

FACTS – HEMATOLOGY/ONCOLOGY AND ENDOCRINE

Von Willebrand's disease is the most common hereditary bleeding disorder. It is inherited in an autosomal dominant fashion and affects 1% of the general population. vWf mediates adhesion of platelets to the blood vessel wall following vascular damage (it also serves as the carrier protein for plasma factor VIII and deficiency of vWf may cause a secondary deficiency in factor VIII). Most patients have mild to moderate bleeding, usually involving the mucocutaneous surfaces – excessive bruising, epistaxis, menorrhagia, and postoperative hemorrhage, particularly after mucosal surgery such tonsillectomy or wisdom tooth extraction. Laboratory findings reveal a prolonged bleeding time, a prolonged aPTT, and a low quantitative assay for vWf antigen and activity (ristocetin cofactor assay). Treatment includes DDAVP (induces vWf release from endothelial cells and is used for mild to moderate bleeding and for prophylaxis before surgery) and cryoprecipitate (contains intact vWf and is used for more severe bleeding or extensive surgeries).

Hemophilia is an X-linked recessive disorder that occurs in 1 in 5,000 males, with 85% having factor VIII deficiency and 10-15% having factor IX deficiency (Christmas disease). Clinical hallmark features in affected males include hemarthrosis (involving the knees, elbows, and ankles) and deep soft-tissue bleeding. CNS bleeding is the most dreaded complication and is usually the result of head trauma. Laboratory findings include a prolonged aPTT, normal PT/bleeding time/platelet count/platelet function assay, and low factor VIII protein activity in the presence of normal von Willebrand's factor assay. Treatment includes prevention of trauma and replacement of factor VIII or IX with factor VIII or IX concentrates (should increase plasma levels to 100% if any concerns of significant head trauma). DDAVP may cause the release of stored factor VIII from the patient's own cells and may be useful in mild hemophilia.

Sickle cell disease is caused by a single amino acid substitution of valine for glutamic acid on the number 6 position of the B-globin chain of Hgb. The mutation results in polymerization (stacking) of Hgb within the RBC membrane when the RBC is exposed to low oxygen or acidosis. Diagnosis of SS disease is now usually made at birth through state newborn screening programs with a hemoglobin electrophoresis. More common in African-Americans, clinical characteristics are not generally present until protective Hgb F declines (by 6 months of age). Clinical features include painful bone crisis (most common crisis, ischemia/infarction of bone or marrow, deep/gnawing/throbbing pain lasting 3 – 7 days, includes acute dactylitis – painful swelling of digits of the hands and feet), acute abdominal crisis (abdominal pain and distension from sickling within the mesenteric artery), stroke (dysarthria, hemiplegia), priapism (painful, sustained erection), acute chest syndrome (new pulmonary infiltrate associated with respiratory symptoms such as cough/SOB/chest pain with hypoxemia), sequestration crisis (rapid accumulation of blood in the spleen, occurring in patients less than 6 years of age, with abdominal pain and distension/pallor/ fatigue/ shock/ high mortality), and aplastic crisis (temporary cessation of RBC production often caused by parvovirus B19 with acute decreases in hemoglobin and reticulocyte count). Other presentations include serious bacterial infection (the leading cause of death, the result of

decreased splenic function, encapsulated bacteria – *Streptococcus pneumoniae*/*Haemophilus influenzae*/*Neisseria meningitidis*, osteomyelitis with salmonella species). Preventative care includes hydroxyurea (agent to increase Hgb F), daily oral penicillin prophylaxis, daily folic acid, routine immunizations that should include yearly influenza vaccine/23-valent polysaccharide pneumococcal vaccine/meningococcal vaccine, and serial transcranial Doppler ultrasounds to identify patients at risk for stroke. Median life expectancy is in the 40s with long-term complications including delayed growth and puberty, cardiomegaly, cor pulmonale, gallstones, poor wound healing, avascular necrosis of the femoral head, and diminished cognition in school performance.

ITP (idiopathic thrombocytopenic purpura) is the most common acquired platelet abnormality in childhood. It typically occurs 1–4 weeks after a viral infection in a perfectly healthy 1-4-year-old child with the abrupt onset of cutaneous bleeding (petechiae, bruising) or mucous membrane bleeding (epistaxis, gum bleeding). Laboratory findings reveal a thrombocytopenia with large platelets and normal WBCs/RBCs. Treatment includes supportive care and avoidance of medications that suppress platelet production or alter function (sulfonamides, aspirin ...). Very low platelet counts or active bleeding warrant treatment with IVIG, corticosteroids, or anti-D immunoglobulin. Platelet transfusions are generally avoided because transfused platelets are rapidly destroyed. Most cases resolve spontaneously within months, although chronic ITP occurs in 10 – 20% (more common in older children and is often treated with splenectomy).

TEC (transient erythroblastopenia of childhood) is the most common form of pure red blood cells aplasia in children. This acquired disorder occurs in previously healthy children who present with severe anemia, often between 1 and 3 years of age. This normocytic, normochromic anemia is slow in onset and presents with signs and symptoms of anemia. Laboratory findings include a decreased hemoglobin (average at presentation is 5.7 g/dL), decreased reticulocytes, and severe erythroid hypoplasia on bone marrow. All patients recover, with 60% requiring a transfusion for support until recovery. TEC must be distinguished from Diamond-Blackfan anemia (DBA), a much more serious disorder. DBA typically presents with a severe anemia in the first year of life, often with pallor recognized at or soon after birth. Approximately 25% of patients demonstrate physical anomalies (craniofacial, renal, cardiac, short stature, triphalangeal thumbs). Laboratory findings reveal a decreased hemoglobin (average of 4 g/dL at diagnosis), decreased reticulocytes, increased Hgb F, erythroid hypoplasia on bone marrow examination, and elevated red blood cell adenosine deaminase levels. Unlike TEC, DBA does not remit spontaneously and frequent RBC transfusions, corticosteroids, and bone marrow transplantation may be indicated.

Autoimmune hemolytic anemia (AIHA) occurs when antibodies are misdirected against the RBCs. Most cases (70 to 80%) occur in infants and young children and are preceded by a respiratory infection. Presenting features include the acute onset of pallor, jaundice, hemoglobinuria, and fatigue. The spleen is usually enlarged and is the primary site for destruction of IgG-coated RBCs. Laboratory findings show a severe anemia, spherocytes on blood smear, prominent reticulocytosis, and a positive direct Coombs test (detects coating of antibodies on the surface of RBCs or complement). Corticosteroids are typically the treatment of choice for severe anemia.

Macrocytic (megaloblastic) anemias are characterized by large RBCs with MCV >95. The two major causes in children are folic acid and vitamin B₁₂ deficiencies. Folic acid deficiency often presents with failure to thrive, chronic diarrhea, and irritability and is most often associated with infants who are exclusively fed with goat's milk – a low serum folic acid is diagnostic and treatment includes dietary folic acid. Vitamin B₁₂ deficiency presents with anorexia, a smooth red tongue, and neurologic manifestations (ataxia, hyporeflexia, positive Babinski responses) and is most often caused by inadequate dietary intake (strict vegetarian or vegan diet, an inherited inability to secrete intrinsic factor (juvenile pernicious anemia), or inability to absorb vitamin B₁₂ (terminal ileum condition such as Crohn's disease or a sequela of intestinal resection from NEC) – diagnosis is made by documentation of low serum vitamin B₁₂ level and treatment is by monthly intramuscular vitamin B₁₂ injections.

Hereditary spherocytosis is the most common inherited abnormality of the RBC membrane and occurs predominantly in persons of Northern European ancestry. There is a deficiency or abnormality of the structural RBC membrane protein spectrin that causes the RBC to assume its spherical shape. Inheritance is usually autosomal dominant. Signs and symptoms include splenomegaly by 2 to 3 years of age, pallor, weakness, jaundice, pigmentary gallstones, and aplastic crises, most commonly associated with parvovirus B19 infection. Infants may present with only prolonged jaundice and anemia. Laboratory findings show an elevated reticulocyte count, hyperbilirubinemia, spherocytes on blood smear, and abnormal RBC fragility with osmotic fragility studies. Treatment includes transfusions and eventual splenectomy, often delayed until after 5 years of age to decrease the incidence of invasive disease caused by encapsulated bacteria.

Glucose-6-phosphate dehydrogenase deficiency (G6PD) is the most common RBC enzymatic defect. This X-linked recessive disorder is most common in males of Mediterranean, Arabic, Asian, and African ethnic groups. G6PD enzyme is critical for protecting the RBC from oxidative stress and its deficiency results in RBC damage when the RBC is exposed to oxidants, including infection, fava beans, and several drugs (sulfa, salicylates, antimalarials). Significant hemolysis occurs 24-48 hours after exposure to an oxidant, with resultant abdominal pain, splenomegaly, jaundice, and significant anemia. Diagnosis is made by measuring the actual enzyme activity of G6PD in RBCs. Treatment involves transfusions as needed.

Fanconi anemia, or congenital aplastic anemia, is the most frequently reported of the rare inherited bone marrow failure syndromes. Clinical features include skeletal abnormalities (short stature and absence or hypoplasia of the thumb and radius), onset of bone marrow failure at a mean of age 7 years (presents with ecchymosis and petechiae), skin hyperpigmentation (including café-au-lait spots), and renal abnormalities. Treatment includes transfusions of RBCs and platelets as needed, and bone marrow transplant from an HLA-compatible donor, if available.

Wiskott-Aldrich syndrome is an X-linked recessive disorder characterized by the triad of combined immunodeficiency (recurrent infections with encapsulated organisms such as *H. influenzae* and *S. pneumoniae*), thrombocytopenia characterized by small platelets, and eczema. Diagnosis is made by CBC, revealing thrombocytopenia and small platelets, decreased IgM, defective antibody response to polysaccharide antigens, and defective cellular immune function. Although splenectomy can cure the thrombocytopenia in more than 90% of patients, the therapy of choice is human leukocyte antigen (HLA)-matched bone marrow transplantation.

Cyclic neutropenia is characterized by regular, periodic oscillations in the number of polymorphonuclear neutrophils in the peripheral blood. Fever, oral ulcers, and stomatitis may occur during the neutropenia, with cycles lasting an average of 21 days. Diagnosis is made by documenting the cyclic nature of the neutropenia by obtaining serial neutrophil counts during a two to three month period. Management requires prompt identification and treatment of infections, including careful attention to oral and dental hygiene, with possible prophylactic administration of granulocyte colony-stimulating factor. The most common cause of transient neutropenia in children is viral infection. Common viruses known to cause neutropenia include hepatitis A and B, influenza, measles, rubella, varicella, RSV, and EBV. Neutropenia usually develops within the first 24–48 hours of the illness and may persist for 1 to 2 weeks.

ALL (acute lymphoblastic/lymphocytic leukemia) is the most common childhood cancer with a peak incidence occurring at 2 – 6 years of age, more common in Caucasian males. Fever and bone or joint pain are the most common symptoms (may manifest as refusal to bear weight). Pallor, bruising, hepatosplenomegaly, and lymphadenopathy are the most common signs. Epistaxis, anorexia, fatigue, testicular pain and swelling, and abdominal pain may also be present. ALL is suggested by a CBC that demonstrates anemia and thrombocytopenia with variable WBC count (high in 1/3, normal in 1/3, and low in 1/3). Leukemic blasts are often seen and confirmation is by bone marrow evaluation demonstrating marrow replacement by lymphoblasts. Favorable prognostic factors at time of diagnosis include age 1 to 10 years of age, female, white, less than 50,000 cells/mm³, and no chromosomal translocation. Management involves three stages: induction (aims to destroy as many cancer cells as possible, often involves corticosteroids, vincristine, and L-asparaginase and intrathecal methotrexate, remission is induced in 95% of patients), consolidation (involves the continuation of systemic chemotherapy agents and prophylactic regimen to prevent CNS involvement), and maintenance therapy (daily and periodic chemotherapy during remission for up to 3 years). Complications during treatment often occur and include: infection (often life-threatening and associated with neutropenia – absolute neutrophil count less than 500 cells/mm³ – often with Gram-negative bacteria); opportunistic infections with HSV, *Pneumocystis carinii* pneumonia, and fungi; metabolic complications such as tumor lysis syndrome (hyperuricemia may result in renal insufficiency, hyperkalemia may result in cardiac dysrhythmias, and hyperphosphatemia may result in hypocalcemia with tetany), and medication-induced complications (pancreatitis from L-asparaginase and corticosteroids, cardiomyopathy from doxorubicin, and cystitis from

cyclophosphamide). Cranial irradiation may result in MR, learning problems, hormonal problems (growth delay, hypothyroidism, hypopituitarism), and secondary malignancy. The outlook for patients with ALL is generally good with an overall long-term survival occurring in 85% of patients.

AML (acute myelogenous leukemia) represents 15–20% of childhood leukemias and is frequently associated with Down syndrome or with ionizing radiation and can occur as a secondary malignancy resulting from chemotherapy – CNS involvement occurs more commonly in AML, and symptoms and signs include fever, hepatosplenomegaly, bruising and bleeding, gingival hypertrophy, and bone pain. Laboratory findings may include pancytopenia or leukocytosis, and diagnosis is suggested by clinical features and blood smear demonstrating leukemic myeloblasts with Auer rods. Confirmation is by morphologic analysis and immunophenotyping of cells obtained by bone marrow biopsy. AML requires very intensive therapy to induce remission and bone marrow transplant is recommended once patients are in remission.

Neuroblastoma is a malignant tumor of neural crest cells and may arise anywhere along the sympathetic ganglia chain and within the adrenal medulla. Neuroblastomas are the second most common solid tumor (after brain tumors), have a peak incidence in the first 5 years of life, and occur 75% of the time in the abdomen or pelvis, 20% in the posterior mediastinum, and 5% in the neck. Although most commonly presenting as a firm abdominal mass that often crosses the midline, clinical presentations vary depending on location of the tumor – mediastinal location leads to respiratory distress and/or an incidental radiographic finding; cervical location often leads to tracheal compression or Horner's syndrome (myosis, ptosis, anhidrosis); catecholamine effect such as flushing/hypertension/headaches/sweating; acute cerebellar atrophy with ataxia and opsoclonus/myoclonus ("dancing eyes,/dancing feet"); and metastatic disease such as periorbital ecchymosis and proptosis, skin nodules with "blueberry muffin" appearance, bone pain or limp, and hepatomegaly. Diagnosis is made by excessive excretion of urinary catecholamines (VMA and HVA) and a positive bone marrow biopsy (CT or MRI is generally used to assess tumor spread and skeletal survey or bone scan is used to assess for metastasis to bone). Management includes surgery +/- chemotherapy/radiation. Good prognosis occurs in children <1 year of age and in patients with stage 1 and 2 disease. Spontaneous regression without treatment may occur in young infants with stage 4S disease.

