus B19, a DNA virus. Erythema infectiosum is a benign, self-limited exanthematous illness. Humans are the only known host and they transmit the virus by way of respiratory secretions and blood. The disease is seen most commonly in spring and has an incubation period that ranges from 4 to 28 days. The patient usually presents with mild systemic symptoms, including low-grade fever, headache, and upper respiratory tract symptoms such as pharyngitis. Some patients (mostly adults and adolescents) may have arthritic symptoms. Clinically, an intensely red "slapped cheek" appearance is followed by a lacy, reticulated maculopapular rash on the trunk and proximal extremities. The palms and soles are not affected. The rash lasts anywhere from 2 to 40 days (average, 11) but has a tendency to recur with exposure to heat or physical exertion for a period of several weeks. The patient is not contagious once the rash appears. Diagnosis of erythema infectiosum is usually made on clinical grounds, as routine laboratory studies are not widely available for detection of parvovirus B19. Detection of viral DNA in fetal blood aids in making a diagnosis of B19-induced fetal hydrops. Treatment consists of supportive care only.

   Coxsackievirus A16 (**choice A**) is the cause of hand-foot-and-mouth disease, a viral illness that presents clinically with characteristic oral and distal extremity lesions. The incubation period is 3 to 6 days, after which a flu-like prodrome ensues and lasts 2 to 3 days. Oral lesions begin as erythematous macules that evolve rapidly into vesicles and then ulcerate. They are painful and interfere with eating. Cutaneous lesions in the form of asymptomatic vesicles with an oval erythematous halo are typically present on the hands, feet, and buttocks. They tend to follow skin lines and are oval in shape. Coxsackievirus infection is highly contagious. Transmission occurs by way of direct contact with nasal or oral secretions, fecal material, or aerosolized droplets in a fecal-oral or oral-oral route.

   Herpes simplex virus type 1 (**choice B**) is the cause of orolabial herpes simplex ("cold sore"). Although initial infection is usually asymptomatic, a vesicular gingivostomatitis may develop in up to 1% of people, chiefly in children and young adults. By adulthood, almost 100% of the population has antibodies confirming infection, but only approximately 10% develop recurrent episodes of orolabial herpes.

   Human herpesvirus type 6 (**choice C**) causes roseola infantum, a common cause of sudden, unexplained high fever in children 6 to 36 months of age. Prodromal fever may be accompanied by convulsions and lymphadenopathy. Suddenly, on approximately the fourth day, the fever drops and a morbilliform erythematous rash consisting of rose-colored discrete macules appears on the neck, trunk, and buttocks, and sometimes on the face and extremities. Complete resolution occurs in 1 to 2 days.

   Rubella virus (**choice E**) is the cause of rubella, a viral infection that occurs most commonly in adolescents and young adults. The patient presents with retroauricular and posterior occipital lymphadenopathy, pharyngitis, low-grade fever, and upper respiratory infection symptoms. A fleeting, erythematous, maculopapular rash begins on the face and spreads to the body, lasting up to 3 days. Forchheimer spots may be present on the soft palate, aiding in making the clinical diagnosis. The prognosis in children is excellent. If the infection occurs during pregnancy, however, it may lead to the congenital rubella syndrome.

6. **The correct answer is C.** Hemophilia A follows an X-linked recessive inheritance pattern. Thus, the risk that the abnormal X-chromosome gene will be passed from

the mother to the male or female offspring is 50%. However, because males possess a single X-chromosome, all the males who inherit the abnormal gene will develop hemophilia. Thus the risk of *disease* is 50% for male offspring. On the other hand, females possess two X-chromosomes, one from the father and one from the mother. Thus, none of the females who inherit one abnormal gene will develop the disease. Hence, the risk of *disease* is 0% for female offspring, not 25% (**choice A**). Instead, female offspring with one abnormal gene are asymptomatic carriers, like the mother.

The risk of disease among male and female offspring is different in X-linked recessive inheritance (**choice B**), due to the presence of two X-chromosomes in females.

Since fathers do not pass on the X-chromosome to male offspring, the incidence of the disease in males is not affected by paternal disease (**choice D**).

A significant number of patients with hemophilia have a negative family history due to a high rate of spontaneous mutation in the factor VIII gene. In such cases the risk to the second male offspring cannot be ascertained without determining the mother's carrier status. However, due to the fact that one of the maternal uncles has hemophilia A, it is reasonable to presume that the mother is a carrier for hemophilia A (**choice E**). This can be further confirmed by laboratory tests on the mother.

7. **The correct answer is C.** This boy has a typical case of measles (morbilli, rubeola), an infectious disease caused by the Morbillivirus and transmitted through respiratory droplets. It usually occurs in unimmunized preschool children or in high school and college students. Measles is extremely contagious and up to 100% of unimmunized contacts contract the disease. Measles is associated with cough, coryza, conjunctivitis, and fever. Koplik spots are usually present on the buccal mucous membrane opposite the premolar teeth. They appear as bluish-white spots. The exanthem typically develops on or around the fourth day on the forehead and behind the ears and then spreads "like a shower" to the face, trunk, and limbs. At first it is macular, and then dull red papules form with a tendency to coalesce into irregular concentric patterns or become more diffusely confluent. From the sixth to the twentieth day the rash starts fading, leaving behind brownish discoloration and fine scaling. In very severe forms it may become hemorrhagic. Active immunization with the live attenuated vaccine (MMR, measles-mumps-rubella) has reduced the incidence of measles infection.

   Hepatitis B vaccine (**choice A**) prevents infection with the hepatitis B virus. Hepatitis B can cause acute and chronic liver disease. The clinical presentation ranges from subclinical hepatitis to symptomatic hepatitis and, in rare instances, fulminant hepatitis. Long-term complications of hepatitis B include cirrhosis and hepatocellular carcinoma. Perinatal or childhood infection is associated with few or no symptoms, but it has a high risk of becoming chronic.

   Influenza vaccine (**choice B**) is indicated in patients with chronic lung disease, heart disease, immunosuppression, and hemoglobinopathies, and in healthcare workers and patients in nursing homes and long-term care facilities. It is not routinely administered to healthy children. Influenza presents with similar prodromal symptoms as measles, but photophobia, Koplik spots, and skin exanthems are not seen in influenza.

   Meningococcal vaccine (**choice D**) is not a routine school vaccine, although it is recommended or required by some colleges. Indications include anatomic or functional asplenia, patients with complement deficiencies, and travelers to endemic areas. Meningococcal infection presents with meningeal symptoms (e.g., headache, nuchal rigidity, and photophobia) and an increased number of white blood cells in the cerebrospinal fluid (CSF), i.e., pleocytosis.

   Varicella vaccine (**choice E**) is scheduled for all children 12 to 18 months of age and all children who have no history of varicella. The prodrome of fever, headache, malaise, and coryza is followed by a typical intensely pruritic vesicular rash that appears in crops.

8. **The correct response is D.** The term premature adrenarche (or pubarche) refers to growth of pubic hair before the age of 8 years in girls (9 years in boys) without the appearance of any other signs of sexual maturation. Sparse hair grows on labia majora and the mons pubis in girls. The girls generally have height and osseous maturation that are more advanced than their peers. Premature pubarche is a benign condition and does not portend the presence of a serious underlying illness. However, in those cases where pubic hair is accompanied by clitoromegaly or other evidence of masculinization, further endocrine investigation is warranted.

   Variants of congenital adrenal hyperplasia (also known as nonclassical forms) can present with virilization in young girls (**choice A**). However, other evidence of masculinization—such as clitoromegaly, change in voice, or growth acceleration—are generally present.

   Exogenous androgens—for example, in the form of anabolic steroids—can produce hirsutism and clitoromegaly (**choice B**). However, there is no history of ingestion of androgens in the present case.

Onset of secondary sexual characteristics in girls before the age of 8 years is considered to be precocious puberty, and not normal pubertal development (**choice C**).

Girls with Turner syndrome (45,XO) have delayed sexual development (**choice E**). They can present in the teenage years with pubic and axillary hair without breast development. However, short stature is an important hallmark of this syndrome, unlike the case vignette.

9. The correct answer is B. This girl has the typical presentation of roseola infantum (exanthema subitum), a febrile illness that usually occurs in children up to 5 years of age. Human herpesvirus 6 is responsible for this infection, and the incubation period is 5 to 15 days. Infection usually occurs early in life, with peak incidence at 6 to 15 months. The classic presentation is high fever up to 41.0 C (106.0 F) that lasts 3 to 4 days with minimal physical findings that may include rhinorrhea, conjunctivitis, and mild occipital lymphadenopathy. Suddenly, on approximately the fourth day, the fever decreases and a morbilliform erythema consisting of rose-colored discrete macules appears on the neck, trunk, and buttocks, and sometimes on the face and extremities. The mucous membranes are spared. Complete resolution occurs in 1 to 2 days. No diagnostic studies are necessary. The diagnosis is based on the age of the patient, history, and physical findings. Supportive therapy with antipyretics and fluids is sufficient.

Measles (**choice A**) is an infectious disease caused by the Morbillivirus and is transmitted through respiratory droplets. Measles is associated with cough, coryza, conjunctivitis, and fever. Koplik spots are usually present on the buccal mucous membrane opposite the premolar teeth. They appear as bluish-white spots. The exanthem typically develops on or around the fourth day on the forehead and behind the ears and then spreads "like a shower" to the face, trunk, and limbs. At first it is macular, and then dull red papules form with a tendency to coalesce into irregular concentric patterns or become more diffusely confluent. From the sixth to the twentieth day the rash starts fading, leaving behind brownish discoloration and fine scaling.