Wilms tumor (nephroblastoma) is a tumor of the kidney and is the most common childhood renal tumor. 75% of cases occur in children younger than 5 years of age. Associated genetic findings or syndromes include Beckwith-Wiedemann syndrome (hemihypertrophy, macroglossia, visceromegaly, omphalocele), deletion of the short arm of chromosome 11, and WAGR syndrome (Wilms tumor, aniridia, genitourinary abnormalities, and mental retardation). Wilms tumor most commonly presents with an asymptomatic abdominal mass often detected by a parent while bathing or dressing the child, but may also present with abdominal pain, hematuria, hypertension, or associated congenital anomalies (genitourinary malformations, hemihypertrophy, or sporadic aniridia). Diagnosis is made typically by abdominal ultrasound/CT/MRI, chest x-ray to rule out pulmonary metastasis, UA, and biopsy. Treatment includes prompt surgery and chemotherapy +/- radiation. Outcome is usually excellent, with an overall cure rate greater than 90%.

Malignant bone tumors in children are most often due to osteosarcoma and Ewing's sarcoma. Osteosarcoma is the most common malignant bone tumor and most often occurs in males during the peak of the rapid growth spurt of adolescence. 50% of tumors occur near the knee and are associated with pain, swelling, and a soft tissue mass. Radiographic findings reveal periosteal reaction with a "sunburst" appearance in the metaphyseal region of the bone. Ewing's sarcoma is the second most common malignant bone tumor and most often occurs in males during adolescence – it is characterized as a small, round, blue cell tumor. 95% have a chromosomal translocation between chromosomes 11 and 21. It most commonly affects the flat bones and diaphysis of tubular bones (axial skeleton, humerus, femur). Typical presentation includes pain, swelling, and a soft tissue mass with a periosteal reaction with "onionskin" appearance. Osteoid osteoma is a common benign bony tumor of childhood, often presenting in males from ages 5 to 24 years of age. It typically presents with a sharp, boring pain, often of the femur or tibia, that is worse at night, and almost without exception completely relieved by NSAIDs. Tenderness at the site of the osteoid osteoma and limp or atrophy of the involved limb may be noted. On plain radiographs, the tumor typically produces a radiolucent nidus with surrounding reactive bone. Although the lesion and this pain will resolve eventually, surgical excision of the nidus offers dramatic relief for patients who have persistent pain. Osteochondromas are the most common benign tumors in children, presenting as a painless, hard, and nontender mass that is usually located in the metaphysis of long bones and demonstrated radiographically as a bony outgrowth from the cortex.

Brain tumors are the second most common childhood cancer (after leukemia) and are the most common solid tumors, accounting for 20% of all childhood cancers. Glial cells are most common (40–60% of brain tumors) and include astrocytomas followed in frequency by primitive neuroectodermal tumors (PNETS), ependymomas, and craniopharyngiomas. Infratentorial tumors (located in the posterior fossa) are most common in children (medulloblastoma is the most common, followed by cerebellar astrocytoma and brainstem glioma). Initial nonspecific symptoms are caused by increased intracranial pressure and are often worse during sleep or upon awakening. Symptoms may include headaches, vomiting, drowsiness or irritability, abnormal behavior, ataxia, seizure, and head tilt. Physical exam may reveal an enlarged or bulging fontanelle, nystagmus, papilledema, cranial nerve abnormalities (especially 6th nerve palsy), and lethargy or irritability. Neuroimaging by CT or MRI is critical for diagnosis and management – MRI is generally preferred because it is better able to visualize the temporal lobes, cranial base, and brainstem. Surgery is the principal treatment followed with possible radiation therapy and chemotherapy.

Hodgkin's disease is a cancer of the antigen-processing cells found within the lymph nodes or spleen – it is associated with EBV infection. More common in older children and adolescents, Hodgkin's presents with painless lymphadenopathy, most commonly in the supraclavicular or cervical regions. The hallmark histologic feature is the Reed-Sternberg cell. Non-Hodgkin's lymphoma is a very aggressive cancer, slightly more common than Hodgkin's, and associated with immunodeficiency states such as HIV/Wiskott-Aldrich syndrome/prior EBV infection. There is a male predominance and the incidence increases

after 5 years of age. Painless lymphadenopathy in the abdominal, mediastinal, and supraclavicular nodes is the most common presenting feature. Lymphoma must be considered as a possible cause in any child older than 3 years of age presenting with intussusception. Burkitt's lymphoma is endemic in Africa where it presents as a jaw mass.

Hypothyroidism presents with a suboptimal growth velocity (less than 5 cm per year or 2 inches per year) with a delayed bone age, goiter, myxedema ("puffy skin"), and amenorrhea or oligomenorrhea in adolescent females. Congenital hypothyroidism is the most common metabolic disorder with an incidence of 1 in 4,000 births, most often caused by thyroid dysgenesis. Thyroid hormone is essential for normal brain growth during the first 2 years of life. Most newborns are asymptomatic at birth and have an unremarkable physical examination, but by several months develop lethargy, constipation, a large anterior and posterior fontanelle, protruding tongue, umbilical hernia, myxedema, mottled skin, hypothermia, delayed neural development, and poor growth, often with a history of neonatal jaundice and poor feeding. Treatment should begin immediately with L-thyroxine, and if treatment is delayed until signs are present, most patients will have suffered permanent neurologic sequelae (mental retardation). Hashimoto's disease (chronic lymphocytic thyroiditis) is an autoimmune disorder which is the most common cause of acquired hypothyroidism. Most common in females, the typical clinical features vary from asymptomatic to goiter to short stature to transient hyperthyroidism. Laboratory findings reveal a low free T4, increased TSH, and antithyroid antibodies (antithyroglobulin, antimicrosomal, or antiperoxidase antibodies). Management includes thyroid hormone replacement with L-thyroxine to normalize the TSH level.

Hyperthyroidism presents with clinical features including eye problems (lid lag and exophthalmos), thyroid gland enlargement, tachycardia with palpitations, skin warm and flushed, CNS problems (remarkable for nervousness and fine tremors with history of fatigue and difficulty concentrating in school), and pubertal problems (delays in menarche and gynecomastia in boys). Graves disease is an autoimmune disorder and is the most common cause of hyperthyroidism in childhood with a female predominance. Laboratory findings include increased T3 and T4 levels with suppressed TSH level in the presence of TSI (thyroid-stimulating immunoglobulin). Management includes antithyroid medications (PTU and methimazole are the two most commonly used medications – although both inhibit thyroid hormone synthesis, PTU also impairs the peripheral conversion of T4 to T3. These medicines are usually the first-line treatments), subtotal thyroidectomy (may be considered if antithyroid medication fails), and ablative therapy with radioactive iodine (often used in adolescents if noncompliance with medication is an issue, its use eventually results in permanent hypothyroidism, controversial because of long-term risks of radiation exposure).

Diabetes mellitus classically presents with polyuria, polydipsia, polyphagia, nocturia, and occasionally enuresis. With type 1 diabetes (insulin deficiency), symptoms progress to weight loss, vomiting, and dehydration with 25% of patients presenting in DKA. With type 2 diabetes (insulin resistant), obesity, acanthosis nigricans, and a strong family history are more common, often presenting in late adolescence or adulthood. Management includes insulin for type 1 diabetes mellitus and oral hypoglycemic agents for type 2. Long-term

complications include microvascular complications (diabetic retinopathy, nephropathy, neuropathy), macrovascular complications (adulthood-atherosclerotic disease, hypertension, heart disease, stroke), and DKA when ill or noncompliant. DKA (hyperglycemia usually greater than 300 mg/dL with ketonuria and a serum bicarbonate level less than 15 mmol/L) treatment is directed initially at restoration of circulating blood volume (most important early step with boluses of normal saline), correction of electrolyte disorders (especially the low total body potassium level), and normalization of excess serum levels of glucose, lipids, and ketones. The risk of cerebral edema usually occurs 6 to 12 hours into therapy with risk factors including patients younger than 5 years of age, initial drops in serum glucose levels faster than 100 mg/dL per hr, and fluid administration greater than 4L/m² per 24 hrs, with mortality rate being as high as 70%.

Growth hormone deficiency presents on the growth chart with a patient demonstrating poor growth velocity (less than 2 inches or 5 cm per year). Causes include brain tumors (craniopharyngioma most commonly), prior CNS irradiation, trauma, and congenital midline defects (single central maxillary incisor, cleft palate, microphallus). Imaging studies include a delayed bone age and MRI of the head to rule out a CNS lesion. Laboratory studies reveal a low IgF-1 level and poor response on growth hormone stimulation testing. Treatment includes daily subcutaneous injections of recombinant growth hormone.

Addison disease, the result of autoimmune destruction of the adrenal gland, usually presents with slowly evolving manifestations of deficiencies in cortisol, aldosterone, and sex steroid hormones. Onset occurs in late childhood/adolescence and is characterized by nausea, vomiting, diarrhea, abdominal pain, and cravings for salt. Anorexia, weight loss, muscle weakness, and fatigue are present. The clinical finding of increased pigmentation of the skin (accentuation of freckles, dark palmar creases, persistent "suntan") is the hallmark. Laboratory findings include hyponatremia, hyperkalemia, and hypoglycemia. The disease is characterized by a decreased level of cortisol and aldosterone and a high serum ACTH level. The most definitive diagnostic test is an ACTH stimulation test which reveals no increase in cortisol above resting levels. Management of an adrenal crisis requires immediate intervention to prevent a fatal outcome – normal saline boluses, glucose infusion, and glucocorticoid replacement with an IV bolus of hydrocortisone sodium succinate.

CAH (congenital adrenal hyperplasia), an autosomal recessive congenital enzyme deficiency in the adrenal cortex, is a classic example of primary adrenal insufficiency of childhood, and is the most common cause of ambiguous genitalia when no gonads are palpable. The enzyme deficiency leads to an underproduction of cortisol or aldosterone and a build-up of precursors that shunt into another pathway leading to increased production of androgens. The three main enzymes deficiencies include: 21-hydroxylase deficiency (accounts for 90% of cases, often salt-wasting, and presents in females with ambiguous genitalia or at 1 – 2 weeks of life in both boys and girls with FTT/vomiting/electrolyte abnormalities), 11B-hydroxylase deficiency (hypertensive and hypokalemic due to increased aldosterone precursors), and 3B-hydroxysteroid dehydrogenase deficiency. Patients with 21-hydroxylase deficiency have increased 17-hydroxyprogesterone levels. Management includes cortisone administered at

doses that sufficiently suppress ACTH production, mineralocorticoid replacement (florinef – if deficient with aldosterone), and frequent monitoring of growth velocity/physical examination/bone age. Prenatal diagnosis of CAH can be determined by molecular analysis of fetal DNA.

Cushing's syndrome, a characteristic pattern of obesity with associated hypertension, is a result of abnormally high blood levels of cortisol resulting from hyperfunction of the adrenal cortex. Causes include increased cortisol production due to adrenal tumors (including carcinomas), ectopic ACTH production by non-pituitary tumors, or exogenous glucocorticoids. A constellation of signs and symptoms is present: rounded face ("moon facies"), truncal obesity, stretch marks, "buffalo hump", masculinization, impaired growth, hypertension, hyperglycemia. Typical laboratory findings include elevated serum cortisol levels with loss of diurnal rhythm and failure to suppress cortisol with a dexamethasone suppression test.

Precocious puberty is the appearance of signs of pubertal maturation before age 8 in a girl (breast or pubic hair), or before age 9 in a boy (testicular enlargement, penile enlargement, or pubic or axillary hair). Premature thelarche is a common and benign condition that usually presents between 1 and 4 years of age with visible or palpable breast tissue only and no other secondary sex characteristics – it is a variation of normal pubertal development due to transient elevations in estrogen levels from either functional ovarian cysts or fluctuations in pituitary gonadotropin secretion – no work-up is necessary unless there is pubic hair development or a rapid growth spurt. Premature adrenarche is the early onset of pubic or axillary hair that occurs without the development of breast tissue or enlarged testes – more common in girls than boys, growth is normal without advancement of bone age, and no treatment is indicated. True/isosexual/central precocious puberty is the early onset of gonadotropin-mediated puberty with earlier than usual activation of the hypothalamus, presenting in females with breast development/pubic hair/rapid growth and in males with testicular enlargement/pubic hair/rapid growth. In girls, most cases are idiopathic. In boys, a pathologic cause for precocious puberty is identified in 90%. CNS abnormalities that may cause isosexual or precocious puberty include hydrocephalus, CNS infections, benign hypothalamic hamartomas, malignant tumors such as astrocytomas and gliomas, and severe head trauma. Evaluation includes elevations in FSH/LH/sex steroids, an advanced bone age, an MRI of the head in all boys and in very young girls with any neurologic symptoms, and a GnRH stimulation test revealing a dramatic increase in LH in response to injection of synthetic GnRH. Heterosexual gonadotropin-independent/peripheral precocious puberty is independent of the HPGA – caused by the peripheral production of male or female sex steroids and not FSH- or LH-mediated. (The hallmark of PPP is a flat response on GnRH stimulation testing because the HPGA has not been activated. Causes may differ in boys and girls, but in general include exposure to exogenous sex steroids, gonadal tumors, adrenal tumors, and nonclassic CAH. McCune-Albright syndrome is another well known cause – characteristic bony changes/polyostotic fibrous dysplasia, skin findings such as the "coast of Maine" café-au-lait spots, and endocrinopathies.

Metabolic syndrome is a constellation of findings that is characterized by ethnicity-specific waist circumference criteria or a BMI greater than 30 kg/M^2 plus two of the following: fasting triglyceride concentration greater than 150 mg/dL , HDL less than 40 mg/dL in men and 50 mg/dL in women, hypertension manifested by systolic blood pressure of 130 mmHg or greater and diastolic pressure of 85 mmHg or greater, fasting plasma glucose value of 100 mg/dL or greater or previously diagnosed type 2 diabetes. Definitions in childhood are not established, but they usually consist of similar criteria adjusted to age-appropriate values greater than 85th percentile for blood pressure, weight and waist circumference.

Turner syndrome occurs when only one X chromosome is present. Clinical features include short stature, webbed neck, shield chest, swelling of the dorsum of hands and feet (congenital lymphedema at birth), ovarian dysgenesis, cardiac defects including left-sided heart lesions (coarctation of the aorta and bicuspid aortic valve), and renal malformations (most commonly a horseshoe kidney). TS should be suspected in any girl who has unexplained short stature or delayed or absent puberty. Because of the high incidence of heart and kidney malformation, echocardiography and renal ultrasonography are warranted soon after birth.

DiGeorge syndrome is caused by a defect in the structures derived from the 3rd and 4th pharyngeal pouches. Clinical features include craniofacial findings (short palpebral fissures, small chin, and ear anomalies), cardiac findings (aortic arch anomalies, VSDs, tetralogy of Fallot), and thymus and parathyroid hypoplasia causing cell-mediated immunodeficiency and severe hypocalcemia. It often presents with a hypocalcemic seizure in a child with a heart murmur. Diagnosis is based on FISH probes to detect the deletion on chromosome 22.

Klinefelter syndrome is the most common cause of male hypogonadism and infertility. Chromosomal analysis reveals an XXY genotype. Risk increases with advancing maternal age. Clinical features include a male with tall stature and long extremities, hypogonadism with delayed puberty, gynecomastia, and variable intelligence and behavioral findings.

TEXAS TECH UNIVERSITY HEALTH SCIENCES CENTER
PEDIATRICS

QUESTIONS ON HEMATOLOGY/ONCOLOGY AND ENDOCRINE

1. A 5-year-old girl has had recurrent epistaxis and easy bruising. Laboratory evaluation reveals: prothrombin time, 11 seconds (control, 11 to 13 sec); activated partial thromboplastin time, 45 seconds (control, 28 to 36 sec); and platelet count, 240,000/mm³.

Among the following, the MOST likely cause of these findings is

- A. factor IX deficiency
- B. Glanzmann thrombasthenia
- C. lupus anticoagulant
- D. vitamin K deficiency
- E. von Willebrand disease

PLATELETS STICKS TO VESSEL, FACTOR 8 STABILIZATION

2. A 2-year-old boy presents for evaluation of a chronic pruritic eruption. His medical history is remarkable for recurrent epistaxis, otitis media, and pneumonia. Physical examination reveals erythematous, slightly scaling patches on the trunk and in the antecubital and popliteal fossae. Petechiae are present profusely.

Of the following, these findings are MOST suggestive of

- A. acrodermatitis enteropathica
- B. ataxia telangiectasia
- C. atopic dermatitis
- D. Langerhans cell histiocytosis
- E. Wiskott-Aldrich syndrome

X - LINKED, SEVERAL INFECTIONS, ECZEMA, COMBINED IMMUNODEFICIENCY
SMALL PLATELETS

3. In examining a 4-year-old girl who is new to your practice, you discover that she has rudimentary thumbs and is well below the 5th percentile for both weight and height. You also observe irregular hyperpigmentation on the trunk and anogenital areas.

Of the following, the MOST likely hematologic disorder associated with these findings is

- A. acute lymphoblastic leukemia
- B. VATER syndrome
- C. Diamond-Blackfan anemia
- D. Fanconi anemia
- E. thrombocytopenia and absent radii (TAR) syndrome

FANCONI'S SYNDROME → PROXIMAL TUBULE PROBLEM

FANCONI'S ANEMIA → 7-8 YEARS, BONE MARROW FAILURE

4. A 10-year-old girl complains of increasing fatigue several days after recovering from an upper respiratory tract infection. Findings on physical examination include obvious pallor, mild scleral icterus, and a spleen tip palpated 3 cm below the left costal margin. Laboratory studies reveal hemoglobin of 6.2 g/dL, reticulocyte count of 31%, and indirect bilirubin of 2.8 mg/dL. Red cell indices, white blood cell count, differential count, and platelet count are normal. Past medical history is unremarkable. She had a normal hemoglobin concentration at 1 year of age and has not had any further laboratory tests until this visit.

Of the following, in addition to a complete blood count, the MOST appropriate laboratory test to order for this girl is

- A. direct Coombs test *Auto immune (RBC vs virus confusion) ↑ retic count*
- B. glucose-6-phosphatase dehydrogenase (G6PD) assay
- C. haptoglobin concentration
- D. hemoglobin electrophoresis
- E. osmotic fragility *SPHEROCYTOSIS*
5. An African-American mother brings her previously healthy 2-year-old son to the emergency department because he looked pale when he woke up this morning. She also reports fever and decreased activity over the last few days. He was diagnosed with a urinary tract infection and given trimethoprim-sulfamethoxazole 3 days ago. On physical examination, he is markedly pale and appears jaundiced. His heart rate is 170 beats/min, and his respiratory rate is 30 breaths/min. The rest of his physical findings are unremarkable. Laboratory evaluation reveals a hemoglobin concentration of 5 g/dL.

Of the following, the MOST likely cause of this boy's anemia is

- A. aplastic anemia
- B. glucose-6-phosphate dehydrogenase deficiency
- C. hereditary spherocytosis
- D. sickle cell hemolytic crisis
- E. transient erythroblastopenia of childhood
6. A healthy 2-day-old boy born at term undergoes circumcision prior to discharge from the hospital. Bleeding was noted at the site of circumcision 10 hours after the procedure and has increased steadily over the past 4 hours. Findings on physical examination are unremarkable except for bleeding along 2 to 3 mm of the surgical site; there are no petechiae or purpura.

Of the following, the MOST likely cause of the bleeding is

- A. disseminated intravascular coagulation
- B. factor VIII deficiency hemophilia
- C. immune thrombocytopenia purpura
- D. neonatal alloimmune thrombocytopenia
- E. von Willebrand disease

7. A 10-month-old African girl who recently emigrated from Nigeria presents with tender swelling of her feet. Pertinent laboratory findings include a hemoglobin concentration of 9.7 g/dL and serum bilirubin of 3.5 mg/dL. Radiographs of her feet demonstrate some cortical thinning and irregular areas of radiolucency.

Among the following, the MOST likely diagnosis is

- A. juvenile rheumatoid arthritis
 - B. kwashiorkor
 - C. malaria
 - D. multifocal osteomyelitis
 - E. sickle cell disease
- DACTYLITIS: PAINFUL SWELLING OF HANDS AND FEET*
AFRICAN AMERICAN: SICKLE CELL

8. A 3-year-old girl recovering from a viral illness is pale and jaundiced. Findings include: splenomegaly; hemoglobin, 5.9 g/dL; mean corpuscular volume, 92 fL; mean corpuscular hemoglobin concentration, 37 g/dL (normal, 32.1 to 36.5 g/dL); white blood cell count, 13,500/mm³, with a normal differential count; platelet count, 431,000/mm³; and reticulocyte count, 34%. Her father and paternal uncles underwent cholecystectomies in their 20s.

Of the following, the MOST likely diagnosis is

- A. Blackfan-Diamond syndrome
 - B. glucose-6-phosphate dehydrogenase deficiency
 - C. hereditary elliptocytosis
 - D. hereditary spherocytosis
 - E. pyruvate kinase deficiency
- Young/NAIVE = CHOLECYSTECTOMIES*

9. A 10-month-old girl has had eight episodes of fever, mouth sores, and irritability over the past 6 months that last several days and resolve spontaneously. Physical examination reveals ulcers in the oral mucosa, gingivitis, and cervical adenopathy. Results of laboratory evaluation include: white blood cell count, 3,000/mm³; 3% neutrophils, 84% lymphocytes, and 13% monocytes on differential count; hemoglobin, 11.0 g/dL; and platelet count, 334,000/mm³.

The MOST important test(s) to obtain is (are)

- A. bone marrow examination
 - B. Epstein-Barr virus titers
 - C. Serial absolute neutrophil counts
 - D. Serum B₁₂ concentration
 - E. Serum folate concentration
- Cyclic neutropenia, 9-21 days cycles of blood counts*

10. An 8-year-old child is brought to you because of leg pain. The pain is greatest at night and is relieved by ibuprofen. There is some local tenderness over the thigh and slight atrophy of the affected limb. A radiograph of the femur reveals a radiolucent nidus surrounded by sclerotic bone, and a bone scan shows increased uptake in that area.

The MOST likely explanation for these findings is

- A. Ewing sarcoma - DIAPHYSIS, CHINA (11,22)T
- B. hairline fracture
- C. osteochondroma
- D. osteogenic sarcoma
- E. osteoid osteoma LOTS OF PAIN @ NIGHT, SENSITIVE TO IBUPROFEN

11. The parents of a 5-year-old boy who has severe factor VIII deficiency hemophilia bring him in for evaluation after he fell off his bicycle. He was not wearing a helmet. He had no loss of consciousness and appears uninjured except for a small hematoma over the right side of his forehead.

Of the following, the MOST appropriate next step is to

- A. admit for observation without therapy
- B. discharge home
- C. infuse with factor VIII BLEED LATE, MUST GIVE FACTOR VIII 100%
- D. obtain computed tomography
- E. treat with desmopressin

12. A 15-month-old boy develops fatigue and pallor over several weeks. Physical examination results are otherwise normal. Laboratory studies reveal: hemoglobin, 2.9g/dL; white blood cell count, 6,400/cu mm ($6.4 \times 10^9/L$); differential count, 43% neutrophils, 49% lymphocytes, and 8% monocytes; platelets, 298,000/cu mm ($298 \times 10^9/L$); mean corpuscular volume, 78 fL; reticulocyte count, 0.1%; indirect bilirubin, 0.2 mg/dL; and fetal hemoglobin, 2% (normal for age).

NORMAL MCV = 75-95

Of the following, the MOST likely diagnosis is

- A. anemia of chronic disease
- B. Diamond-Blackfan anemia CONGENITAL RBC ANEMIA, 1ST YEAR, INTENSIVE TX
- C. Fanconi anemia
- D. iron deficiency anemia
- E. transient erythroblastopenia of childhood 1ST COUPLE OF YEARS COMMON IN PEDIATRICS, RBC PRECURSORS STOP WORKING MOST COMMON PURE RBC APLASIA

13. A 6-year-old girl complains of pain in her legs. Physical examination reveals petechiae, purpura, and marked pallor. Her spleen is palpable 3 cm below the left costal margin. Laboratory findings include: hemoglobin, 6.3 g/dL; white blood cell count, 2,400/mm³; and platelets, 22,000/mm³.

Of the following, the MOST likely diagnosis is

- A. acute lymphoblastic leukemia
- B. aplastic anemia
- C. autoimmune hemolytic anemia
- D. Henoch-Schönlein purpura
- E. immune thrombocytopenic purpura

14. You are evaluating an 8-month-old infant for anemia. At 6 months of age, iron supplementation was prescribed when a complete blood count (CBC) revealed anemia. Follow-up CBC shows that the anemia has persisted, although the child's mother insists she has been giving the iron supplements. The infant has been exclusively bottle-fed with fresh goat's milk since age 4 months. Cereal and baby foods have not yet been introduced.

Of the following, the MOST likely cause for this infant's persistent anemia is

- A. folate deficiency ↓ IN GOATS MILK, MACROCYTIC, MEGALOBLASTIC ANEMIA
- B. iron deficiency
- C. vitamin B₁₂ deficiency
- D. vitamin E deficiency
- E. zinc deficiency

15. A 2-year-old girl presents for evaluation of fussiness, low-grade fever, and what her parents describe as "growing pains." On physical examination, you palpate a nontender mass deep in the right periumbilical area and note mild purple discoloration of the eyelids.

Of the following, the MOST likely diagnosis is

- A. hepatoblastoma
- B. ALL
- C. intussusception
- D. neuroblastoma METASTATIC TO EYES → NEUROBLASTOMA
- E. Wilms tumor MASS, BLOOD IN URINE, ↑BP

16. During a routine health supervision visit, you note several 1 to 1.5 cm purpuric lesions located over both tibias of a previously well 16-month-old boy. There is no purpura in other areas, no petechiae, and no mucosal bleeding.

Of the following, the most appropriate INITIAL diagnostic approach is to obtain

- A. a bleeding time
- B. a complete blood count with platelet count
- C. a factor VIII activity
- D. no laboratory studies
- E. the partial thromboplastin time and prothrombin time

17. A 6-year-old boy currently receiving chemotherapy for acute lymphoblastic leukemia presents with a fever of 38.8°C (102°F) and symptoms of an upper respiratory tract infection. He does not have an indwelling intravenous access catheter. Laboratory findings include: hemoglobin, 11.5g/dL; white blood cell count, 6,400/mm³; and platelets, 380,000/mm³.

Of the following, the MOST important study to obtain next is

- A. absolute neutrophil count
- B. cerebrospinal fluid analysis
- C. chest radiograph
- D. liver function studies
- E. urine culture

18. A 14-year-old boy has had increasing pain and swelling in his left thigh over the past 2 weeks. He has been afebrile. Peripheral blood counts are normal, but erythrocyte sedimentation rate is 75 mm/h. Radiography demonstrates an osteolytic lesion in the midshaft of the femur with a surrounding soft-tissue mass. There is an "onionskin" appearance to the bony lesion.

Of the following, the MOST likely diagnosis is

- A. aneurysmal bone cyst
- B. Ewing sarcoma
- C. lymphoma
- D. osteogenic sarcoma
- E. osteomyelitis

19. The mother of a previously healthy 4-year-old boy asks you to evaluate an abdominal mass she discovered on her son while bathing him. On physical examination, his blood pressure is 130/70 mmHg and his pulse is 80 beats/min. A large, firm, nontender mass is palpable in his right upper quadrant. Urinalysis is remarkable for the presence of microscopic hematuria.

Of the following, the MOST likely diagnosis for this child is

- A. hepatoblastoma
- B. neuroblastoma
- C. pheochromocytoma
- D. renal cell carcinoma
- E. Wilms tumor *Association w eye problems, ANEMIA, Assoc w Beckwith/Wiedemann*

20. You are evaluating a previously healthy 5-year-old boy who has a 2-day history of a febrile illness. On physical examination, his temperature is 101.3°F (38.5°C), and other vital signs are stable. He appears well, has no significant lymphadenopathy or hepatosplenomegaly, and has no focus of infection. A complete blood count demonstrates a hemoglobin of 12g/dL, white blood cell count of 2,800/mm³ with 25% granulocytes, and a platelet count of 200 x 10³/mcL (200 x 10⁹/L).