Rubella (**choice C**) is a viral infection caused by the rubella virus that occurs most commonly in adolescents and young adults. The patient presents with retroauricular and posterior occipital lymphadenopathy, pharyngitis, low-grade fever, and upper respiratory infection symptoms. A fleeting, erythematous, maculopapular rash begins on the face and spreads on the body, lasting up to 3 days. Forchheimer spots may be present on the soft palate, aiding in making the clinical diagnosis. The prognosis in children is excellent. If the infection occurs during pregnancy, however, it may lead to the congenital rubella syndrome.

Scarlet fever (**choice D**) is the result of pyrogenic exotoxins produced by beta hemolytic streptococcus group A. Fever and sore throat precede the skin eruption by several days. The rash begins on the face and neck and spreads downward, sparing palms, soles, and the perioral area (circumoral pallor). Tiny scaly papules appear on the trunk, giving the skin the texture of sandpaper. Fine desquamation ensues from the face and neck down, whereas palms and soles often desquamate in a "gloves and socks" fashion. The erythema is accentuated in skin folds, where petechiae may also occur. The most characteristic mucosal lesion is the strawberry tongue: protruding, enlarged, erythematous papillae and a thick white coating. A diagnosis of scarlet fever may be made clinically and is supported by throat cultures or rapid strep testing.

Varicella (**choice E**), or chickenpox, is caused by the varicella zoster virus. Children between the ages of 5 and 9 years are affected most commonly. Clinically, chickenpox presents as a rash, low-grade fever, and malaise. In the immunocompetent patient, this is usually a benign illness that is associated with lassitude and body temperatures of 37.8 to 39.4 C (100.0 to 103.0 F) of 3 to 5 days' duration. The skin lesions include maculopapules, vesicles, and scabs in various stages of evolution. These lesions, which evolve from maculopapules to vesicles over hours to days, appear on the trunk and face and rapidly spread to involve other areas of the body. Successive crops appear over a 2- to 4-day period. Lesions can be found on the mucosa of the pharynx and the vagina also. Immunocompromised individuals are at greater risk for visceral complications, which occur in 30 to 50% of cases and are fatal 15% of the time.

10. The correct answer is D. This patient has neurofibromatosis, an autosomal dominant disorder that occurs as a result of an abnormality of neural crest differentiation during embryogenesis. The diagnosis of type 1 neurofibromatosis requires two or more of the following criteria:

1) six or more café-au-lait macules of more than 5 mm in greatest diameter in prepubertal individuals and more than 15 mm in diameter in postpubertal individuals;
2) two or more neurofibromas of any type or one plexiform neurofibroma;
3) freckling in the axillary and/or inguinal region;
4) optic glioma;
5) two or more Lisch nodules;

6) a distinctive osseous lesion, such as sphenoid dysplasia or thinning of the long-bone cortex with or without pseudoarthrosis; and
7) a first-degree relative (parent, sibling, or offspring) with the disease.

To make the diagnosis of neurofibromatosis type 2, one of the following is needed:

1) bilateral eighth nerve masses (acoustic neuroma); or
2) a parent, sibling, or child with neurofibromatosis type 2.

There is no specific therapy for neurofibromatosis. Genetic counseling should be done and prenatal testing is available. Therapy should include early identification and treatment of complications. A pediatric ophthalmologist should perform an annual ophthalmology examination.

Jaw cysts (**choice A**) are typically seen in the basal cell nevus (Gorlin) syndrome, in association with multiple basal cell carcinomas of the skin and pitting of the palmoplantar surfaces.

Osteopoikilosis (**choice B**) is characteristic of Goltz syndrome, or focal dermal hypoplasia, an X-linked dominant disorder that affects only females. Linear and whorled, Blaschko line–associated papules and plaques with fat herniation are noted on the skin. Mild to moderate mental retardation is typical and patients may have an associated convulsive disorder.

Osteopathia striata (**choice C**) is a manifestation of Buschke-Ollendorf syndrome, where connective tissue nevi are the hallmark of skin involvement.

"Train track" calcifications of the skull (**choice E**) were historically used in the diagnosis of Sturge-Weber syndrome, a neurophakomatosis characterized by port wine stains of the V1 (±V2, V3) trigeminal area of the face, leptomeningeal vascular anomalies with or without associated seizure disorders, and possible ipsilateral glaucoma.

11. The correct answer is E. The clinical suspicion in this case is vesicoureteral reflux, which is best demonstrated by filling the bladder with dye and doing fluoroscopy while the child voids.

Cystoscopy (**choice A**) as a rule is not indicated for the workup of suspected reflux. It might be needed to further define abnormal anatomy, for instance, an ectopic ureter. It is not, however, the best initial choice for this question.

Intravenous pyelogram (**choice B**) or ultrasound (**choice C**) can demonstrate the presence of dilation of the urinary tract, which might have been produced by vesicoureteral reflux. The reflux itself, however, is best demonstrated with the voiding cystourethrogram. Because it is so safe, ultrasound often is used to begin urologic workups in children. Had the question asked for the standard next step in management, ultrasound might have been a reasonable answer.

Retrograde urethrogram (**choice D**) is used to evaluate urethral injuries in male patients with pelvic fracture and blood at the meatus.

12. The correct answer is C. This patient has rubella (German measles), a viral infection characterized by a maculopapular exanthem and tender lymphadenopathy of the posterior occipital, retroauricular, and cervical lymph nodes. It is seen infrequently because of widespread vaccination efforts. The incubation period is 14 to 21 days. Patients are contagious 2 days before the rash begins and 5 to 7 days after the rash. Clinically, pharyngitis, low-grade fever, and upper respiratory tract infection symptoms may be the presenting sign or rubella may begin with tender lymphadenopathy. The exanthem begins on the face and rapidly spreads to the entire body in the form of erythematous macules and papules that resolve in 3 days. Retroauricular, posterior cervical, and postoccipital lymph nodes are characteristically enlarged and slightly tender. Forchheimer spots are rose spots seen on the soft palate that may precede the skin rash. Adolescent females may have polyarthritis. The diagnosis usually is made clinically from the history and physical examination but should be confirmed by serologic tests or a viral culture. Supportive treatment is given. The prognosis of rubella in children is excellent and complications are uncommon. A pregnant woman who contracts rubella in the first trimester of pregnancy, however, has a high probability that intrauterine transmission will give rise to the congenital rubella syndrome.

His 4-month-old niece (**choice A**) is highly unlikely to contract rubella because of her very young age. Rubella most commonly affects adolescents and young adults.

His 15-year-old sibling (**choice B**), although likely to contract rubella because of his age, has an excellent prognosis and complications would be uncommon.

His 60-year-old grandmother with systemic lupus erythematosus (**choice D**) and his 75-year-old grandfather with diabetes mellitus (**choice E**) are highly unlikely to contract rubella because of their age. The accompanying systemic diseases (i.e., diabetes mellitus and systemic lupus erythematosus) do not carry an increased risk for infection with this virus, nor are they a known risk factor for complications.

13. The correct answer is D. Osgood-Schlatter disease is a traction apophysitis of the tibial tubercle near the insertion of the patellar tendon. The condition is most common in boys during late childhood or adolescence, especially in athletes. Diagnosis is based on characteristic presentation and examination findings. An x-ray is useful to exclude other conditions. The only treatment required is a restriction or decrease in physical activity. Complete resolution occurs when tibial tubercle fuses to the tibia as part of bone maturation, usually over the following 1 to 2 years. Therefore, reassurance that the condition is self-limiting is appropriate.

Acute leukemia can occasionally present with bone pain in children. The presence of systemic symptoms, organomegaly, lymphadenopathy, bruising, or pallor would make it necessary to obtain a complete blood count (**choice A**) in a child presenting with bone or joint symptoms.

Erythrocyte sedimentation rate (**choice B**) is elevated in most rheumatic conditions. The absence of systemic symptoms and lack of physical findings affecting the knee joint in this patient make the diagnosis of a rheumatic disorder less likely.

In the presence of typical physical signs, magnetic resonance imaging (**choice C**) is not indicated, and referral to an orthopedic surgeon (**choice E**) is not necessary.

14. The correct answer is E. Oral thrush is common in the newborn period and arises from infection of the mucous membrane by *Candida*. Infants become colonized by acquiring the infection from the mother during delivery. The use of antibiotics can lead to development of thrush. Other individuals who are susceptible are those with immunodeficiency (congenital or acquired) and diabetes mellitus. The whitish plaques of thrush can be confused with milk curds in newborns, but the latter can be easily removed with a spatula and do not result in punctate bleeding spots. Treatment is usually with nystatin suspension that acts locally on the oral mucosa. Clotrimazole and miconazole are also effective therapies.

Amoxicillin (**choice A**) is an antibiotic commonly used to treat respiratory infections in children. It has no antifungal activity.

Amphotericin B (**choice B**) is an antifungal agent that is used parenterally to treat serious systemic fungal infections. It has no role in treating infections confined to the skin or oral mucosa.

The frequency of oral thrush is no different in formula-fed infants, and change to formula feeding (**choice C**) is not indicated.

Although sometimes very mild cases of thrush may not require treatment (**choice D**), a newborn who is fussy or has feeding problems should be appropriately treated.

15. The correct answer is B. Infectious croup, or laryngotracheobronchitis (LTB), is an acute, self-limited illness in children under the age of 3 years. The most frequent etiology of LTB is parainfluenza virus. The illness resolves in 3 to 7 days and usually does not require hospitalization. Diagnosis is based on the clinical picture, or sometimes on x-ray of the airway, which shows characteristic narrowing of the subglottic area described as pencil-point sign or steeple sign. No further studies are required to make the diagnosis.

Haemophilus influenzae type B (**choice A**) can cause epiglottitis, a severe and potentially fatal illness in children between 3 and 5 years of age. The patients present with high fever, rapidly progressive airway obstruction, and toxic look. This illness has become rare due to immunization.