Of the following, the MOST appropriate management is to

- A. administer amoxicillin for 10 days
 - B. initiate a trial of granulocyte colony-stimulating factor
 - C. obtain a bone marrow aspirate
 - D. refer him to a hematologist
 - E. repeat the complete blood count in 1 to 2 weeks
21. You are seeing a 14-year-old girl who has sickle cell disease for a routine health supervision visit. She reports that she has had three episodes of right-sided abdominal pain over the past 2 months, one of which lasted several hours. The only remarkable finding on physical examination is scleral icterus. Laboratory evaluation demonstrates hemoglobin of 8.2 g/dL (82 g/L), reticulocyte count of 7.5%, total bilirubin of 4.2mg/dL, and conjugated bilirubin of 0.8 mg/dL.

Of the following, the MOST appropriate diagnostic study is

- A. abdominal computed tomography to evaluate for subacute appendicitis
 - B. abdominal ultrasonography to evaluate for cholelithiasis
 - C. blood culture to rule out *Salmonella* bacteremia
 - D. echocardiography to evaluate for right ventricular dysfunction
 - E. liver/spleen scan to evaluate for splenic infarction
22. An otherwise well 4-year-old boy is brought to your office because his mother has noticed bruising over the past 2 weeks. On physical examination he appears well and has no hepatosplenomegaly or adenopathy. There are scattered petechiae on the right upper arm, resolving bruises on the legs, and no evidence of new bruises. A complete blood count shows a platelet count of 50 x 10³/mcL (50 x 10⁹/L). The white blood cell and differential counts, hemoglobin, and hematocrit are normal.

ITP: Tx below 30 x 10³

Of the following, the BEST next step in the management of this patient is to recommend

- A. administration of intravenous immunoglobulin therapy
- B. administration of oral corticosteroid therapy
- C. avoidance of sulfonamides contribute to bleeding
- D. hospitalization for observation
- E. performance of a bone marrow examination

23. A 15-month-old infant has been breastfed since birth. He eats finger foods (eg, peas, carrots) and occasionally some cereal. His mother adheres to a vegan diet and plans the same for her child. A complete blood count documents anemia.

Of the following, the MOST likely cause of this infant's anemia is a deficiency of

- A. folic acid
- B. niacin
- C. riboflavin
- D. thiamine
- E. vitamin B₁₂ IF, VEGETARIAN DIETS, TERMINAL ILEUM

24. A newborn is recognized clinically to have Down syndrome. The parents are very concerned about the disorder and its manifestations.

Of the following, the MOST important hematologic/oncologic complication is

- A. aplastic anemia
- B. leukemia
- C. macrocytic anemia
- D. platelet dysfunction
- E. thrombocytosis

25. An otherwise healthy 18-month-old boy is at less than the 5th percentile for length; his length was at the 25th percentile at 6 months of age. His weight has been at the 10th percentile since birth. Findings on physical examination are unremarkable except for microphallus.

Of the following, the diagnostic study that is MOST likely to establish the etiology of this boy's decline in growth rate is a(n)

- A. bone age study
- B. complete urinalysis
- C. examination of stool for ova and parasites
- D. growth hormone assay MIDLINE DEFECTS: MICROPHALLUS, SINGLE CENTRAL TOOTH, CLEFT PALATE
- E. sweat chloride test

26. A 5-year-old girl is seen for respiratory distress. She is awake but fatigued. Findings include: tachypnea with rapid, deep respirations; dry mucous membranes; sunken eyes; blood glucose, 1,200 mg/dL; and serum ketones, strongly positive. She is placed on a rapid infusion of fluids with a continuous infusion of insulin. Six hours later she becomes unresponsive, coma ensues, and her posture becomes decorticate.

The MOST likely explanation for these findings is

- A. cerebral edema and herniation
- B. hypocalcemic tetany
- C. hypoglycemic seizures
- D. hypokalemic paralysis
- E. hypophosphatemic coma

27. A newborn has ambiguous genitalia characterized by posterior fusion of the labioscrotal folds and clitoromegaly. Findings include: chromosome analysis, a normal female 46,XX pattern; sodium, 127 mEq/L; potassium, 6.5 mEq/L; markedly increased levels of 17-hydroxyprogesterone and androstenedione. You diagnose the salt-wasting form of congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency.

When discussing options for future pregnancies, the MOST appropriate statement to include is that

- A. CAH is identified best by measurement of amniotic fluid 17-hydroxyprogesterone
 - B. prenatal diagnosis of CAH can be determined by molecular analysis of fetal DNA
 - C. the fetal gender should be determined by ultrasonography because only females are at risk
 - D. the mother should receive dexamethasone therapy throughout all future pregnancies
 - E. there is a 50% risk for an affected child in each future pregnancy
28. A 12-week-old female who was born at home has received no medical care. She has a coarse face, with puffy eyelids, thickened protruding tongue, and thick hair. Her cranial sutures are easily palpable, and posterior and anterior fontanelles are open. Her abdomen is protuberant, and an umbilical hernia is present. Her skin is cold to touch and mottled. No masses are palpable in the neck.

Of the following, the MOST likely long-term sequelae of this infant's condition is

- A. cerebral palsy
 - B. corneal opacities
 - C. deafness
 - D. hydrocephalus
 - E. mental retardation
29. A 6-year-old boy is being evaluated for an episode of generalized tonic-clonic jerking that occurred 2 days ago. His mother states that he did well in school last year, but his teachers report that he has had trouble concentrating over the past 4 months. Findings on physical examination include an ataxic gait, excessive motor activity, and difficulty following verbal commands, apparently from poor auditory processing. He is noted to have hyperpigmentation of the extremities and hyperreflexia too.

Of the following, the MOST likely diagnosis is

- A. acute cerebellar ataxia
- B. adrenoleukodystrophy
- C. attention deficit disorder
- D. encephalitis
- E. learning disability

30. A previously active 12-year-old boy fractured his pelvis in a motor vehicle accident and is immobilized in a cast. He complains of fatigue, decreased appetite, nausea, constipation, and polyuria with hematuria. His mother thinks he is more irritable than usual.

Of the following, the MOST likely cause of these complaints is

- A. hypercalcemia
- B. hyperkalemia
- C. hypermagnesemia
- D. hypocalcemia
- E. hypomagnesemia

31. An 18-year-old female college student presents for evaluation of fatigue. She has had difficulties concentrating on her studies and has lost 8 kg since her last visit 1 year ago. Physical examination reveals a thin woman with a body mass index of less than 15%. Her skin is a bronze color, with prominent freckles on her face, dark palmar creases, and dark-pigmented areolae. A urine pregnancy test is negative.

Of the following, the MOST likely diagnosis is

- A. Addison disease
- B. anorexia nervosa
- C. hyperthyroidism
- D. late-onset congenital adrenal hyperplasia
- E. polycystic ovarian syndrome

32. A 13-year-old girl presents with a history of palpitations, problems with sleeping, and recent weight loss despite increased appetite. Significant findings on physical examination include tachycardia, mild exophthalmos, a palpable thyroid, and a tremor.

Of the following, the MOST appropriate initial treatment for this adolescent is

- A. observation and re-evaluation
- B. oral methimazole
- C. oral thyroxine
- D. thyroid ablation with I^{131}
- E. thyroidectomy

33. A newborn female has a cardiac murmur. Before the cardiologist arrives to evaluate her, she has a seizure. Results of laboratory testing include a serum calcium level of 5.0 mg/dL. Subsequently, echocardiography reveals an aortic arch anomaly.

Of the following, the MOST appropriate test to obtain now is

- A. brainstem auditory evoked responses
- B. electroencephalography
- C. fluorescent in situ hybridization analysis of chromosome 22
- D. peripheral blood chromosome analysis
- E. thyroid function testing

37. A 17-year-old boy presents for a sports physical. He has a learning disability and is shy. His height is at the 75th percentile, and his body mass index is at the 85th percentile. Physical examination findings include minimal facial hair, bilateral gynecomastia (breast >4 cm in diameter), and small testes (testicular volume of 6 mL).

Of the following, the MOST likely cause of this patient's gynecomastia is

- A. constitutional delay of puberty
 - B. incomplete androgen insensitivity syndrome
 - C. Klinefelter syndrome
 - D. obesity
 - E. pubertal gynecomastia
38. A 3-year-old girl has isolated bilateral breast tissue development with no other signs of pubertal development, such as pubic hair. All growth parameters are at the 50th percentile.

Of the following, the MOST appropriate management is to

- A. measure serum gonadotropin levels
 - B. obtain computed tomography of the head
 - C. obtain pelvic ultrasonography
 - D. obtain radiography of the hand and wrist to determine bone age
 - E. reassure the parents that such breast development usually regresses spontaneously
- Premature thelarche : BREASTS - HAIR > NORMAL
HAIR - BREASTS > NORMAL
BREAST + HAIR = ABNORMAL
39. An 8-year-old girl presents to the emergency department with a history of vomiting, weight loss, and rapid breathing. Findings on physical examination include lethargy, tachycardia, tachypnea, cool extremities, and pallor. Initial laboratory evaluation reveals a venous pH of 7.10 and a blood glucose concentration of 650 mg/dL. Urinalysis is positive for glucose and ketones.

Of the following, the MOST immediate life-threatening complication of this child's illness is

- A. cerebral edema
- B. hyperkalemia
- C. hypocalcemia
- D. hypoglycemia
- E. hypovolemia

34. You are asked to evaluate a 9-year-old child who has short stature and truncal obesity. Her weight is at the 95th percentile and her height is at the 5th percentile for age. Reviewing her growth chart, you note that she had a normal growth pattern (weight and height at the 50th percentile) until 5 years of age when she began to gain weight dramatically.

Of the following, the MOST likely cause for the change in her growth pattern is

- A. Cushing syndrome
 - B. exogenous obesity
 - C. hyperthyroidism
 - D. inflammatory bowel disease
 - E. Prader-Willi syndrome
35. A 14-year-old girl has a 3-month history of a progressively enlarging mass in the left thyroid lobe. She has developed hoarseness over the past 2 weeks. Physical examination reveals a 3 cm firm mass in the left upper lobe of the thyroid gland. There is no associated adenopathy.

Of the following, the MOST helpful test for establishing the diagnosis is

- A. fine-needle aspiration biopsy of the thyroid gland
 - B. measurement of the thyroid-stimulating hormone level
 - C. measurement of the thyroxine level
 - D. radionuclide scan of the thyroid gland
 - E. ultrasonography of the thyroid gland
36. A couple schedules an interview with you prior to the birth of their first child. The expectant mother tells you that her pregnancy has been complicated by the finding of a cystic hygroma on prenatal ultrasonography at 14 weeks' gestation. The couple elected for amniocentesis, and chromosome analysis revealed a 45,X karyotype. The cystic hygroma subsequent has resolved.

Of the following, the two MOST important evaluations to perform soon after birth are

- A. echocardiography and renal ultrasonography *TURNERS SYNDROME, SHORT FEMALES, ANNOUATORY*
- B. head and renal ultrasonography
- C. head ultrasonography and echocardiography
- D. head ultrasonography and ophthalmologic evaluation
- E. ophthalmologic evaluation and echocardiography

40. A 13-year-old girl presents with the chief complaint of delayed puberty. She tires easily, and her school grades have begun to drop. Her growth velocity has decreased over the past 2 years. Thyroid function tests reveals thyroid-stimulating hormone, 12.2 mU/L (12.2 mcIU/mL) (normal, 0.6 to 6.3 mU/L [0.6 to 6.3 mcIU/mL]) and thyroxine, 58 nmol/L (4.5 mcg/dL) (normal, 72.0 to 150.6 nmol/L [5.6 to 11.7 mcg/dL]). Slight symmetric thyroid enlargement is apparent on physical examination.

The MOST likely cause for this girl's illness is

- A. chronic lymphocytic thyroiditis
- B. delayed-onset thyroid dysgenesis
- C. hypothalamic-pituitary hypothyroidism
- D. idiopathic simple goiter
- E. Turner syndrome

41. A 6-year-old girl who has type 1 diabetes presents with a 2-day history of fever, malaise, and diarrhea. She is well hydrated, and findings on physical examination are normal. The urine is positive for ketones, and the blood glucose level is 260 mg/dL. Electrolyte levels, including bicarbonate, are normal.

In addition to providing supplemental insulin as needed, the MOST appropriate management of this patient is to

- A. hospitalize her immediately
 - B. instruct her to check her blood glucose once a day
 - C. instruct her to check her urine for ketones once a day
 - D. instruct her to decrease her total caloric intake
 - E. instruct her to drink large amounts of sugar-free liquids
42. An afebrile 5-year-old boy has had a generalized seizure. Findings on his physical examination are normal. Laboratory evaluation reveals a low serum calcium level of 5.5 mg/dL (5.5 mmol/L), an elevated serum phosphorus level of 7.5 mg/dL, and a normal serum albumin level of 4.0 g/dL.

Of the following, the test that is MOST helpful in establishing the cause of the hypocalcemia is measurement of serum levels of

- A. alkaline phosphatase
- B. amylase
- C. magnesium
- D. parathyroid hormone
- E. vitamin D

43. A healthy 7-year-old Caucasian girl has bilateral breast tissue development and long, downy, pigmented hair over the labia majora, with a few coarse curly hairs. All growth parameters are at the 50th percentile. Findings on the remainder of the physical examination are normal.

Of the following, the MOST appropriate initial management for this patient is

- A. computed tomography of the abdomen
- B. computed tomography of the head
- C. pelvic ultrasonography
- D. radiography of the hand and wrist to determine bone age
- E. reassurance of the parents that such secondary sexual development is within the range of normal

FACTS - NEWBORN AND ADOLESCENCE

Hirschsprung disease occurs in approximately 1 in 5000 births and is a common cause of neonatal bowel obstruction. The typical presentation in the newborn period includes delay or failure to pass meconium, bilious emesis, and abdominal distension. The disease is characterized by a congenital absence of intramural ganglia in the affected segments of the bowel. Clues to a more proximal intestinal obstruction, such as duodenal or jejunal atresia, would include bilious or non-bilious vomiting, minimal abdominal distension, normal passage of meconium, and possible polyhydramnios. A characteristic double-bubble appearance on prenatal ultrasound is diagnostic of duodenal atresia, and 30% of affected infants have Down syndrome.