Respiratory syncytial virus (**choice C**) causes bronchiolitis and pneumonia in infants. It is a much less frequent cause of LTB than parainfluenza virus.

Group A streptococcal infection (**choice D**) presents with a sore throat in older children, often with tonsillar exudates, palatal petechiae, and enlarged, tender, cervical lymph nodes. Stridor is unlikely in streptococcal infections.

Streptococcus pneumoniae (**choice E**) causes otitis media and pneumonia in children. It does not cause croup or inspiratory stridor.

16. The correct answer is B. The two most common causes of microcytic anemia when routine testing is done at the age of 1 year are iron deficiency and thalassemia trait. Between the two, iron deficiency is far more prevalent. In children who have mild anemia (hemoglobin 9.5 to 11 g/dL) in screening complete blood count or hematocrit at 1 year of age, it is appropriate to give a trial of oral iron at a dose of 3 mg/kg/day for 1 to 2 months. If there is no response in the hemoglobin at this time, then iron therapy should be not be continued without further investigations. These should include complete blood count, reticulocyte count, serum iron level, serum ferritin level, and iron-binding capacity. Supportive evidence of insufficient iron in the diet should also be sought; most often there is history of excessive milk intake or use of non–iron-containing infant formula. In patients with thalassemia trait (either alpha- or beta-thalassemia), the blood count looks similar to iron deficiency due to the presence of low hemoglobin and low mean corpuscular volume (microcytosis). In thalassemia-trait, the red cell

distribution width (RDW) remains normal, whereas it is elevated in iron deficiency. Such patients do not respond to oral iron supplements. The child in this vignette has microcytic anemia with a normal RDW and did not respond to ferrous sulfate drops. In addition, his diet seems to contain adequate amounts of iron. Thus he may have thalassemia trait, for which the most informative test is hemoglobin electrophoresis.

Bone-marrow aspiration (**choice A**) is appropriate when aplastic anemia or leukemia is suspected. However, it is not required in this case.

Serum B_{12} level (**choice C**) is useful in diagnosis of megaloblastic anemia, which manifests as increased MCV. It is not needed in patients who have microcytosis.

Serum iron and ferritin level (**choice D**) should be obtained in this child to rule out iron deficiency that has not responded to oral iron therapy. However, these tests will not likely reveal the etiology of the anemia.

Upper and lower gastrointestinal endoscopy (**choice E**) are important tests where iron deficiency is suspected to be due to blood loss from the gastrointestinal tract. It is not required for the child presented in this vignette.

17. **The correct answer is C.** This patient has the classic presentation of gonorrhea. Gonorrhea is a sexually transmitted disease caused by *Neisseria gonorrhoeae*. It affects the mucous membranes of the genitourinary tract and may at times also affect the mucosa of the oropharyngeum, rectum, and conjunctiva. Clinical presentation may vary. Some patients will have urethritis, cervicitis, or dysuria. Other patients with gonorrhea may be asymptomatic. Boys with urethritis will have purulent discharge and burning on urination. Postpubertal girls with symptomatic cervicitis may have purulent discharge, suprapubic pain, and dysuria. The cervix may be inflamed and friable. A culture for gonorrhea should be performed on any discharge from the cervix, urethra, rectum, or eye. If disseminated gonorrhea is suspected, cultures of the blood, pharynx, rectum, urethra, cervix, and synovial fluid (if applicable) should be evaluated. The discharge should also be Gram-stained. A Gram stain indicative of gonorrhea will show Gram-negative intracellular diplococci in polymorphonuclear cells. All patients with gonorrhea should be tested for syphilis and HIV. Ceftriaxone is the treatment of choice. In addition, doxycycline or azithromycin should be administered to treat presumed or proven Chlamydia.

Colorless pyriform flagellates in a saline smear (**choice A**) are diagnostic of trichomoniasis, a sexually transmitted infection with *Trichomonas vaginalis*. It is a common cause of vaginal pruritus, burning, and a frothy leukorrhea. In men, it may cause urethritis, prostatitis, or balanoposthitis (inflammation of the glans and prepuce), but is very frequently asymptomatic.

Chains of Gram-negative bacilli (**choice B**) are found in 50% of smears performed in patients with chancroid, a sexually transmitted infection with *Haemophilus ducreyi*. Clinically, one or more deep or superficial tender ulcers on the genitals are accompanied by painful inguinal adenitis in half of the cases.

Motile spiral spirochetes on darkfield microscopy (**choice D**) are diagnostic of syphilis, a sexually transmitted infection with *Treponema pallidum*. Clinically, the primary lesion is an asymptomatic small red papule or crusted erosion that indurates and assumes a cartilaginous-like consistency over several days. The surface of the erosion typically oozes a serous fluid. Regional lymph nodes are enlarged, nontender, and firm.

Multinucleated giant cells are seen in Tzanck smears (**choice E**) (methylene blue staining) of genital herpes simplex lesions. Clinically, painful grouped vesicles may be accompanied by tender adenitis. Genital herpes is most commonly caused by herpes simplex virus type 2, although contemporary sexual practices have caused an increase in the number of cases caused by herpes simplex virus type 1.

18. **The correct answer is B.** The obvious diagnosis is congenital diaphragmatic hernia, but the emergent issue is the pulmonary hypoplasia on the left and pulmonary hypertension that affects both lungs. Respiratory support is required for a few days before surgical correction is attempted.

As suggested, the infant is not ready to undergo surgical correction yet (**choice A**).

A chest tube (**choice C**) might be needed if positive pressure ventilation were to produce a pneumothorax, but right now that is not what the infant needs.

There is high pulmonary vascular resistance now, and that is part of the problem. To increase it further (**choice D**) would make things worse. Drugs with the opposite effect (such as nitric oxide) may be used.

The infant needs positive pressure ventilation, but if it is done through a mask (**choice E**), part of that gas will end up in the gastrointestinal tract, further compressing the lungs.

19. **The correct answer is A.** Charcot-Marie-Tooth disease (hereditary motor-sensor neuropathy type I) is the most common genetic neuropathy. In this autosomal dominant disease, patients have peroneal muscular atrophy. Most patients are asymptomatic until late childhood or early adolescence. Patients have a history of gait

disturbance, clumsiness, and tripping over their own feet. By the teenage years, patients exhibit pes cavus, tremor, and variable sensory loss (stocking-glove distribution). On physical examination, the peroneal and tibial nerves are most commonly affected. There is muscle wasting of the lower legs, giving them a stork-like appearance. Patients have claw hands with nerves that are palpably enlarged. There is decreased motor and sensory conduction. A sural nerve biopsy demonstrates "onion bulb" formations that surround the axons. This pathologic finding is called interstitial hypertrophic neuropathy. The definitive genetic diagnosis is made with genetic testing of the blood. Orthotics are used to brace the feet and the legs. Parents should be examined and nerve conduction studies performed.

Duchenne muscular dystrophy (**choice B**) is the most common hereditary neuromuscular disease. It is transmitted as an X-linked recessive disease, with approximately 30% new mutations. Clinically, poor head control may be one of the first signs of the disease. There could be mild delay in early gross motor skills. On physical examination, patients will have pseudohypertrophy of the calves. Gower sign is seen by 3 years of age. Hip girdle weakness and a Trendelenburg sign (at 5 to 6 years of age) develop. Patients eventually lose ambulation capabilities. Patients have a poor cough and pharyngeal weakness. Death usually occurs by 18 years of age from respiratory problems and/or, rarely, heart problems.

Friedrich ataxia (**choice C**) is a hereditary metabolic degenerative disease caused by an expanded GAA triplet repeat in the frataxin gene (9q13-q21.1). It is transmitted as an autosomal recessive or dominant trait. Ataxia usually appears before 10 years of age. Cardinal features include progressive limb and gait ataxia, dysarthria, loss of joint position and vibration senses, absent tendon reflexes in the legs, and extensor plantar responses. These patients have an explosive dysarthric speech, and nystagmus. Titubation is present when sitting or standing. The diagnosis is based on clinical signs and symptoms. There is no available treatment. This is a progressive disorder with significant morbidity. Loss of ambulation typically occurs 15 years after disease onset.

Guillain-Barré syndrome (**choice D**) is a postinfectious polyneuropathy that causes demyelination in motor and sometimes sensory nerves. This disease affects patients of all ages. *Campylobacter jejuni* and *Mycoplasma pneumoniae* have been associated with Guillain-Barré. Clinically, an ascending weakness and paralysis that begins in the lower extremities can be seen. This usually occurs 10 days after a nonspecific viral infection. Weakness may progress to include the respiratory muscles, causing respiratory insufficiency. In most cases, there is a spontaneous recovery in 2 to 3 weeks. Involvement of the respiratory muscles may result in respiratory failure.

Werdnig-Hoffman disease (**choice E**) is a degenerative disorder of motor neurons. The disease is transmitted as an autosomal recessive trait. Onset is before 2 years of age and often begins in utero. Patients have generalized weakness and severe hypotonia of the proximal and distal limbs, intercostals, and bulbar muscles. The legs tend to lie in a frog-leg position with hips abducted and knees flexed. Fasciculations are visible in the tongue, and the patient has flaccid quadriplegia. There is no medical treatment to stop progression of the disease. Therefore, only supportive care is given. Most patients die before the age of 2 from respiratory failure and food aspiration.

20. The correct answer is D. This patient has genital herpes, a sexually transmitted infection caused by herpes simplex virus (HSV). HSV is a double-stranded, DNA-containing, enveloped virus that causes a number of problems involving the eye, skin, oral mucosa, central nervous system, and genital tract. There are two types of HSV, type 1 and type 2. HSV-1 causes infections of the mouth, lips, eyes, central nervous system, and, uncommonly, genital disease. HSV-2 is responsible for genital infections and neonatal infections and may cause oral lesions. Genital herpes is more commonly seen in teenagers and adults. It is transmitted by sexual activity. Fever, regional adenopathy, and dysuria may be present in patients with primary genital herpes infection. In girls, the vulva and vagina may have vesicles and ulcers, but the cervix is the primary site of infection. In boys, vesicles and ulcers may be found on the penis.