Tracheoesophageal fistula occurs in approximately 1 in 4000 neonates with the most common form being a blind pouch of the upper esophagus and a TEF connected to the distal esophagus. 50% of infants with TEF have associated anomalies, the most common of which is the VATER association (Vertebral defects, Anal atresia, TE fistula with atresia, and Radial upper limb hypoplasia and Renal defects). Infants who have TEF present with excessive oral secretions and frothing and bubbling at the mouth and nose with frequent episodes of coughing, choking, cyanosis, and respiratory distress. Feeding exacerbates the symptoms. Prenatal maternal polyhydramnios is a clue to the diagnosis. The diagnosis is further presumed when there is an inability or failure to pass a catheter into the stomach, and chest x-ray reveals a catheter coiled in the esophageal pouch. Because barium swallow poses a potential risk of aspiration, it should not be performed as an initial diagnostic test. Failure to pass a catheter through the posterior nose is presumptively diagnostic of choanal atresia (these infants also refuse to take the pacifier or bottle because closure of the mouth results in an inability to breathe).

Necrotizing enterocolitis is an acute fulminating, inflammatory disease, usually of the ileocolic junction, seen most often in premature babies. Typical presentation involves abdominal distension, feeding residuals, and bloody stools. The radiographic hallmark is intramural gas (pneumatosis intestinalis). Abdominal calcifications usually indicate intrauterine perforation of the intestine, most commonly seen with the meconium ileus that often accompanies cystic fibrosis.

Chest x-rays in response to respiratory distress in the neonate will often provide clues to the etiology: the finding of a bilateral diffuse reticulogranular pattern with superimposed air bronchograms is consistent with GBS neonatal pneumonia and hyaline membrane disease; over-inflation with coarse irregular densities is characteristic of meconium aspiration, often seen in post-term neonates with a history of perinatal asphyxia; prominent perihilar streaking with fluid in the fissures is consistent with transient tachypnea of the newborn, often seen in term neonates following C-section delivery, and treated with an enriched-oxygen environment until the pulmonary fluid clears and alveolar ventilation is accomplished, usually within 24 hours of birth.

Neonatal resuscitation should begin with the baby's body and head being dried quickly, using a pre-warmed towel to remove amniotic fluid. Drying prevents evaporative heat loss and provides gentle stimulation, which may initiate or help maintain breathing. If meconium is present, the OB should perform suctioning of the mouth and nasal pharynx prior to delivery of the shoulder/body to reduce the likelihood of aspiration, followed by tracheal suction after delivery of the body if the baby is hypotonic. If the neonate shows any signs of poor respiratory effort or bradycardia, PPV (positive-pressure ventilation) should be initiated. Chest compression should be started when the heart rate remains less than 60 beats per minute despite effective PPV. Epinephrine is indicated when the heart rate remains below 60 beats per minute despite at least 30 seconds of coordinated chest compressions and ventilation.

Hyperviscosity syndrome is a common and potentially dangerous situation that is most often caused by polycythemia, meaning a central venous hematocrit 65% or higher. This leads to plethora (a ruddy, deep red-purple appearance), hyperbilirubinemia, and cyanosis. Other findings include lethargy, tachypnea, hypoglycemia, and feeding disturbances. The incidence of polycythemia is highest in infants at high altitude, postterm, SGA, in the recipient of a twin-twin transfusion, in infants of diabetic mothers, in children with Down syndrome, and with delayed clamping of the umbilical cord. Symptomatic infants should undergo partial exchange transfusion involving removal of the infant's blood and replacement with isotonic fluid.

Vitamin K deficiency, in its classic form, presents most commonly from the 2nd to 7th day of life in healthy, breast-fed infants who have not received vitamin K prophylaxis (aka being born at home). The presentation includes melena, large cephalohematomas, intracranial hemorrhage, and bleeding from the umbilical stump, injection sites, or after circumcision. If bleeding is severe, the neonate may need FFP in addition to vitamin K.

Cyanotic congenital heart disease is most often caused by the five T's (truncus arteriosus, transposition of the great vessels, tricuspid atresia, tetralogy of Fallot, total anomalous pulmonary venous return). TGV is the most common cardiac cause of central cyanosis in the first month of life, often presenting without a heart murmur. TOF is the most common cardiac cause of central cyanosis presenting after the first month of life.

Neonatal abstinence syndrome is a term used to describe the withdrawal from opioids that occurs when an infant is born to a mother with a history of chronic opiate use/abuse. Infants typically present with a high-pitched cry, irritability, hyperactive reflexes, tremors and/or seizures, sweating, fever, poor feedings, and diarrhea. The most physiologic treatment for neonatal opiate withdrawal is oral paregoric. On the contrary, if mothers are given opiates like Stadol immediately prior to delivery only, then Naloxone, a narcotic antagonist, is often used in the delivery room to reverse the narcotic-induced respiratory depression, but its use in chronic opiate-exposed neonates may precipitate an acute onset of seizures.

Maternal tobacco smoking is often associated with growth failure (SGA) in a developing fetus. Maternal use of cocaine during pregnancy can cause placental abruption and fetal death.

Four components must be present to make a definite diagnosis of fetal alcohol syndrome – documented exposure to alcohol in utero, prenatal and postnatal growth deficiency (length, weight, and height), neurologic dysfunction (developmental retardation, ADHD, speech and language delay), and a typical set of dysmorphic facial features (hypertelorism, short palpebral fissures, a flat philtrum, midface hypoplasia, and a thin upper lip).

HIE (hypoxic-ischemic encephalopathy) due to perinatal asphyxia is the single most common cause of neonatal seizures in infancy, most often occurring within the first 24 hours after birth, typically clonic and multifocal. Intracranial hemorrhage accounts for approximately 10% of neonatal seizures (subarachnoid hemorrhage is typical in the term infant with onset of seizures occurring on the 2nd postnatal day; IVH typically occurs in the premature infant).

Congenital syphilis during pregnancy has a transmission rate approaching 100%, with fetal or perinatal death occurring in 40%. Among survivors, manifestations have traditionally been divided into early (during the first 2 years of life) and late (after 2 years of life) stages. The most common clinical manifestation of early congenital syphilis is periostitis. Late manifestations include Hutchinson triad (Hutchinson teeth, interstitial keratitis, cranial nerve VIII deafness), saddle nose, mulberry molars.

The most important determinant of neurodevelopmental outcome in a very low-birthweight infant is the length of gestation. Physical characteristics and neurologic development are used to assess gestational age – postterm (cracked, peeling skin; minimal if any vernix caseosa; and deep creases over the entire surface of the sole of the foot) and preterm (gelatinous, red translucent skin; few or no creases on the sole of the foot; minimal if any breast buds; and, in males, testes are undescended with faint rugae of the scrotum).

Chlamydia trachomatis pneumonia presents at 1 to 3 months of age with tachypnea, cough, inspiratory crackles, eosinophilia, a history of purulent conjunctivitis, and the absence of fever/wheezes. Chlamydia should be treated orally with a macrolide (the eyedrops do not prevent pneumonia).

Neonatal conjunctivitis (eye redness and discharge) is often diagnosed based on the age at presentation:

- Less than 48 hours – chemical conjunctivitis due to silver nitrate or Erythromycin eyedrops.
- 2 to 5 days – N gonorrhoeae
- 5 to 14 days – Chlamydia trachomatis

Infants of diabetic mothers are at increased risk of: hypoglycemia secondary to fetal pancreatic insulin secretion, polycythemia, RDS secondary to delayed surfactant production, hypocalcemia, and other malformations (particularly congenital heart disease/septal hypertrophy, and variants of caudal regression syndrome).

Adolescence is a time during which there may be several physiologic variations of normal:

1. Gynecomastia – breast tissue enlargement occurs in about 70% of adolescent males between ages 12 and 15, usually between Tanner stage 2 and 3. The breast tissue is 2 to 3 cm in size, firm, and mildly tender, often asymmetric. This is due to a temporary imbalance between circulating estrogens and androgens. In general, reassurance that this is a benign condition that will resolve in 1 to 2 years is all that is needed. If galactorrhea is present, then drug use, especially marijuana, should also be a consideration.
2. Leukorrhea – a physiologic vaginal discharge that usually precedes menstruation by 3 to 6 months. It consists of a white, odorless, mucoid discharge that may reveal sheets of vaginal epithelial cells without significant inflammation, with a low pH and gram stain showing rod-shaped organisms (lactobacilli). The management is reassuring the patient and her parents that this type of discharge is normal for her age.
3. Menstrual patterns can be quite variable in the first 2 years following menarche because most early menstrual cycles are anovulatory. Therefore, for a few years following menarche, it is common for an adolescent to experience irregular menstrual bleeding patterns, such as several months of amenorrhea or intermittent prolonged menses.

Persons younger than 18 years of age are considered minors. Typically, the consent of a parent or legal guardian is required before providing medical care to minors, although there are several exceptions. These exceptions include consent to emergency care, diagnosis/treatment/prevention of STDs, contraception, diagnosis/management of pregnancy, management of rape/sexual abuse, and the diagnosis/treatment of mental health problems including substance abuse. Certain minors who have achieved a certain status may also be able to consent for their own or their child's healthcare, including those who are living apart from their parents, are the parent of a child, or are in the armed services.

Adolescents are the age group with the highest rate of STDs. Gonorrhea and chlamydia can present with urethritis in males or with mucopurulent cervicitis and PID in females. Estimates of coinfection in adolescent women range from 25% to 60%, therefore treatment of females with cervical discharge should include effective treatment for gonorrhea (a single intramuscular dose of Ceftriaxone, or alternative therapies like Cefixime/Ciprofloxacin/Ofloxacin) and chlamydia (a single oral dose of Azithromycin, or consider Doxycycline/Erythromycin).

The three leading causes of death among adolescents in the United States are accidents, homicide, and suicide. Motor vehicles are responsible for the overwhelming majority of accidental deaths in the United States with alcohol being involved in approximately 40% of fatalities. The second most common type of unintentional accidental injury leading to death in adolescents is drowning, which often occurs in lakes and involves alcohol too. Homicide has become the leading cause of death for black adolescent males in our country and is the second most common cause of death among teenagers, with firearms being implicated more commonly than knives. The suicide rate of adolescents has tripled over the past several decades, with the increase in suicide deaths most pronounced in white males. Girls are 4 times more likely than boys to attempt suicide, but boys are 4 times more likely than girls to die from a suicide attempt.

Bulimia nervosa is an eating disorder that is characterized by binge eating. These recurrent episodes, which are defined as consuming an amount of food larger than most people would eat in a discrete period, happen in addition to some inappropriate compensatory activity to prevent weight gain, such as self-induced vomiting, laxative abuse, misuse of diuretics or enemas, fasting, or vigorous exercise. The affective component of this disorder is the lack of control during the bingeing episodes followed by depression or self-deprecating thoughts. These should occur at least two times per week for three months to meet the diagnosis of bulimia. Classic clinical manifestations of this illness include salivary gland enlargement secondary to stimulation by binge eating and vomiting, dental enamel erosion by gastric acids, bruises or calluses over the dorsum of the 2nd to 5th metacarpophalangeal joints due to self-stimulation of the gag reflex, wide fluctuations in body weight with intermittent lower extremity edema, and electrolyte abnormalities such as hypokalemic, hypochloremic metabolic alkalosis in patients with a history of chronic vomiting or acidosis if a history of chronic laxative abuse. The management team should include the primary pediatrician, a psychologist, and a nutritionist. Indications for hospitalization might include failure of outpatient treatment, dehydration, severe electrolyte disturbances, and EKG abnormalities (prolonged QT interval, severe bradycardia, dysrhythmias).

Anorexia nervosa is characterized by an internal fear of becoming obese not diminished by weight loss, distortion of body image, refusal to maintain body weight over age/height minimum, and amenorrhea in postmenarchal females due to hypothalamic/pituitary dysfunction. The chronic emaciation affects the myocardium leading to bradycardia, decreased QRS amplitudes, prolonged QT intervals, and nonspecific ST segment changes – these are the most likely causes of sudden death in anorectics.

Constitutional delay of growth in adolescence (CDGA) occurs in the majority of adolescents who exhibit pubertal delay and short stature. It is characterized by a normal or near-normal rate of linear growth below but parallel to the 5th percentile during the prepubertal years, delayed skeletal and sexual maturation during adolescence, and a normal final adult height. In CDGA, the bone age always is delayed, usually is equal to the height age, and is less than the chronological age. The prepubertal growth pattern of children who have familial short stature mirrors that of children who have CDGA. However, children who have familial short stature usually experience a normal onset of puberty, have a bone age that is consistent with chronologic age, and have a final adult height that is short.

Polycystic ovarian syndrome (PCOS) is the most common cause of persistent irregular menses in women of reproductive age. Typical characteristics are related to increased androgen production and anovulation. Chronic hyperandrogenism may present as hirsutism, acne, or rarely clitoromegaly. Hirsutism is defined as the growth of coarse hair in the androgen-dependent areas of the body, including the sideburn area, chin, upper lip, periareolar area, chest, low abdomen, and thighs. Increased androgen production in PCOS often leads to elevated serum concentrations of testosterone and androstenedione. Most of these patients will be obese.

Turner syndrome is characterized by short stature and primary amenorrhea. These females may also have a history of congenital lymphedema, webbed neck, heart problems (coarctation of the aorta), kidney problems (horseshoe kidney). The work-up of a child with short stature and absent menstruation by age 16 should include a karyotype.

Puberty is the development of secondary sexual characteristics and the maturation of gonadal function.

Females – the normal age range of onset of puberty in females is 8 to 13 years old, with the average being 10.5 years. The first sign of puberty in girls is breast enlargement, with the normal progression going from breast buds (thelarche), followed by the appearance of pubic hair, growth spurt, and then menarche. Pubarche follows thelarche by about 6 months. The growth spurt starts soon after thelarche and reaches peak growth velocity 1 year later (fairly early in pubertal progression), which is 1 year before menarche. Menarche typically occurs 2 years after thelarche begins, usually at around 12 years, 9 months and during stage 4 Tanner development. If breast development is at stage 5 without any pubic hair, then one should consider androgen insensitivity (testicular feminization). Conversely, the lack of breast development when pubic hair development is complete suggests an excess of androgens, a lack of estrogen, or congenital absence of breast tissue.

Males – the normal age range of onset of puberty in boys is 9.5 to 13.5 years, with the average being 11.5 years. The first sign of puberty in boys is enlargement of the testes, with the normal progression being enlargement of the testes, then the appearance of pubic hair, then the linear growth spurt. In males, the peak height velocity or maximum linear growth is a late pubertal event that typically occurs during Tanner stage 4, which is often 1 to 2 years later than the growth spurt in females.

TEXAS TECH UNIVERSITY HEALTH SCIENCES CENTER
PEDIATRICS

QUESTIONS ON NEWBORN AND ADOLESCENCE/GYNECOLOGY

1. A term newborn develops bilious vomiting at 48 hours of age. She has not passed meconium, and her abdomen is distended. Physical examination shows a normal appearing perineum. No other anomalies are apparent.

Of the following, the MOST likely diagnosis is

- A. annular pancreas
 - B. anorectal malformation
 - C. duodenal atresia
 - D. Hirschsprung disease
 - E. jejunal atresia
2. A newborn delivered at 36 weeks' gestation has excessive oral secretions and choking spells. The maternal history is notable for polyhydramnios. The infant's abdomen is somewhat distended and tympanitic.

Of the following, the BEST procedure to evaluate this infant's symptoms is

- A. attempt to pass a catheter through the posterior nose
 - B. attempt to pass a nasogastric tube into the stomach
 - C. barium swallow
 - D. fiberoptic endoscopy
 - E. transillumination of the chest
3. A 3-week-old preterm neonate, who has been receiving full enteral nutrition, develops abdominal distension, feeding residuals, and bloody stools. Physical examination reveals abdominal tenderness, hypothermia, apnea and lethargy. An abdominal radiograph (Figure A92A) is obtained.

Of the following, the radiographic finding MOST specific for the diagnosis of necrotizing enterocolitis is

- A. abdominal calcification - *not common unless ruptured*
- B. bowel wall thickening
- C. distended bowel loops
- D. intramural gas (*Pneumatosis intestinalis*)
- E. intraperitoneal fluid

4. A newborn who weighs 4,700 g and whose estimated gestational age is 37 weeks has a blood glucose concentration of 18 mg/dL (1.00 mmol/L) at 30 minutes after birth. Maternal history is significant for type 1 diabetes mellitus. Blood count reveals polycythemia. Echocardiographic examination reveals septal hypertrophy of the heart.

Of the following, the MOST important cause of hypoglycemia in this infant is

- A. abnormal gluconeogenesis
 - B. abnormal glycogenolysis
 - C. depleted liver glycogen stores
 - D. excess catecholamine secretion
 - E. increased pancreatic insulin secretion
5. A preterm neonate has tachypnea, expiratory grunting, nasal flaring, subcostal retractions, and cyanosis shortly after birth. The mother had rupture of membranes 36 hours prior to vaginal delivery and has developed uterine tenderness and fever.

Of the following, the MOST likely chest radiographic finding in this infant is

- A. diffuse reticulogranular pattern
 - B. displacement of the mediastinum
 - C. fine curvilinear lucencies
 - D. overinflation with coarse densities
 - E. prominent perihilar streaking
6. A 3.7-kg male infant is delivered at 38 weeks' gestation by scheduled repeat cesarean section to a 24-year-old multigravid woman who has intact fetal membranes. The Apgar score is 8 at both 1 and 5 minutes. Physical examination of the infant at 10 minutes after birth reveals mild intercostal retractions and a respiratory rate of 80 breaths/min. He is acyanotic and has a peripheral oxygen saturation of 88% on room air. There is no heart murmur. A chest radiograph reveals expansion of the lungs to nine anterior ribs, perihilar streaking, and a fluid density in the right horizontal fissure.

Of the following, the BEST therapy for this infant is

- A. intravenous ampicillin and gentamicin
- B. intravenous furosemide
- C. intravenous prostaglandin E1
- D. supplemental oxygen by hood
- E. tracheal intubation and surfactant

7. A 4-hour-old term infant has blue hands and feet. His extremities are slightly cold to touch, and chest auscultation reveals normal heart and breath sounds.

Of the following, the MOST appropriate plan of action is to

- A. measure blood viscosity
 - B. measure methemoglobin concentration
 - C. observe the infant
 - D. obtain chest radiography
 - E. order echocardiography
8. A newborn is delivered at 32 weeks' gestation by emergent cesarean section following spontaneous placental abruption. During resuscitation in the delivery room, the infant receives 100% oxygen and positive pressure ventilation through an endotracheal tube at a rate of 60 breaths/min. She has adequate chest excursions, but remains poorly perfused. Auscultation reveals a heart rate of 54 beats/min.

Of the following, the MOST appropriate next step in resuscitation is to

- A. administer a bolus of albumin
 - B. administer intratracheal epinephrine
 - C. begin chest compressions *PPV - CHEST COMP - EPI*
 - D. infuse intravenous sodium bicarbonate
 - E. inject intramuscular naloxone hydrochloride
9. A term infant is noted to be plethoric at birth. Pregnancy and delivery were uncomplicated. Initial laboratory findings of hemoglobin, 25 g/dL and hematocrit, 74% on a capillary sample are confirmed on a venous sample.

Of the following, the complication MOST likely to occur in this infant is

- A. hyperbilirubinemia
- B. hyperglycemia
- C. hypophosphatemia
- D. leukocytosis
- E. sepsis

10. A 5-day-old breastfed term neonate born at home develops bruising and gastrointestinal bleeding. He is otherwise well. Laboratory findings include: partial thromboplastin and prothrombin times, greater than 3 min; serum bilirubin, 5.6 mg/dL; platelet count 240,000/mm³; and hemoglobin concentration, 17 g/dL.

Of the following, the MOST likely cause of the bleeding is

- A. disseminated intravascular coagulation
- B. factor VIII deficiency hemophilia
- C. factor IX deficiency hemophilia
- D. liver disease
- E. vitamin K deficiency

DM 2 to 7

11. A 3-hour-old term infant has cyanosis, no cardiac murmur, tachypnea, and no retractions. Pulse oximetry reveals an oxygen saturation of 60% in the right hand and 75% in the right foot.

Of the following, the MOST likely cause of the cyanosis is

- A. group B streptococcal sepsis
- B. persistent fetal circulation
- C. pulmonary valve atresia
- D. tetralogy of Fallot
- E. transposition of the great vessels

12. A term newborn who is small for gestational age is irritable and inconsolable. Physical examination reveals generalized hypertonia, posturing, tremors, exaggerated startle responses, and skin excoriations. His cry is high-pitched, and his suck and swallow are weak and uncoordinated. The maternal history is significant for use of opiates throughout the pregnancy.

Of the following, the MOST appropriate drug for management of this infant is

- A. chlorpromazine
- B. diazepam
- C. naloxone
- D. paregoric
- E. phenobarbital

13. Among the following, the MOST common adverse effect of maternal smoking on the fetus is

- A. abruptio placentae
- B. cleft lip and palate
- C. decreased birthweight
- D. hypospadias
- E. intrauterine death

14. The mother of an infant being relinquished for adoption is said to have consumed alcohol regularly throughout pregnancy. The adoption agency asks you to determine whether the child has fetal alcohol syndrome.

Which of the following constellations of features BEST describes the fetal alcohol syndrome?

- A. Elfin facies, irritability, and supra-aortic stenosis *WILLIAMS SYNDROME*
- B. Growth deficiency with microcephaly, developmental delay, and short palpebral fissures
- C. Intrauterine growth retardation, triangular-shaped face, and clinodactyly of the fifth finger *RESSEK SYNDROME*
- D. Short stature, webbed neck, and pulmonic stenosis *NOVEMBER*
- E. Weakness, club feet, immobile face, and inadequate respirations

15. You are attending the emergency delivery by cesarean section of a primiparous woman. The gestation was complicated by pregnancy-induced hypertension. Deep variable fetal heart rate decelerations were noted during labor. At delivery, the infant is acrocyanotic with poor tone; spontaneous movement and minimal respiratory effort are present.

Of the following, your INITIAL management is to

- A. ascertain the heart rate and assign a 1-minute Apgar score
- B. begin tactile stimulation and provide blow-by oxygen supplementation
- C. dry all skin surfaces and clear the oropharynx
- D. initiate bag-mask ventilation
- E. insert an umbilical catheter and administer naloxone

16. A term newborn presents with bilious vomiting shortly after birth. Her abdomen is distended slightly, and facial features are characteristic of Down syndrome. She has passed a normal meconium stool. The pregnancy was complicated by polyhydramnios.

Of the following, the MOST likely diagnosis is

- A. duodenal atresia
- B. Hirschsprung disease
- C. meconium ileus
- D. midgut volvulus
- E. pyloric stenosis

17. A 4.0-kg term infant is delivered vaginally 6 hours after rupture of fetal membranes. The amniotic fluid is stained with thick meconium.

If the infant is limp, pale, and cyanotic at birth, the BEST sequence of care will be to

- A. administer bag/valve/mask positive pressure ventilation immediately after delivery
- B. have the delivering physician notify the physician caring for the infant of the history after completing the delivery
- C. suction the infant's mouth and hypopharynx after delivery and administer blow-by oxygen
- D. suction the infant's mouth and hypopharynx after delivery of the head and assess the need for further resuscitation after the 1-minute Apgar score is assigned
- E. suction the infant's mouth and hypopharynx after delivery of the head, followed by tracheal suctioning after delivery of the body

18. A newborn infant is delivered by emergent cesarean section at 41 weeks' gestation following a pregnancy complicated by a prolapsed umbilical cord and meconium-stained amniotic fluid. At 6 hours of age, the infant has a generalized tonic-clonic seizure.

Of the following, the MOST likely explanation for this seizure is

- A) hyponatremia
- B. hypoxic-ischemic encephalopathy
- C. intracranial hemorrhage
- D. meningitis
- E. pyridoxine dependency

19. A young woman who had a negative serologic test for syphilis in her first trimester is retested at 34 weeks' gestation. Her serologic test is now positive, and she is treated with penicillin for 2 days before spontaneous delivery of the infant.

Of the following, the clinical finding in this newborn that would be MOST consistent with the diagnosis of congenital syphilis is

- A. interstitial keratitis
- B. intrauterine growth retardation
- C. periostitis
- D. pneumonitis
- E. sensorineural deafness

20. Very low-birthweight (<1,500 g) infants are at increased risk for neurodevelopmental abnormalities from conditions such as intraventricular hemorrhage, periventricular leukomalacia, retinopathy of prematurity, and hearing deficit.

Of the following, the MOST important determinant of neurodevelopmental outcome is

- A. antenatal obstetric management
 - B. infant gender
 - C. length of gestation
 - D. maternal education
 - E. socioeconomic status
21. Term pregnancies extend from 38 to 42 weeks after conception. Pregnancies beyond 42 weeks are referred to as postterm.

Of the following, the physical finding on the newborn examination that would be MOST characteristic of a postterm infant is

- A. absence of creases on the soles of the feet
 - B. body weight below the 10th percentile
 - C. cracked, peeling skin
 - D. elevated weight-to-length ratio
 - E. excessive vernix caseosa
22. A 900-g male infant is delivered vaginally to a woman who had no prenatal care.

Of the following, the physical finding that is MOST consistent with prematurity rather than intrauterine growth retardation is

- A. creases over entire sole of foot
- B. descended testes with deep rugae of the scrotum
- C. formed and firm pinna with instant recoil
- D. gelatinous translucent skin
- E. raised areola and 3 mm breast buds

23. A 2-week-old infant is admitted to the hospital due to poor feeding, intermittent vomiting, and lethargy. He is evaluated for sepsis, oral feedings are stopped, and he is given maintenance intravenous fluids and antibiotics. After 3 days, all culture results are negative, and the baby is alert. Feedings are restarted using a cow milk formula, and 2 days later the baby is obtunded. Results of laboratory tests show metabolic acidosis with elevated anion gap, elevated lactate, mild hypoglycemia, hyperammonemia, and neutropenia.

Of the following, the MOST likely diagnosis is

- A. lactose intolerance
- B. celiac sprue
- C. organic acidemia
- ~~D.~~ galactosemia (6/6/2 sepsis)
- E. fatty acid oxidation defect

24. Physical examination of a 6-week-old afebrile infant reveals a respiratory rate of 74 breaths/min and a few crackles on inspiration. The 17-year-old mother, who identifies several sexual partners in the past, reports that the infant has had a loud cough but otherwise has been well except for some yellow eye drainage a few weeks ago.

Among the following, the MOST likely cause of the infant's findings is

- A. *Bordetella pertussis*
- B. *Chlamydia trachomatis*
- C. Influenza type A virus
- D. Respiratory syncytial virus
- E. *Neisseria gonorrhoeae*

25. A 13-year-old boy is seen in your office because of breast enlargement over the past 6 months. He denies pain, galactorrhea, and marijuana use. He is taking no medications. Physical examination reveals sexual maturity rating (Tanner) stage 2 genitalia and pubic hair growth and asymmetric breast buds beneath each nipple, with the left measuring 1 cm and the right 3 cm.

Of the following, the most appropriate INITIAL management for this boy is

- A. computed tomography of the head with enhancement
- B. magnetic resonance imaging of the abdomen
- C. serum beta-human chorionic gonadotropin level
- D. reassurance that this is a normal occurrence
- E. ultrasonography of the testes

26. You are discussing with a group of medical students the circumstances under which minor adolescents legally are permitted to provide consent for health care.

Of the following, the statement that you are MOST likely to include in your discussion is that in most states, a minor adolescent

- A. cannot consent to confidential care for drug or alcohol abuse
- B. cannot consent to medical care, even if living apart from his or her parents
- C. is authorized to provide consent for the medical care of his or her child
- D. is not permitted to consent to testing for sexually transmitted diseases
- E. may consent to pregnancy termination without parental notification or consent

27. The PEAK height velocity in boys occurs

- A. 1 year before the peak velocity in girls
- B. At the time the voice begins to deepen
- C. Prior to peak muscle mass development
- D. When testicular volume is 3 mL
- E. When the sexual maturity rating is stage 4

28. Menarche is MOST likely to occur

- A. at 14 years of age
- B. 1 year before peak height velocity
- C. 2 years after thelarche
- D. When body fat reaches 12% of body weight
- E. When the sexual maturity rating is stage 2

29. An 18-year-old girl complains of heavy menstrual periods that last 8 to 10 days. Physical examination reveals a creamy, greenish vaginal and cervical discharge that contains many leukocytes and Gram-negative diplococci in pairs. A pregnancy test is negative and syphilis serology is pending.