Diagnosis may be confirmed by the Tzanck test on scrapings from the herpetic lesions. An early vesicle, not a pustule or crusted lesion, is unroofed, and the base of the lesion is scraped gently with a scalpel blade. The material is placed on a glass slide, air-dried, and stained with Giemsa or Wright stain. Multinucleated giant cells with intranuclear inclusions suggest the presence of herpes, but culture or immunofluorescence testing must be performed to identify the specific virus. ELISA and immunofluorescent techniques may be helpful. The diagnosis may also be made clinically.

Acyclovir is the treatment for herpes. Valacyclovir, a prodrug of acyclovir and famciclovir, and a prodrug of penciclovir are other oral antiherpes drugs used to treat genital herpes. Genital herpes is a risk factor for human immunodeficiency virus infection. Genital herpes may be present without symptoms. During this latent infection, herpes virus is being shed and sexual partners are

at risk. If the female patient with recurrent genital herpes is pregnant and has active lesions, then the infant passing through the birth canal is at risk for acquiring infection.

The clinical picture of this patient is typical for primary genital herpes (fever, regional adenopathy, and vesicles and ulcers on the genitals). It would be reasonable to perform a viral culture and typing to determine the strain of virus that caused the infection, but performing a bacterial culture (**choice A**) is not indicated because there are no signs of bacterial infection.

A complete blood count (**choice B**) would not aid in the diagnosis of primary genital herpes.

A KOH preparation (**choice C**) is performed on scaling skin lesions when a fungal etiology is suspected. The edge of such a lesion is scraped gently with a scalpel blade; the removed scale is collected on a glass microscope slide and treated with 1 to 2 drops of a solution of 10 to 20% KOH. KOH dissolves keratin and allows easier visualization of fungal elements. Brief heating of the slide accelerates dissolution of keratin. When the preparation is viewed under the microscope, the refractile hyphae will be seen more easily when the light intensity is reduced. This technique can be used to identify hyphae in dermatophyte infections, pseudohyphae and budding yeast in *Candida* infections, and fragmented hyphae and spores in tinea versicolor.

Even though a skin biopsy (**choice E**) is a minor surgical procedure, it is not necessary to perform an invasive procedure that might result in a permanent scar in a straightforward case that can be easily confirmed with a simple Tzanck preparation from the base of a lesion.

21. **The correct answer is E.** It is the responsibility of the treating physician to report any suspected child abuse to the appropriate agency immediately. It is also the law in most states. All states have an identified agency responsible for the investigation of all such cases. The agency is required to make a determination about the most suitable placement for the child during the investigation and thereafter. In many states, this agency is called the Department of Child and Family Services.

The physician may pursue any diagnostic investigations or treatment considered necessary for the child's care, even if parental consent is not available (**choice A**).

It is not appropriate for the physician to confront the baby-sitter (**choice B**) or any other person involved in the child's care. What is appropriate is for the physician to obtain a thorough history in a nonconfrontational and nonjudgmental manner. The physician's role is to gather all of the medical facts. It is the role of the state agency or police to question potential suspects and conduct the remainder of the investigation.

Many hospitals have an identified department or person who can facilitate referrals for suspected child abuse. Often this individual is a social worker (**choice C**). Typically, there is a social worker on call for a hospital 24 hours a day to handle such situations. It is not appropriate to wait until the following morning to contact a social worker.

In cases of suspected child abuse, the physician's duty to protect the child against further injury overrides any consideration of confidentiality (**choice D**).

22. **The correct answer is F.** Neonates exhibit several innate responses that appear to promote infant-maternal interaction. A neonate will orient its head toward a human voice and learn to respond preferentially toward its own mother's voice. A neonate will also fix its eyes on the human face, and preferentially fixates on a familiar face such as its mother's.

The grasp reflex (**choice A**) is elicited by tactile stimulation of the midpalm and leads to flexion of the digits. It allows the newborn infant to grasp its mother, just as an arboreal primate would. The grasp reflex disappears at age 3 months.

The rooting reflex (**choice B**) is elicited by lightly stimulating the infant's cheek and observing the reflexive attempts to bring the stimulating object (such as a nipple) to the mouth.

The asymmetric tonic neck reflex (**choice C**) is an early reflex pattern that appears at age 2 to 4 weeks and disappears at age 6 months. This reflex is not seen immediately after birth because the newborn has high flexor tone throughout the body.

The normal newborn cries immediately following a painful stimulus. A latency period (**choice D**) is not normal and may be present in an infant who is neurologically depressed or impaired.

A newborn infant is unable to roll over from back to stomach (**choice E**). This milestone is usually achieved at age 4 to 8 months.

23. **The correct answer is D.** The combination of delirium, mydriasis, dryness of skin and mucous membranes, tachycardia, and fever suggest cholinergic (muscarinic) blockade that most commonly would be caused by ingestion of atropine-like drugs. The drug of choice for therapy, and as a diagnostic aid, is physostigmine. Physostigmine is a potent peripheral and central anticholinesterase. Like many other anticholinesterases, it penetrates the blood-brain barrier to counteract the

central effects of the cholinergic blocking agents. It must be administered cautiously and the patient monitored in an intensive care unit because of the tendency of this drug to cause cardiac arrhythmias and hypotension.

Edrophonium (**choice A**) would have muscarinic and cholinergic effects peripherally, but not centrally. Furthermore, its duration of action is for a very short period of time.

Phenobarbital (**choice B**) would further impair the patient's sensorium. A preferable anticonvulsant would be diazepam.

Phenothiazine (**choice C**) has cholinergic blocking action and would only worsen the patient's condition.

Thiamine (**choice E**) would be ineffective, although not harmful.

24. The correct answer is B. The greatest risk of foreign-body aspiration is among toddlers. Small toys (or parts of toys), seeds, and nuts are among the objects most frequently implicated in such incidents. Unfortunately, a history of choking is not always elicited and the diagnosis should always be suspected when evaluating unexplained respiratory symptoms in this age group. Localized wheezing and unequal breath sounds are important diagnostic clues during examination. The diagnosis is confirmed in most cases by chest x-ray, which shows either collapse or hyperinflation (due to air-trapping) of the affected lung. Bronchoscopy is diagnostic in doubtful cases and the foreign body can be removed at the same time.

Some patients with chronic cough may have aberrant blood vessels or congenital anomalies that compress the bronchus and may be visible on barium swallow (**choice A**). Gastroesophageal reflux can also be detected by barium swallow, as can an esophageal pH probe study (**choice C**). Such patients often become symptomatic during infancy.

Asthma is the most common etiology of recurrent cough in children. Children often have longer duration of symptoms and wheezing at the time of presentation. In this case, the etiology of symptoms could be asthma, however the absence of previous symptoms and the localized signs on lung examination make it important to rule out foreign body in this age group. If asthma is suspected, a trial of inhaled bronchodilators should be given as a therapeutic trial rather than performing skin allergen testing (**choice D**).

Cystic fibrosis is always possible in children with chronic respiratory symptoms (**choice E**). In this child, the absence of gastrointestinal symptoms, short duration of cough, and normal growth would make cystic fibrosis an unlikely diagnosis and a sweat chloride test unnecessary.

25. The correct answer is B. This patient has the typical clinical features of spinal muscular atrophy type I (infantile form), or Werdnig–Hoffman disease, a degenerative disorder of motor neurons. The primary pathology is atrophy of anterior horn cells in the spinal cord and of motor nuclei in the brain stem, with secondary atrophy of motor nerve roots and of muscle. The disease is transmitted as an autosomal recessive trait. Onset is before 2 years of age and often begins in utero. Patients have generalized weakness and severe hypotonia of the proximal and distal limbs, intercostals, and bulbar muscles. The legs tend to lie in a frog-leg position, with hips abducted and knees flexed. Fasciculations are visible on the tongue, and the patient has flaccid quadriplegia. Tendon stretch reflexes are absent. Infants have normal intelligence. The electromyelogram shows fibrillation and evidence of muscle denervation. The serum enzyme determinations (creatine kinase) are normal or slightly elevated. Muscle biopsy shows evidence of denervation, and nerve biopsy shows slowed conduction. There is no medical treatment to stop progression of the disease. Therefore, only supportive care is given. Most patients die before the age of 2 from respiratory failure and food aspiration.

Patients with Werdnig-Hoffman disease have normal intelligence, facial movements, sensation, and sphincter function. The disease is not associated with severe mental retardation (**choice A**).

Werdnig-Hoffman disease is inherited as an autosomal recessive gene defect located on chromosome 5. The defective gene is a neuronal apoptotic inhibitor protein. The gene mutation is, hence, not located on the X-chromosome (**choice C**).

Serum creatine kinase levels are either normal or only slightly elevated in this disease. They are not the diagnostic test of choice (**choice D**). The diagnosis is made by demonstrating atrophy of the anterior horn cells on autopsy or by identification of mutations in the neuronal apoptotic inhibitor protein gene in the affected patient.

It is incorrect to say that this child will never attain normal sphincter function (**choice E**); this disease does not affect the sphincter muscles, and function there is normal.

26. The correct answer is E. Trisomy 21, or Down syndrome, occurs in 1 in 600 live births and is the most common trisomy syndrome. About 95% of the cases are due to trisomy of chromosome 21, and the remaining 5% are due to translocations involving chromosome 21 and another chromosome. Cardiac defects are seen in nearly 40% of patients, with atrioventricular canal defect being a typical cardiac malformation. Gastrointestinal malformations (particularly duodenal

atresia) are also seen in 5% of cases. Other problems include variable degrees of mental retardation, characteristic facial appearance (described in the case vignette), and hypotonia. The risk of leukemia is increased in patients with Down syndrome.