Of the following, the BEST antibiotic therapy for this patient is

- A. amoxicillin
- B. amoxicillin and erythromycin
- C. ceftriaxone
- D. ceftriaxone and azithromycin
- E. spectinomycin and doxycycline

30. The three LEADING causes of death among adolescents in the United States are

- A. accidents, cardiovascular disease, malignancy
- B. accidents, homicide, suicide
- C. cardiovascular disease, homicide, suicide
- D. homicide, malignancy, suicide
- E. homicide, infections, malignancy

31. An otherwise healthy 12-year-old boy is at the 5th percentile for height.

Of the following findings, the one that BEST supports a diagnosis of constitutional growth delay is

- A. bone age of 12 years
- *B. growth velocity of 5 cm/y
- C. high gonadotropin levels
- D. sexual maturity rating 3 pubic hair
- E. testicular length of greater than 2.5 cm

32. A previously healthy 13-year-old girl has secondary amenorrhea. Menarche occurred at 12 years of age and periods occurred monthly until approximately 3 months ago. She has Tanner or sexual maturity rating (SMR) 4 breast development and SMR 3 pubic hair development. Findings on physical examination are unremarkable. Urine pregnancy test is negative.

Of the following, the NEXT step in the management of this patient who has secondary amenorrhea is to

- A. measure serum androgen levels
- B. measure serum gonadotropin levels
- C. measure serum thyroxine concentration
- D. obtain pelvic ultrasonography
- o E. schedule follow-up in 3 months

33. Physical examination of a 17-year-old girl reveals swelling of both the parotid and submandibular glands, epigastric tenderness and intermittent edema. Serum electrolyte concentrations include: sodium, 131 mEq/L; potassium, 2.3 mEq/L; chloride, 90 mEq/L; and bicarbonate, 32 mEq/L.

Of the following, the best INITIAL step in the management of this adolescent is to

- *A. ask her about her eating behaviors
- B. measure the serum amylase concentration
- C. obtain a consultation for upper endoscopy
- D. order a barium swallow
- E. prescribe oral potassium supplements

34. You are seeing a 10-year-old girl for a health supervision visit. Her mother asks how much of an increase in her daughter's height to expect annually and whether this amount will change during puberty.

Of the following, the height velocities that are MOST likely to occur are

	<u>Average Prepubertal</u>	<u>Adolescent Growth Spurt</u>
A.	1 to 2 cm/y	5 to 6 cm/y
B.	3 to 4 cm/y	7 to 8 cm/y
C.	5 to 6 cm/y	9 to 10 cm/y
D.	7 to 8 cm/y	11 to 12 cm/y
E.	9 to 10 cm/y	13 to 14 cm/y

35. You are evaluating a healthy 16-year-old boy for short stature. His growth chart reveals height measurements following the 5th percentile and weight measurements following the 25th percentile. Findings on physical examination include Sexual Maturity Rating (Tanner) stage 2 pubic hair and stage 3 genitalia. You diagnose constitutional delay of growth and adolescence.

Radiographic studies MOST likely would show a bone age

- A. Equal to the chronologic age
 - B. Greater than the chronologic age
 - C. Greater than the height age
 - D. Less than the chronologic age
 - E. Less than the height age
36. An obese 16-year-old girl comes to your office complaining of infrequent menstrual periods since menarche at age 13. Her menstrual periods occur every 2 to 3 months and last 3 to 4 days. She denies a history of visual changes, headaches, or increased facial or body hair. Significant findings on the physical examination include velvety, hyperpigmented verrucous skin (Figure Q157A) on the nape of the neck and in the axillae and scattered erythematous papules and pustules (Figure Q157B) over her cheeks and shoulders. She has Sexual Maturity Rating (Tanner stage) 5 breasts and genitalia.

Of the following, the study MOST likely to have abnormal results is a(n)

- A. computed tomographic scan of the adrenals
- B. serum glucose level
- C. serum prolactin level
- D. serum testosterone level
- E. ultrasonographic scan of the pelvis

37. A 15-year-old girl is worried because she has not begun menstruating. Breast growth began at age 14. Her mother and sister began menstruating at age 12. The girl's weight is at the 25th percentile for age, and her height is below the 5th percentile. Small breast buds are noted on physical examination, but there is no pubic hair.

Of the following, diagnostic studies are MOST likely to show

- A. abnormal karyotyping
- B. decreased serum testosterone
- C. decreased thyroid stimulating hormone
- D. elevated erythrocyte sedimentation rate
- E. elevated serum prolactin

38. A 12-year-old premenarchal girl has had a persistent vaginal discharge for 3 months. Physical examination findings include Sexual Maturity Rating (Tanner) stage 3 breasts and genitalia. The hymen is annular and smooth. A thin, white, odorless discharge is noted in the introitus. Microscopic analysis of the fluids reveals vaginal epithelial cells, rod-shaped organisms, and a few white blood cells.

Of the following, the MOST appropriate treatment for this girl is

- A. azithromycin orally
- B. cefixime orally
- C. clotrimazole cream vaginally
- D. metronidazole orally
- E. reassessment in 2 to 3 months

Physiologic
Leukorrhea

FACTS – CARDIOVASCULAR AND RESPIRATORY

Kawasaki disease (mucocutaneous lymph node syndrome) is an acute febrile vasculitis of childhood. It is most common in children of Asian descent, presents in kids younger than 5 years of age, and is the most common cause of acquired heart disease in children in the U.S. (has replaced rheumatic heart disease). Diagnostic criteria include fever lasting at least 5 days plus 4 of the 5 following: bilateral nonpurulent conjunctivitis, oropharyngeal changes, cervical adenopathy, truncal rash, changes in the distal extremities including edema/redness/desquamation of palms and soles. The primary risk is the development of coronary artery aneurysms in untreated patients. Treatment includes anti-inflammatory therapy with IVIG and aspirin.

Marfan syndrome is an autosomal dominant connective tissue disorder that affects primarily the ocular (upward lens subluxation), cardiovascular (aortic root dilatation with or without dissection/mitral valve prolapse/aortic regurgitation), and skeletal findings (tall stature, arachnodactyly, joint laxity, chest wall deformities, scoliosis). The most serious cardiovascular abnormality associated with Marfan's is aortic root dilatation with possible dissection or rupture leading to pericardial tamponade and sudden death – chronic beta-blocker treatment has been shown to slow the progression of aortic root enlargement. Homocystinuria is an amino acid metabolism defect that presents very similar clinically to Marfan's with the exception that these children have downward lens subluxation and a hypercoagulable state (increased risk of stroke, myocardial infarction, and DVTs).

The incidence of congenital heart disease is about 0.5%-1% of the general population. Acyanotic CHD includes VSD, ASD, and PDA. VSDs are the most common congenital heart disease and usually present with a harsh holosystolic murmur that is best heard over the lower left sternal border, often presenting between birth and 2 months of age and becoming progressively louder as the pulmonary resistance physiologically drops. As the size of the VSD decreases, the intensity of the murmur increases. Most infants who have large ventricular defects exhibit tachypnea, tachycardia, gallop rhythm, sweating with feedings, diminished volumes of feedings, hepatomegaly, and failure to thrive at about the second to third month of life. (One of the congenital obstructive left heart lesions should be suspected in an infant who presents with congestive heart failure outside of the newborn nursery but within the first month of life, including hypoplastic left heart syndrome, critical neonatal aortic coarctation, and critical aortic valve stenosis.) ASDs often present outside of infancy and in older children with a LUSB murmur related to the increased pulmonary blood flow, as well as a fixed split heart sound resulting from the relatively prolonged time for the dilated right ventricle to empty its contents during systole. PDAs present with a continuous, "machinery-like" murmur that is loudest at the LUSB. Closure of a PDA can be accomplished with IV indomethacin (for symptomatic neonates with normal platelet counts and renal function) or transcatheter closure (preferred in infants who weigh more than 2 kg).

Supraventricular tachycardia is a relatively common cardiac emergency among infants and presents with irritability, poor feedings, tachycardia (heart rates above 220 beats per minute) and decreased peripheral pulses. The best means of converting this rhythm to sinus rhythm is to block atrial to ventricular conduction within the AV node, both by vagal maneuvers or drugs. Applying an ice bag to cover the entire face of an infant for 5 to 10 seconds is a very effective and first-line method to create a powerful vagal nerve discharge and block the AV node. Other means of controlling SVT include IV adenosine, digoxin, verapamil, and synchronized direct current cardioversion.

Innocent murmurs are very common in the pediatric population. A Still's murmur is the most common – presenting at 2-7 years, this SEM is best heard at the mid-left and left lower sternal border and is described as vibratory or musical in quality, loudest when the child is supine. A venous hum is another common innocent murmur that is often continuous and best heard at the upper sternal borders under the clavicles, usually when the child is upright and disappears when the child is supine or turns the head far to either side.

Acrocyanosis (cyanosis restricted to the hands and feet) is common in healthy infants and is thought to be caused by vasomotor instability or circulatory sluggishness. Cyanosis in young infants is detected most reliably in the mucous membranes, tongue, and lips. The most common cause of central cyanosis in the first month of life is transposition of the great vessels. The most common cause of cyanosis after the first month of life is tetralogy of Fallot (infundibular pulmonic stenosis, RVH, VSD, and overriding aorta). Children who have cyanotic CHD are at risk for developmental delay and poor school performance.

Prostaglandin E₁ is a potent dilator of the ductus arteriosus and is indicated to treat neonates who have life-threatening forms of congenital heart disease, especially if patency of the ductus is essential:

1. It is the only source of pulmonary arterial blood flow (pulmonary atresia).
2. It is the only source of systemic arterial blood flow (aortic atresia, hypoplastic left heart syndrome, severe coarctation of the aorta).
3. It is the only means of bidirectional mixing (complete transposition of the great vessels).

Down syndrome (trisomy 21) children carry about a 50% risk of having congenital heart disease, primarily AV septal defects (endocardial cushion defects), VSDs or ASDs. Accordingly, the AAP recommends that all infants who have Down syndrome receive a cardiac evaluation to exclude congenital heart disease. Other genetic syndromes are also associated with certain heart defects (Turner's – coarctation of the aorta, Marfan's – aortic dissection and mitral valve prolapse, Kawasaki's – aortic aneurysms, William's – supraaortic stenosis, congenital rubella – pDA, Noonan's – pulmonic stenosis, Holt-Oram – ASD).

The most common arrhythmia of the fetus and neonate is isolated atrial ectopy. Asymptomatic nonconducted premature atrial beats are very common in healthy neonates and present as intermittent pauses in an otherwise normal cardiac rhythm. The EKG will reveal premature P waves within the ST segment of the QRS complex that precedes the pause, and when these beats occur this soon, the AV node is still refractory and does not conduct it through to the ventricle.

Pericarditis is an inflammation of the pericardial space around the heart, most often caused by viral infections (Coxsackievirus, echovirus, adenovirus, influenza, parainfluenza, and EBV). Symptoms include fever, dyspnea, malaise, and chest pain most intense while supine and relieved with sitting upright. Physical examination includes a pericardial friction rub, distant heart sounds if the effusion is large, pulsus paradoxus, and hepatomegaly. Myocarditis is an inflammation of the myocardium and is one of the more common causes of sudden death in young athletes. It often follows a viral or flu-like illness with symptoms such as dyspnea, malaise, and abdominal complaints like nausea, vomiting, and anorexia. Physical examination may show resting tachycardia, gallop heart rhythm, hepatomegaly, tachypnea, and pulmonary rales. Costochondritis is a much more common cause of chest pain – this benign condition causes mild to moderate anterior chest pain, usually unilateral, localized with reproducible tenderness on palpation over the costochondral junctions, sharp in quality, exacerbated by deep breathing, and resolving spontaneously with or without NSAIDs.

Long QT syndrome is a cardiac condition that may cause patients to present with cardiac arrest (ventricular tachycardia, torsades de pointes, or ventricular fibrillation), syncope, seizures, presyncope or palpitations. The syndrome may be inherited as an autosomal dominant condition (Romano-Ward syndrome) or in an autosomal recessive pattern with deafness (Jervell and Lang-Nielsen syndrome). The diagnosis is made on the basis of an EKG showing a long QT interval greater than 0.44 seconds. Treatment of symptomatic patients includes a beta-blocker to reduce the symptoms.

Rheumatic fever is a nonsuppurative complication of group A streptococcal infections of the throat that occur in children between the ages of 5 and 15 years. This inflammatory disease affects the heart, CNS, joints, subcutaneous tissue, and skin, and is the second most common cause of acquired heart disease in the United States and a significant cause of cardiovascular morbidity in developing countries. Major criteria, also known as the Jones criteria, include carditis, polyarthritides (migratory), subcutaneous nodules, erythema marginatum, and Sydenham's chorea, with minor criteria including arthralgia, fever, increased acute-phase reactants, and a prolonged P-R interval. The carditis may involve any of the three layers of the heart: the endocardium, myocardium, or pericardium. When signs or symptoms are present, they most often are the result of endocarditis, which most often affects the mitral valve and leads to regurgitation (apical, holosystolic murmur that radiates to the left axilla).

Certain congenital heart lesions are believed to have such a low risk for developing endocarditis that prophylactic antibiotics prior to dental and other procedures seem unwarranted. Congenital lesions in this category include unrepaired secundum atrial septal defect, successfully repaired secundum atrial septal defect, ligated PDA, and successfully repaired VSD. 80% of cases of infective endocarditis occur in children who have structural abnormalities of the heart, most often caused by *Streptococcus viridans* and Staphylococcus species. Antibiotic prophylaxis is recommended before invasive procedures including dental work likely to produce bleeding, surgery (including tonsillectomy), and invasive GI or urologic procedures (this applies to all patients with structural heart disease except secundum ASD; all postoperative cardiac surgery patients for up to 6 months after surgical repair; all postoperative cardiac surgery patients for an indefinite period if any hemodynamic residua of the initial lesion remain).

Syncope is a sudden and transient loss of consciousness and postural control. Although it may be the presenting symptom for a complex and serious medical problem (cardiac arrhythmia, structural cardiac anomaly, neurologic disorder), most adolescents who present with syncope have no overt underlying disease. Neurocardiogenic syncope is the most common form of syncope in the adolescent and is almost always benign. It results from an inappropriate output from the autonomic nervous system that leads to symptomatic changes in heart rate, blood pressure, or both. This occurs most commonly while the person is standing, particularly for prolonged periods of time, but it also can occur with emotional stress or with noxious stimuli such as the sight of blood. Many patients have a prodrome of dizziness, nausea, and change in sound perception or vision, often appearing pale, clammy and confused just prior to the loss of consciousness. An EKG should be performed to look for evidence of heart block, QRS abnormalities, prolonged QT interval, sinus node dysfunction, or the presence of a pre-excitation syndrome such as WPW.