DiGeorge syndrome (**choice A**) is due to abnormalities of chromosome 22, and its features include aortic stenosis, parathyroid and thymic aplasia, ear defects, and cleft palate.

In Prader-Willi syndrome (**choice B**) there is severe hypotonia and failure to thrive during infancy. Older children are obese and have developmental delay.

Both trisomy 13 and 18 (**choices C and D**) are rare and have more severe malformations than trisomy 21, with most patients dying during infancy. In trisomy 13, cleft lip and palate, heart defects, omphalocele, and polydactyly are present. In trisomy 18, the distinguishing features are microcephaly, cardiac defects, overlapping fingers, and prominent calcaneus.

27. The correct answer is D. Bronchiolitis is one of the most common respiratory illnesses in infants during the winter season. It is most commonly due to respiratory syncytial virus (RSV), which causes inflammation of the small airways, causing obstruction and trapping of air. Clinically, this manifests as coughing, wheezing, and hyperinflated lung fields on chest x-ray. Often there is a mild fever and a prominent rhinitis, which can increase the breathing difficulty by causing nasal blockage. The diagnosis, based upon demonstration of RSV antigen in the nasopharyngeal aspirate, is both rapid and specific.

Bronchoscopy (**choice A**) is very important when foreign-body aspiration is suspected. Toddlers form the highest risk group for aspiration of foreign body, which is uncommon in very young infants.

Chest x-ray (**choice B**) will show hyperinflation of lung fields or sometimes interstitial infiltrates in infants with bronchiolitis. The changes are not specific for RSV infection.

Complete blood count (**choice C**) is helpful for diagnosis of bacterial pneumonia (leukocytosis with increased neutrophils) and pertussis (lymphocytosis). It is generally not helpful in diagnosing RSV infection.

Throat swab (**choice E**) is used when acute pharyngitis is suspected. This is usually a problem in older children who present with sore throat and tonsillar enlargement with exudates. Rapid diagnosis of streptococcal infection is possible on throat swabs obtained at the office.

28. The correct answer is B. Retropharyngeal abscesses can develop in preschool children due to the presence of lymph nodes behind the posterior pharyngeal wall. It is usually a complication of acute pharyngitis, although some cases arise as a result of penetrating trauma or osteomyelitis affecting the cervical vertebrae. Group A *Streptococcus*, *Staphylococcus aureus*, and anaerobes are the most common causative agents. There is sudden onset of high fever and the enlarging abscess causes pain and obstruction, which prevent the child from swallowing saliva. The neck is held in hyperextension and the breathing can become difficult. A lateral neck x-ray will show increase in retropharyngeal soft tissue and help to confirm the diagnosis. CT of the neck can help in cases where the diagnosis is in doubt.

A complete blood count (**choice A**) is likely to show leukocytosis with increased neutrophils in such patients, but is a nonspecific finding.

The pain can cause patients with retropharyngeal abscess to hold their necks in rigid extension, thus raising the possibility of meningitis (**choice C**). However, the presence of upper airway obstruction and normal sensorium should make this diagnosis less likely and so a lumbar puncture would not be helpful in establishing a diagnosis.

Nasopharyngeal aspirate (**choice D**) can be cultured to diagnose viral infections. Retropharyngeal abscess is a bacterial illness, and thus this test is not helpful in this patient.

Throat swab (**choice E**) is useful for diagnosis of bacterial pharyngitis, but does not help in establishing a diagnosis of retropharyngeal abscess.

29. The correct answer is C. Erythema toxicum is a common condition in the neonatal period, manifesting as a transient rash of unknown etiology. The diagnosis is made on characteristic visual appearance of vesicles and erythema. However, if the vesicular fluid were observed under a microscope it would reveal eosinophils. The rash disappears within a few days without specific treatment.

Allergy to cow's milk protein (**choice A**) is uncommon and does not present this early. More often, there are prominent gastrointestinal symptoms and blood loss in such infants.

Atopic dermatitis (**choice B**) is a common skin disorder among infants that begins between 1 and 6 months of age, with red, itchy papules most prominent over the cheeks.

Intrauterine infections (**choice D**) can cause several forms of rash in newborns, but the most common presentation is with petechial rash. The infants tend to be small for gestational age and have organomegaly, microcephaly, chorioretinitis, or other manifestations to support the diagnosis.

Sepsis (**choice E**) can produce a rash due to coagulopathy and thrombocytopenia. However, such infants look sick and are unable to continue with normal feeding. Temperature instability, especially hypothermia, can be observed in newborns with sepsis.

30. The correct answer is C. While less frequent in full-term infants, neonatal sepsis can be a serious, life-threatening illness in newborns. Newborn infants who develop a temperature (generally over 38 C) must be presumed to have sepsis until proven otherwise. Localizing signs are generally not found in this age group, but should be carefully looked for. Such patients should be admitted on an urgent basis, and complete blood count, chest x-ray, and blood, urine, and cerebrospinal fluid cultures should be obtained. Broad-spectrum antibiotics should be started until the infant is afebrile and the culture results are negative.

It is not appropriate to monitor newborns with fever at home, due to the risk of bacterial sepsis. Thus, acetaminophen (**choice A**) or arranging another visit in 2 days (**choice B**) are both unsuitable options in the management of this patient.

While a CBC should be sent for this child, a normal blood count does not rule out bacterial sepsis (**choice D**).

Neonatal sepsis requires treatment with parenteral antibiotics due to risk of septicemia or meningitis. Thus, oral amoxicillin (**choice E**) should not be started in this patient.

31. The correct answer is D. This patient exhibits features of Turner syndrome, which has an abnormal karyotype (45,XO). The absence of one X chromosome leads to streak gonads and primary ovarian failure. Thus, there is absence of normal breast development during adolescence. Due to the presence of adrenal androgens, axillary and pubic hair development may occur. Short stature is an important sign of Turner syndrome and becomes more apparent when the peers undergo the growth spurt associated with normal puberty. In addition to the abnormal physical features described in the case vignette, Turner syndrome is associated with renal anomalies, cardiovascular anomalies, hypertension, hearing abnormalities, and, in the newborn period, edema of the hands and feet.

Abdominal ultrasound (**choice A**) will reveal absence of normal ovarian structure and the presence of any renal anomalies. However, these features are not sufficient to confirm a diagnosis of Turner syndrome.

Gonadotropin level (**choice B**) in the serum is elevated in the presence of primary ovarian failure. When the failure of puberty is due to hypothalamic causes, serum gonadotropin levels are suppressed.

Patients with congenital growth hormone deficiency manifest severe growth failure with normal body proportions (**choice C**). The growth pattern and physical features in the present case more strongly favor a diagnosis of Turner syndrome.

Acquired hypothyroidism (**choice E**) leads to marked deceleration of growth after an initial period of normal growth. Girls with Turner syndrome have increased incidence of thyroid abnormalities.

32. The correct answer is D. This infant has ambiguous genitalia. The most common cause of ambiguous genitalia is female pseudohermaphroditism, and congenital adrenal hyperplasia (CAH) is the most common cause of female pseudohermaphroditism. 21-Hydroxylase enzyme deficiency is the most common enzyme deficiency and accounts for 90% of congenital adrenal hyperplasia. It is critical that the precise etiology be delineated so that any urgent metabolic abnormalities can be treated safely and quickly. In female pseudohermaphroditism, the gonadal tissue is represented by ovaries. The chromosomal analysis shows 46,XX. Overexposure to androgens in utero causes severe masculinization of the external genitalia. CAH patients display marked enlargement of the phallus, which excretes urine through a single urogenital sinus opening.. Many of these infants experience salt wasting caused by reduced aldosterone production. Salt wasting may cause a low plasma sodium with high renin and potassium concentrations. Death may occur in the neonatal period if the ensuing electrolyte abnormalities are unrecognized and untreated. Dehydration secondary to vomiting can lead to circulatory collapse.

An arterial blood gas (**choice A**) is not helpful in the treatment of the underlying condition. Although the gas will reveal an acidosis, the source of the dehydration and lethargy is electrolyte abnormalities.

Karyotype (**choice B**) is important to determine the exact chromosomal makeup of the infant. These results help in determining the sex of the infant for rearing, however, and are not available immediately. It is an important test, but not one that will help this lethargic, dehydrated infant in the intensive care unit.

Pelvic ultrasound (**choice C**) can be used to determine the presence or absence of various intraabdominal organs or the presence of nonpalpable testes. It does not provide definitive answers, however, and will not help this dehydrated infant.

In patients with CAH caused by 21-hydroxylase or 11-beta-hydroxylase enzyme deficiency, the 17-alpha-hydroxyprogesterone serum level (**choice E**) is elevated. This piece of knowledge is important for determining the underlying etiology, but in the intensive care unit, the patient's electrolytes are of higher priority.

33. The correct answer is D. By making a family tree you can see that the described pattern of inheritance is consistent with X-linked dominant transmission. In this case the disease is manifested in female heterozygotes as well as carrier males (hemizygotes).

Albinism (**choice A**) and congenital adrenal hyperplasia (**choice B**) are inherited in an autosomal recessive fashion. In autosomal recessive inheritance, there is a statistical probability that one of four children will be affected with the disease, two of four will be carriers, and one of four will be completely normal. There is no gender preference for the disease.

Bruton agammaglobulinemia (**choice C**) and Duchenne muscular dystrophy (**choice E**) are inherited in an X-linked recessive fashion. The usual transmission is from heterozygous females to male offspring. Family history reveals that the disorder is only found in certain male relatives, and commonly in maternal uncles.

34. The correct answer is A. Myelomeningocele is a neural tube defect and the most severe form of dysraphism involving the vertebral column. The etiology is unknown; however, it is thought that agents such as drugs, radiation, malnutrition, and genetics have a role in adversely affecting normal central nervous system development. After 1991, 50% of cases of neural tube defects in the United States are related to nutritional deficiency of folic acid and are thus preventable. Offspring in a family with one affected child are at increased risk of neural tube defect. The biochemical marker for neural tube defects is alpha-fetoprotein acetylcholinesterase (AFP). This substance is excreted from the fetus and leaks into the amniotic fluid when there is failure of the neural tube to close. Attention to nutrition in the prenatal period, with prenatal vitamins and folic acid, decreases the risk. The U.S. Public Health Service recommends intake of folic acid at a dosage of 0.4 mg/day for all women anticipating pregnancy. Since 1996, mandatory folic acid fortification of enriched cereal grain has been implemented in the United States.

Vitamin A (**choice B**) deficiency results in a skin eruption that consists of keratotic papules of varying size over the shoulders and extremities that may spread to involve most of the body surface except palms and soles (where there are no pilosebaceous follicles). On the face it resembles acne because of large comedones that develop. The whole skin displays dryness and fine scaling. Eye findings are prominent and often pathognomonic. They include night blindness (nyctalopia), an inability to see bright light, xerophthalmia, xerosis corneae, and keratomalacia. Vitamin A deficiency is a major cause of blindness in children in Third World countries.

Vitamin B_2 (**choice C**) deficiency manifests with the oral-ocular-genital syndrome. Angular cheilitis and cheilosis are prominent. The tongue is atrophic and magenta-colored. A seborrheic dermatitis–like eruption affects the eyebrows, glabella, and nasolabial folds of the face. Confluent dermatitis of the scrotum or vulva is present and extends onto the thighs. Photophobia and blepharitis angularis also occur. All the signs respond dramatically to 5 mg of riboflavin (vitamin B_2) daily.

Vitamin B_6 (**choice D**) deficiency occurs in patients with uremia or cirrhosis, and with some medications. Skin changes are prominent and include a seborrheic dermatitis–like eruption, atrophic glossitis with ulceration, angular cheilitis, conjunctivitis, and intertrigo. The patients may be somnolent and confused, and may display signs of neuropathy.

Vitamin K (**choice E**) deficiency occurs in patients with malabsorption syndromes or liver disease, or as a side effect of certain medications (coumarin, salicylates, cholestyramine). The result is a decrease in the vitamin K–dependent clotting factors II, VII, IX, and X. Purpura, hemorrhage, and ecchymoses occur in the skin. Treatment is with 5 to 10 mg/day of intramuscular vitamin K. In acute, life-threatening situations, fresh frozen plasma will provide the deficient clotting factors until their synthesis can be resumed.

35. The correct answer is A. Friedrich ataxia is a hereditary metabolic degenerative disease caused by a repeat expansion of a gene that codes for the mitochondrial protein frataxin, resulting in an overload of iron in mitochondria. It is transmitted as an autosomal recessive or dominant trait. Ataxia usually appears before 10 years of age. Cardinal features include progressive limb and gait ataxia, dysarthria, loss of joint position and vibration senses, absent tendon reflexes in the legs, and extensor plantar responses. Patients have dysarthric speech and nystagmus. On physical examination, patients have ataxia and absent deep tendon reflexes. The lower

extremities are usually more involved than the upper extremities, and there is also loss of vibration and position sense. Skeletal deformities, such as high arched foot (pes cavus), hammertoes, or kyphoscoliosis, may be present. Titubation is present when sitting or standing. The diagnosis is based on clinical signs and symptoms. There is no available treatment. This is a progressive disorder with significant morbidity. Loss of ambulation typically occurs 15 years after disease onset. More than 95% of patients are wheelchair-bound by age 45 years. Patients with Friedrich ataxia may develop hypertrophic cardiomyopathy, with progression to intractable congestive heart failure causing death.

Refsum disease (**choice B**) is a neurocutaneous syndrome that is characterized by the accumulation of phytanic acid in plasma and tissues. Patients are unable to degrade phytanic acid because of a deficient activity of phytanoyl-CoA hydroxylase. Peripheral polyneuropathy, cerebellar ataxia, retinitis pigmentosa, and ichthyosis are the major clinical components. The symptoms evolve slowly and insidiously from childhood through adolescence and early adulthood.

Spastic hemiplegia (**choice C**), a form of cerebral palsy, is a disorder of impaired motor functioning and posture with onset before or at birth or during the first year of life. It is a nonprogessive disorder and varies widely in its causes, manifestations, and prognosis. The most obvious manifestation is impaired ability of voluntary muscles. Spastic hemiplegia involves only one side of the body. Cognitive function may be spared, but seizures are common.

Werdnig-Hoffman disease (**choice D**) is a degenerative disorder of motor neurons. The primary pathology is atrophy of anterior horn cells in the spinal cord and of motor nuclei in the brain stem, with secondary atrophy of motor nerve roots and of muscle. The disease is transmitted as an autosomal recessive trait. Onset is before 2 years of age and often begins in utero. Patients have generalized weakness and severe hypotonia of the proximal and distal limbs, intercostals, and bulbar muscles.

Wilson disease (**choice E**) is an autosomal recessive degeneration of basal ganglia characterized by increased copper deposition in the brain, liver, kidney, and cornea, and low serum copper and ceruloplasmin levels. A family history is often present. Clinically, patients may have tremor, drooling, "fixed smile," dysarthria, and choreoathetosis. Children may have hepatomegaly. Almost all patients with neurologic disease have Kayser-Fleischer rings, i.e., a yellow-brown deposit at the limbus of the cornea. The diagnosis of Wilson disease is confirmed by low serum copper and ceruloplasmin levels, increased urine copper concentrations after the administration of penicillamine, and a rise in the level of copper in the liver.

36. The correct answer is E. Lesch-Nyhan syndrome is an X-linked disorder of purine metabolism deficiency of hypoxanthine-guanine phosphoribosyl transferase. It is, therefore, seen almost exclusively in boys. This metabolic deficiency leads to an excess of uric acid. Infants with Lesch-Nyhan syndrome have no apparent neurologic dysfunction until a delay in motor development occurs in the first few months. This is usually the first abnormality noted. In addition, patients have self-destructive behavior patterns. On physical examination, there is evidence of self-mutilation, choreoathetosis, spasticity, psychomotor retardation, gouty arthritis, and renal calculi. Initially, uric acid crystalluria and microhematuria may present as "orange sand" noticed in the diapers. Plain abdominal radiographs (**choice D**) will not detect calculi composed of uric acid, oxypurine metabolites, or allopurinol because they are radiolucent. However, they are easily seen by ultrasonography. The diagnosis may be made by the presence of dystonia and self-mutilation. The definitive diagnosis is made by analysis of the hypoxanthine-guanine phosphoribosyl transferase enzyme. Treatment consists of allopurinol for renal complications. Behavior modification, restraints, and removal of teeth are needed to reduce anxiety and stabilize mood.

Cerebrospinal fluid analysis (**choice A**) does not reveal any abnormality in patients with Lesch-Nyhan syndrome.

Computerized tomography of the brain (**choice B**) and magnetic resonance imaging of the brain (**choice C**) generally do not reveal any structural abnormalities or signal changes in the gray or white matter. Even if there is mild loss of brain volume, it is most often too small to be detected in routine imaging studies. Neuroimaging of the spinal cord may reveal early degenerative joint disease with possible damage to the spinal cord or nerve roots.

37. The correct answer is C. This patient has fulminant hepatic failure associated with hepatolenticular degeneration. Liver biopsy is the criterion standard for diagnosis. Hepatolenticular degeneration (Wilson disease) is an autosomal recessive degeneration of basal ganglia characterized by increased copper deposition in the brain, liver, kidney, and cornea, and low serum copper and ceruloplasmin levels. A family history is often present, and screening asymptomatic family members identifies 25% of cases. Initial presentation of Wilson disease occurs between adolescence and 40 years of age. The clinical presentation varies, although it has been noted that hepatic failure is more common in children whereas psychiatric symptoms are more common in adults. Clinically, patients may have tremor, drooling, "fixed smile," dysarthria, and choreoathetosis. Children

may have hepatomegaly. Almost all patients with neurologic disease have Kayser-Fleischer rings, i.e., a yellow-brown deposit at the limbus of the cornea. The diagnosis of Wilson disease is confirmed by low serum copper and ceruloplasmin levels, increased urine copper concentrations after the administration of penicillamine, and a rise in the level of copper in the liver.

Analysis of 24-hour urine collection (**choice A**) will reveal increased urinary copper excretion in excess of 100 mg/dL. Further increase in copper excretion after penicillamine administration is used to confirm the diagnosis. Nevertheless, a liver biopsy remains the gold standard for diagnosis.

Computerized tomography of the brain (**choice B**) is used to assess the presence of hypodense regions in the basal ganglia of patients with Wilson disease. Other possible findings include ventricular dilatation, brainstem atrophy, and posterior fossa atrophy.

Skeletal radiologic survey (**choice D**) is not recommended as part of the workup for Wilson disease in children because the musculoskeletal abnormalities do not develop until adulthood.

Slit-lamp examination of the eye (**choice E**) is necessary for confirming the presence of Kayser-Fleischer rings, copper granules in the stromal layer of the eye. They are always present with neurologic disease, and may be present in hepatic disease as well. Kayser-Fleischer rings fade and disappear with appropriate therapy.

38. **The correct answer is D.** Although uncommon, primary osteosarcoma, a malignant bone tumor, occurs most frequently in adolescents and teens. The distal femur is the most common location for osteosarcoma, but it may present almost anywhere in the appendicular skeleton. In the past, treatment for osteosarcoma was limited to amputation with a very low survival rate; however, with chemotherapy and limb salvage surgery, the mortality of osteosarcoma has been decreased significantly.

A benign bone cyst (aneurysmal or unicameral) (**choice A**) is generally not associated with thigh swelling or pain. Radiographically, cysts are radiolucent and do not appear to form bone.

There is no mention of a distal femur fracture in the patient's history, and although callous formation may be difficult to interpret on radiographic examination, the patient's history should give one that diagnosis; thus, **choice B** is incorrect.

The patient does not have a history of fever, chills, or night sweats and has an enlarging mass in her distal femur; this is not suggestive of osteomyelitis (**choice C**).

Paget disease is a pathologic condition that results in abnormal bone turnover and is not associated with enlarging, bone-forming masses (**choice E**).

39. **The correct answer is A.** This patient has the classic clinical presentation of Rett syndrome. Rett syndrome (pervasive developmental disorder) is a neurodegenerative disorder that affects only females. The onset usually occurs at 1 year of age with loss of developmental milestones and acquired microcephaly. On physical examination, there is a loss of purposeful movement, social withdrawal, stereotypical hand movements (hand wringing), and acquired microcephaly. The patient may have sighing, intermittent apnea, and autistic behavior. Currently, diagnosis of Rett syndrome is made if the patient meets defined clinical criteria. The diagnosis is supported by a positive mutational analysis of MECP2 (methyl-CpG binding protein-2). The gene is located on chromosome X. However, as many as 20% of females meeting the full clinical criteria for Rett syndrome may have no identified mutation. It is considered to be fatal in males. Because no cure exists, treatment is palliative and supportive. A multidisciplinary approach to care for persons with Rett syndrome is recommended. If the patient has seizures, anticonvulsants should be administered. These patients may have generalized tonic-clonic seizures. Death occurs during adolescence or the third decade, and may be caused suddenly from a cardiac arrhythmia.

NF1 (**choice B**) mutations or deletions are responsible for the development of type 1 neurofibromatosis. The gene is located on chromosome 17. These patients are at increased risk of developing nervous system neoplasms, including plexiform neurofibromas, optic gliomas, ependymomas, meningiomas, astrocytomas, and pheochromocytomas. Neurofibromas may undergo secondary malignant degeneration and become sarcomas.

PTEN (**choice C**) is a tumor suppressor gene located on chromosome 10q23. Mutation of this gene results in the multiple hamartoma syndrome (Cowden disease), characterized by the development of trichilemmomas, oral mucous papillomas, acral keratoses, breast cancer in women, and medullary carcinoma of the thyroid in both men and women.

SPINK5 (**choice D**) is a serine protease inhibitor of Kazal type responsible for the genetic defect that results in Netherton syndrome, a rare autosomal recessive genodermatosis of cause characterized by erythroderma, trichorrhexis invaginata (bamboo hair), ichthyosis linearis circumflexa, atopic diathesis, and failure to thrive.

TSC1 (**choice E**) is the gene responsible for tuberous sclerosis. It is located on chromosomes 9 and 16.

Tuberous sclerosis is a neurocutaneous syndrome characterized by mental retardation, facial fibroangiomas, hypopigmented spots of the skin, and epilepsy.

40. The correct answer is B. Most single congenital malformations follow the pattern of multigenic, or non-Mendelian, inheritance. Both genetic and environmental factors are thought to be involved in this form of inheritance. Most common congenital malformations fall into this category. The recurrence risk of the same congenital malformation occurring in a second offspring is generally in the range of 4 to 6%. It is worth noting that the concordance rate among monozygotic twins is well below 100%, indicating the role of environmental factors.

By its very nature, multigenic inheritance does not involve specific genes or cytogenetic markers. Thus, chromosomal studies (**choice A**) are generally not helpful in most isolated common congenital malformations such as clubfoot, pyloric stenosis, or cleft lip.

The risk of recurrence is higher for conditions that clearly follow Mendelian inheritance. These are single-gene defects such as sickle-cell anemia or cystic fibrosis. In these recessively inherited diseases, the recurrence risk in the second child is 25% (**choice C**).

As compared with the normal population, the risk of recurrence is increased about 20 to 50 times in congenital malformations that follow the pattern of multigenic inheritance (**choice D**).

Although the incidence of certain chromosomal defects (such as trisomy) increases with higher maternal age, the same is not true of diseases such as cleft palate, pyloric stenosis, or clubfoot that follow multigenic inheritance (**choice E**).

41. The correct answer is E. This child demonstrates signs of impending respiratory failure and should be immediately intubated in the emergency room. Patients in status asthmaticus should be evaluated for altered mental status, retention of carbon dioxide, and development of acidosis. These patients are at risk of dying due to respiratory failure unless the condition is recognized in time. Children with severe asthma may have a silent chest with no wheezing due to poor air exchange. They are unable to talk in full sentences due to hypoxia, and they need to breathe in while talking.

Children presenting with status asthmaticus may be dehydrated due to tachypnea and decreased intake of fluids. Thus, although intravenous fluids (**choice A**) are required in the management of this child, they should be provided after the breathing has been stabilized.

Methylprednisolone (**choice B**) or other steroids are essential in treating status asthmaticus. However, the onset of action is not immediate and thus other steps must be taken to improve ventilation.

Sodium bicarbonate (**choice C**) is sometimes used for severe acidosis, especially where ongoing bicarbonate loss is suspected. It should not be used in patients with inadequate ventilation.

It is not adequate to admit the child for observation alone (**choice D**), given the extreme respiratory distress.

42. The correct answer is C. This patient has the typical presentation of Duchenne muscular dystrophy. The diagnosis is confirmed by muscle biopsy. Duchenne muscular dystrophy is the most common hereditary neuromuscular disease. It is caused by a mutation in the dystrophin gene at Xp2.1. It is transmitted as an X-linked recessive disease, with approximately 30% new mutations. Clinically, poor head control may be one of the first signs of the disease. There could be mild delay in early gross motor skills. On physical examination, patients will have pseudohypertrophy of the calves. Gowers sign (pushing the knees to stand up) is seen by 3 years of age. Hip girdle weakness and a Trendelenburg sign (at 5 to 6 years of age) develop. Patients eventually lose ambulation capabilities. Patients have a poor cough, and pharyngeal weakness. Cardiomyopathy also develops. Diagnosis is made by muscle biopsy demonstrating necrosis, fat cells, and fibrous tissue. Creatine kinase is greatly elevated.

Becker muscular dystrophy is also caused by a deficiency of dystrophin, but the age of onset is later and the course of the disease is slower. Pseudohypertrophy is prominent, and pes cavus deformities are present. Cardiac and nervous system involvement is unusual.

An electrocardiogram (**choice A**) is helpful in uncovering sinus arrhythmia with deep Q waves and elevated right precordial R waves, but has no value in confirming the diagnosis of Duchenne muscular dystrophy.

Electromyography (**choice B**) is not diagnostic but it narrows the differential diagnosis, effectively excluding primarily neurogenic processes.

Radiographs of the spine (**choice D**) are important for screening and evaluating the degree of scoliosis in patients who are wheelchair-bound, but has no value in confirming the diagnosis of Duchenne muscular dystrophy.

Serum creatinine phosphokinase (**choice E**) is always increased in patients with Duchenne muscular dystrophy or Becker muscular dystrophy, up to 50 to 100 times the reference range. If a child has very elevated creatinine phosphokinase and proximal muscle weakness, further testing to confirm the diagnosis is indicated.

43. The correct answer is D. The clinical presentation of cystic fibrosis is variable, with a significant number of infants manifesting pulmonary symptoms. These can be chronic cough, recurrent pneumonia or bronchiolitis, wheezing, atelectasis, or hemoptysis. Infants who present with a picture of acute bronchiolitis repeatedly or outside the winter season should be suspected to have an ongoing problem such as asthma, cystic fibrosis, foreign body, or congenital malformation. In this case, hyperexpanded lungs, failure to thrive, and presence of loose, foul-smelling, greasy stools (indicative of exocrine pancreatic involvement) should lead to referral for performing a sweat chloride test. Elevated sweat chloride (>60 mEq/L) in the presence of suggestive symptoms is diagnostic of cystic fibrosis.

Children with persistent or recurrent pulmonary symptoms should be evaluated for possible foreign-body aspiration by bronchoscopy (**choice A**). The presence of failure to thrive and intestinal symptoms make this a less likely diagnosis.

Chest x-ray (**choice B**) is indicated in this case and would reveal hyperinflation and patchy atelectasis. These findings are not specific for cystic fibrosis.

Certain patients with protracted pulmonary symptoms and diffuse lung changes on x-ray require a lung biopsy (**choice C**) to establish a diagnosis. Sweat chloride testing is a more definitive and less invasive method of diagnosing cystic fibrosis, compared with lung biopsy.

Children with malabsorption may require GI endoscopy (**choice E**) to obtain a mucosal biopsy for diagnosis. However, prominent respiratory symptoms in this case indicate the presence of a systemic disorder, and sweat chloride testing is the appropriate diagnostic test.

44. The correct answer is C. This girl has pediculosis capitis (head lice). Microscopic examination of the snipped hair would help identify nits attached to hair shafts. Itching is the principal manifestation of this infestation and the lice should not be overlooked because of impetigo of the scalp or suboccipital lymphadenopathy, both secondary to scratching. Dermatitis of the neck, shoulders, and urticaria may also occur as a hypersensitivity reaction to the pediculus. It presents as red, scaly, lichenified plaques on the posterior neck and shoulders. Head lice can survive up to 2 days, or longer, off the scalp, so shared hats, headsets, combs, brushes, bedding, clothing or upholstery easily transmits pediculosis. Nits are mainly found on the hairs in the postauricular and occipital scalp as small, whitish, 0.5-mm nodules. They are easily spotted in dark-haired individuals but may be camouflaged well in blondes. Nits should not be mistaken for epidermal scale or hair casts, both of which are easily slid down the hair shaft, whereas nits are firmly attached and cannot be easily moved. In addition, nits fluoresce under Wood's light and are easily viewed on snipped hairs under the microscope.

Sabouraud's dextrose agar (**choice A**) is the most commonly used medium in medical mycology and serves as the basis for most of the morphologic descriptions of these fungi. Cultures should be maintained at room temperature (26 C) for up to 4 weeks before they are discarded as showing no growth.

The hair penetration test (**choice B**) is used to identify certain fungal cultures in vitro. Species of Trichophyton will penetrate a hair shaft placed in the medium, whereas those of Microsporum, for example, will not.

Potassium hydroxide hair preparation (**choice D**) is used to diagnose tinea capitis. The hair shaft is immersed in a 20% potassium hydroxide solution for several hours, and then examined under the microscope. This helps dissolve the hair keratins and permits visualization of the fungi.

The pull test (**choice E**) is utilized in the diagnosis and assessment of alopecia areata, a disorder in which round or oval patches of hair precipitously fall out leaving a smooth but otherwise normal skin surface with visible hair follicle orifices but no hair shafts. The pull test is performed at the periphery of such patches by firmly grasping approximately 50 hairs close to the scalp and firmly pulling. If more than 5 hairs are liberated from the scalp by this maneuver, the inflammatory process leading to hair loss is still active in that area of the scalp.

45. The correct answer is E. Mothers are advised to place their infants "back to sleep." Studies looking at SIDS rates during the introduction of such "Back to Sleep" campaigns have found a decline in occurrences of SIDS by nearly 40%, though a causal relationship has not yet been proven.

To the chagrin of their parents, many infants refuse to sleep in one position. In general, any comfortable position (**choice A**) other than prone significantly reduces the infant's chances of SIDS. The supine sleeping position followed by sleeping on either side (**choices B and D**) is safer than the prone position. The prone sleeping position (**choice C**) has been correlated with an increased risk for SIDS in several retrospective studies. Although the exact cause of increased risk with sleeping in the prone position has not been proven, this position is hypothesized to increase the risk for smothering and potentiating apneic episodes.

46. The correct answer is C. This patient meets the criteria for Kawasaki syndrome, the most common vasculitis among children age 5 years and younger. Patients must have an otherwise unexplained fever, together with four of the five following symptoms: conjunctivitis, mucous membrane changes, cervical lymphadenopathy, rash, and swelling or desquamation of the hands or feet. The disease is most common in children of Japanese descent and often can be mistaken for scarlet fever or a nonspecific viral syndrome. Prompt diagnosis and treatment with aspirin and intravenous immunoglobulin is essential. Up to a quarter of untreated patients develop dilated coronary arteries that are prone to stenosis and thrombosis.

Acetaminophen (**choice A**) can mask the fevers and treat some of the associated symptoms, such as arthritis. Aspirin (**choice B**) has been found to be much more effective, however, because of its anti-inflammatory effects and its antithrombotic effects. Aspirin alone, however, does not reduce the incidence of coronary artery aneurysm, which intravenous immunoglobulin (IVIG) does. As such, the first-line treatment for Kawasaki syndrome is combination therapy with IVIG and aspirin.

Plasmapheresis (**choice D**) and prednisone (**choice E**) generally are not used for the treatment of Kawasaki syndrome, though both may be used in rare instances as salvage therapy. Steroids, effective in most forms of vasculitis, have been associated with increased occurrence of coronary artery aneurysms. Plasmapheresis, on the other hand, has mainly been limited by technical considerations and the availability of a more tested medical treatment.

47. The correct answer is A. This patient has Turner syndrome, usually caused by absence or abnormality of an X chromosome, resulting at least functionally in a patient with a genotype of 45,XO. In addition to primary amenorrhea, sterility, classic physical findings of micrognathia, and a "shield chest," these patients are at an increased risk for coarctation of the aorta. Up to 20% of patients with Turner syndrome have this condition, which can cause primarily upper extremity hypertension.

Essential hypertension (**choice B**) is the most common cause of hypertension, accounting for 90 to 95% of cases. It is rare, however, to have essential hypertension in such a young patient. The associated physical findings point toward Turner syndrome, commonly associated with coarctation of the aorta, a well-described cause of secondary hypertension.

Ovarian failure (**choice C**) is part of Turner syndrome. Strands of nonfunctional connective tissue replace the ovaries. Ovarian failure, however, does not cause hypertension. Rather the associated cases of coarctation may result in hypertension.

Renal artery stenosis (**choice D**) and renal failure (**choice E**) are not commonly associated with this condition and are unlikely to be a cause of this patient's symptoms.

48. The correct answer is D. Spina bifida occulta is the most benign form of dysraphism. There is a defect of the closure of the posterior vertebral arches and laminae, usually at L5 and S1. The meninges do not herniate through the bony defect. The spinal defect is covered by skin rendering the underlying neurologic defect occult or hidden. These children do not have associated hydrocephalus or Chiari II malformations. Orthopedic malformations include extremity asymmetry and foot deformities. However, most children will be asymptomatic. Typically, a dermal sinus or patch of hair may be present over the defect. A spinal roentgenogram shows a defect in the closure of the vertebral arches. Lamina defects, hemivertebrae, scoliosis, widening of interpedicular spaces, and butterfly vertebrae may all be seen on x-ray. Surgical repair of these lesions is most effectively performed in a prophylactic fashion.

Ash leaf macules (**choice A**) are hypopigmented macules present on the skin of patients with tuberous sclerosis. They are usually the first visible cutaneous marker of this congenital disorder and may be present as a single or multiple lesions. Tuberous sclerosis patients may also develop angiofibromas of the face (adenoma sebaceum) and periungually (Koenen tumors). Calcifications may be present in the central nervous system, leading to a seizure disorder that usually begins in teenage or adolescent years. Often, there is mental retardation of variable degree.

A Becker nevus (**choice B**) is visible as a tan or brown macule present at birth, most commonly on the shoulder area. It is characterized by flattened and broad rete ridges that have an increased number of melanocytes present at the dermoepidermal junction. About 15% will have an associated intradermal melanocytic nevus. At puberty, thick, dark terminal hairs may develop within the lesion. There are no associated central nervous system abnormalities.

Mongolian spots (**choice C**) are flat blue or slate-gray lesions with well-defined margins, present at birth. Most commonly, they are located in the lumbosacral area, although they may appear on other parts of the skin. Mongolian spots are very common in dark-skinned races, especially Asian, where they are seen in up to 90% of newborns. A total of 5 to 10% of Caucasian newborns

will have one at birth. Most of these lesions resolve in the first few years of life, but in a small percentage of cases they will persist into adulthood. Histopathologically, melanin-containing melanocytes are seen in the dermis. Mongolian spots should be differentiated from the bruises of child abuse, which have variation of color within them and gradually fade into the surrounding skin.

Salmon patch (**choice E**), or nevus flammeus, is a common capillary malformation of the skin, present in at least 15% of newborns. It may persist indefinitely in 5% of newborns and grow in proportion to the growth of the affected body part. A pink-red macule is seen at birth on the glabella or on one upper eyelid, where they appear in about 15% of infants. In this location, nevus flammeus has a tendency to fade and disappear some time during childhood (as opposed to the "stork bite" lesion of the nuchal region, where it is somewhat more common and tends to persist indefinitely).

49. The correct answer is C. The key to the question is the patient's age. Legg-Calve-Perthes disease is a noninflammatory deformity of the proximal femur caused by a vascular insult, leading to osteonecrosis of the proximal femoral epiphysis. It is most commonly seen in boys aged 4 to 8 years with delayed skeletal maturity, but should be considered in any limping child 4 to 12 years of age. Patients may be pain-free or note knee, groin, or back pain. Decreased range of motion of the affected hip and a Trendelenburg gait are also common findings.

This patient is too old for developmental dysplasia of the hip (**choice A**) to be the most likely diagnosis because it is usually recognized within the first 2 years of life.

Although juvenile rheumatoid disease (**choice B**) is a possibility, it too is much less likely than Legg-Calve-Perthes disease. Juvenile rheumatoid arthritis usually affects patients older than the boy in this scenario (greater than 8 years of age) and is much more common in girls.

The patient does not appear septic or have any history of fever, chills, or severe pain with hip range of motion. Thus, septic arthritis (**choice D**) is highly unlikely.

Slipped capital femoral epiphysis (**choice E**) usually occurs in adolescents and is often associated with obesity or endocrine disorders.

50. The correct answer is B. Administration of hepatitis B vaccine and hepatitis B immune globulin immediately after birth appears to give the infant an initially high level of antibodies against the virus; and the vaccine, when all three doses of the vaccine have been administered, affords long-term protection. This is now considered standard care for an infant with a mother who is hepatitis B surface antigen–positive.

Administration of hepatitis B vaccine only (**choice A**), while definitely appropriate to administer to this child, will not result in an initially high level of protective antibodies in the newborn.

Administration of hepatitis B immune globulin only (**choice C**) affords only transient protection to the newborn and still exposes to the child to further infection later in life.

Explaining to the mother that no intervention is indicated (**choice D**) is untrue; furthermore, it could possibly prevent the administration of medications that could possibly protect the newborn from infection. It has also been shown that women who have surface antigenemia with a high level of infectivity have a >90% transmission rate from mother to fetus, as is the case in this scenario.

Waiting to repeat a serologic workup before treatment of the infant (**choice E**) denies the newborn the highest level of protection for postexposure prophylaxis. Current recommendations state that the immune globulin should be administered immediately after birth and the first dose of the vaccine within the first 12 hours of birth.