The primary structural CHDs that can lead to shock in the neonate rely on the patency of the PDA to provide systemic blood flow and include critical aortic stenosis, aortic coarctation, and hypoplastic left heart syndrome. If there is turbulent flow, such as in aortic stenosis or coarctation, there is a corresponding murmur. No turbulent flow, and thus no murmur, may be apparent in patients who have hypoplastic left heart syndrome. Critical aortic stenosis presents with a RUSB murmur and weak distal pulses; aortic coarctation presents with blood pressure elevation in the upper extremities and decreased in the lower extremities before the onset of CHF; HLHS presents with a single loud second heart sound, increased right ventricular precordial activity, and a relatively high room air oxygen saturation for cyanotic CHD (not too much cyanosis) due to the fact that the right ventricle can deliver a high saturation of blood to both the aorta and to the pulmonary arteries.

Hypertrophic cardiomyopathy is LVH in the absence of any systemic or cardiac disease known to cause the hypertrophy. The most typical anatomic finding is asymmetric septal hypertrophy (also termed idiopathic hypertrophic subaortic stenosis). It is the most common cause of sudden death in athletes. Symptoms may be absent until syncope or sudden death occurs or may include chest pain and exercise intolerance. Physical exam reveals a classic harsh, systolic ejection murmur at the apex that is accentuated with physiologic maneuvers such as Valsalva or going from squatting to standing. Its inheritance is autosomal dominant in 60% of cases. Diagnosis is confirmed by EKG (showing ventricular hypertrophy) and echocardiography.

Stridor is a high-pitched respiratory noise caused by turbulent airflow through a constricted passage, described as inspiratory, biphasic, or expiratory. Congenital stridor usually is present at birth, and the most common etiologies include laryngomalacia (70%), bilateral vocal cord paralysis (20%), and subglottic stenosis (5%). These common causes of congenital stridor are diagnosed by fiberoptic laryngoscopy and bronchoscopy. Laryngomalacia presents with stridor shortly after birth, often between 1 and 2 months of age, which worsens during supine positioning, increased crying or agitation, or during an intercurrent viral illness, characterized by an omega-shaped epiglottis that prolapses during inspiration, and improving spontaneously by 2 years of age. Vocal cord paralysis, in addition to inspiratory stridor, causes a weak cry, hoarseness, coughing, and choking, often caused by an underlying condition (congenital central lesions, birth trauma, prior surgical procedures that may damage the recurrent laryngeal nerve like thyroid/cardiac/TEF surgery). Subglottic stenosis manifests as a biphasic stridor with no voice abnormalities and may be congenital or acquired following intubation. Expiratory stridor is most commonly caused by tracheomalacia. Other causes of stridor include vascular compression from an aberrant innominate artery that can compress the anterior trachea or a vascular ring (double aortic arch) that can compress both the trachea and esophagus, often diagnosed by barium swallow, showing a posterior indentation of the esophagus. In patients who have suspected vascular airway compression, MRI or CT is used to define abnormal vascular anatomy. Another cause of progressive stridor is a subglottic hemangioma that typically enlarges over the first few months of life – patients are asymptomatic at birth, then develop biphasic stridor, with gradual involution to complete resolution – fluoroscopy most commonly reveals a unilateral subglottic mass and 50% of affected children have an associated cutaneous hemangioma.

Tuberculosis, caused by *Mycobacterium tuberculosis*, is most common among urban, low-income, and minority groups, with the highest risk including immigrants from highly endemic regions of the world, healthcare personnel, homeless individuals, residents of correctional facilities, and individuals with immunodeficiency. Symptoms include fever, chills, weight loss, cough, hemoptysis, and night sweats. TB infection is a term used to describe an asymptomatic individual with a positive PPD, normal physical exam, and normal chest radiography, vs. tuberculosis disease is a term to describe an individual with signs and symptoms of TB with or without positive findings on chest radiograph.

Extrapulmonary TB disease may present as cervical lymphadenitis (scrofula – the most common form of extrapulmonary TB disease in children), meningitis, skeletal disease (vertebral Pott's disease), and disseminated/miliary disease. The best specimen for the diagnosis of pulmonary TB in any young child or in any older child or adolescent in whom the cough is nonproductive or absent is an early morning gastric aspirate – three aspirates should be submitted, with smears for acid-fast bacilli and culture. Treatment for TB infection is isoniazid for 9 months. Treatment for TB disease includes a regimen of isoniazid, rifampin, and pyrazinamide.

Bronchiolitis is a lower respiratory tract illness in which the bronchioles are obstructed with mucus and edema. Clinical features include a mild upper respiratory tract infection with rhinorrhea, low-grade fever, and congestion, followed within one to three days by wheezing, worsening cough, and dyspnea, with occasional coarse crackles. Radiographic findings include hyperinflation and patchy atelectasis. More than 50% of cases are caused by RSV. On the contrary, infants born to mothers infected with *Chlamydia trachomatis* may present with tachypnea and a persistent, "staccato" cough at 1 to 3 months of age. These infants are typically afebrile, do not wheeze, and have crackles present on physical examination. They often have a history of purulent conjunctivitis.

Congenital diaphragmatic hernia presents with early respiratory distress and a scaphoid abdomen. 90% of cases involve a left-sided hernia with subsequent hypoplasia of the left lung and displacement of the heart to the right of midline, leading to hypoplasia of the right lung too.

Asthma is an inflammatory disorder of the lower airways that is characterized by a chronic nocturnal cough and expiratory wheezing, exacerbated by activity and during a URI. Asthma usually develops in early childhood with 80% of patients reporting symptoms prior to age 6 years. Because albuterol works by bronchodilating the entire lung, areas that are already bronchodilated open to a greater degree than the more constricted areas, thus overinflating and receiving more air while receiving the same perfusion – this leads to ventilation and perfusion mismatching. Chest radiography may reveal overinflation with atelectasis (mimics pneumonia, but has volume loss).

Cystic fibrosis is a multisystem disorder that results in altered exocrine gland secretions, affecting 1 in 2500 Caucasians. Common presentations include meconium ileus at birth, recurrent or chronic respiratory symptoms including pneumonia (*Staphylococcus aureus* and *Pseudomonas aeruginosa*), and malabsorption/steatorrhea/FTT. Diagnosis is most commonly made by an elevated sweat chloride greater than 60 mmol/L. The most common cause of death is bronchiectasis.

Kartagener syndrome (primary ciliary dyskinesia) consists of the triad of chronic sinusitis, bronchiectasis, and situs inversus. Recurrent acute otitis media and nasal polyps are also common. This syndrome is due to an abnormality of the cilia in which absent dynein arms are noted on electron microscopy. The resulting ciliary dysfunction leads to mucus stasis and frequent sinopulmonary infections.

Foreign body aspirations often have a history of a choking episode followed by coughing or wheezing, but such a history is not always available. The physical examination typically reveals tachypnea, asymmetric breath sounds, localized wheezing, or even normal lung findings. Chest radiographs may show normal findings, unilateral hyperinflation, atelectasis, or lobar infiltrate. Older children may cooperate for inspiratory and expiratory radiographs which may accentuate an area of air trapping – lateral decubitus films showing continued air trapping may be more useful for young patients. Rigid bronchoscopy should be performed if there is a strong suspicion for foreign body aspiration.

Epiglottitis is an acute inflammation and edema of the epiglottis, arytenoids, and aryepiglottic folds. This disorder is most common in children 2 to 7 years of age and historically is associated with *Haemophilus influenzae* type b. Clinical features include the abrupt onset of rapidly progressive upper airway obstruction with high fever and toxic appearance, muffled speech and inspiratory stridor, dysphagia with drooling, and sitting forward in a tripod position with neck hyperextension. Complete airway obstruction with respiratory arrest may occur suddenly. A lateral neck radiograph shows the characteristic “thumbprint” sign. Management involves controlled nasotracheal intubation performed in the safest manner possible by an experienced team that includes anesthesiologists and otolaryngologists. Bacterial tracheitis is an acute inflammation of the trachea, most often caused by *Staphylococcus aureus*. The most common presentation is a preceding viral illness followed by the abrupt onset of a rapid respiratory decompensation with high fever, toxicity, mucus and pus in the trachea. Affected children tend to lie flat and do not drool (these findings distinguish this condition from epiglottitis). Chest x-ray reveals an irregular tracheal air column that suggests purulent tracheal debris. Bronchoscopy generally is recommended to establish a diagnosis and to remove thick purulent material to improve the airway and obtain material for culture. Most children are treated with appropriate anti-staphylococcal antibiotics and respiratory support.

Empyema should be suspected when children who have an acute pneumonia fail to respond to therapy or in whom transient improvement is followed by fever, progressive respiratory distress, and physical signs of pleural effusion (dullness on percussion, decreased or absent breath sounds). Empyema is a parapneumonic effusion caused by spread of the inflammatory process to the visceral pleura and the pleural space. The development of a dense layer of exudate on the surface of the lung and the presence of walled-off cavities containing infected fluid can restrict lung function and cause persistence of signs and symptoms of infection, including fever, cough, chest pain, and respiratory distress. Gram-positives (*Streptococcus pneumoniae*, *Staphylococcus aureus*, and group A streptococcus) are the most common bacterial pathogens responsible for empyema.

Mycoplasma pneumoniae is one of the most common causes of pneumonia in older children and adolescents, presenting with low-grade fever, chills, nonproductive cough, headache, pharyngitis, and malaise. Lung examination may demonstrate widespread rales. Diagnosis is suggested by the presence of positive cold agglutinins, but a definitive diagnosis is made by mycoplasma serology. Management includes oral erythromycin or azithromycin.

Apnea is described as a respiratory pause without airflow lasting more than 20 seconds, or a respiratory pause of any duration if accompanied by bradycardia and cyanosis or oxygen desaturation. Apnea of prematurity is caused by neonatal infections, lung disease, hypothermia, hypoglycemia, seizures, maternal drugs, drug withdrawal, anemia (often seen at 4 to 8 weeks of life during the physiologic hemoglobin nadir), and gastroesophageal reflux.

Psychogenic cough (habit cough) has been described as predominantly a daytime cough that disappears after the child falls asleep. The cough is characterized as a dry, repetitive, generally nonproductive "barking seal" cough that is louder whenever attention is paid to it. It does not increase with exercise and does not awaken the sleeping patient.

Hemoptysis is coughing up of blood or the presence of blood in the sputum. Although pulmonary causes may include acute lower respiratory tract infection, cystic fibrosis, foreign body aspiration, and congenital heart disease, the most common cause of hemoptysis is epistaxis, particularly among children who have previously been well.

Vocal cord dysfunction is characterized by an abnormal adduction of the vocal cords during the respiratory cycle (especially inspiration) that produces airflow obstruction at the level of the larynx. It frequently mimics persistent asthma and is often mistreated with high-dosed inhaled and/or systemic corticosteroids, bronchodilators, ER visits, and hospitalizations. Common clinical presentations include wheezing, coughing, a feeling of tightness in the throat, hoarseness and voice change, stridor, shortness of breath, and inspiratory difficulty. Flow-volume loops obtained during symptomatic periods of wheezing show a limitation of inspiratory flow suggesting a variable extrathoracic obstruction. Paradoxical vocal cord motion can be visualized on direct laryngoscopy when patients are symptomatic. The mainstays of treatment for VCD involve teaching the patient vocal cord relaxation techniques and breathing exercises.

Vascular rings are congenital abnormalities of the aortic arch and its major branches resulting in the formation of rings around the trachea and esophagus with varying degrees of compression. Common anomalies include a double aortic arch, right aortic arch with left ligamentum arteriosum, anomalous innominate artery arising further to the left on the arch than usual, anomalous left carotid artery arising further to the right than usual and passing anterior to the trachea, and anomalous left pulmonary artery (vascular sling). If the vascular ring produces compression of the trachea and esophagus, symptoms are frequently present during infancy – chronic wheezing is exacerbated by crying, feeding, and flexion of the neck. Extension of the neck tends to relieve the noisy respiration.

Vomiting is frequent and there may be a brassy cough, pneumonia, or sudden death from aspiration. Imaging tests to help narrow the differential diagnosis include a barium swallow to look for a posterior indentation, echocardiography, MRI, CT, or angiography during cardiac catheterization. Bronchoscopy may be used to determine the extent of airway narrowing.



TEXAS TECH UNIVERSITY HEALTH SCIENCES CENTER
PEDIATRICS

QUESTIONS ON CARDIOVASCULAR AND RESPIRATORY

1. A 9-week-old male infant has had irritability and fever to 104° P (40.0°C) for 8 days. Cultures of blood, urine, and cerebrospinal fluid are negative. A coalescing red maculopapular rash has been present on the trunk and extremities since the second day of the illness. Red scleral conjunctivae without exudate are noted.

Of the following, the MOST likely complication to develop is

- A. aortic thrombosis
B. cerebral infarction
*C. coronary artery aneurysms
D. renal vein thrombosis
E. splenic infarction

KAWASAKI

(B) rev.
LAD
Equiv ESR
1/5 Desquamation
Most common Aneurysm HD

2. A 12-year-old boy has a height that is at the 95th percentile for a 16-year old and a weight that is at the 25th to 50th percentile for his age. He has long fingers, a pectus carinatum, and a midsystolic click followed by a loud honking murmur.

The risk of which of the following is the MOST likely reason to advise against competitive basketball?

- A. commotio cordis with ventricular fibrillation
*B. pericardial tamponade from aortic dissection
C. rhabdomyolysis with severe acidosis
D. sinus of Valsalva rupture with acute aortic regurgitation
E. torsades de pointes with sudden cardiac death

MARFAN

EYES ↑
SKELETAL
HEART - Pericardial Tamp & A/D →
β Blocker

3. During the physical examination of an otherwise healthy 2-month-old infant, you note a harsh grade 3/6 holosystolic murmur that is low-pitched and heard best over the lower left sternal border.

Of the following, the diagnosis MOST consistent with these auscultatory findings is

- A. aortic stenosis RUS
B. atrial septal defect LUS
C. patent ductus arteriosus LUS
D. tetralogy of Fallot
*E. ventricular septal defect

4. A previously healthy 4-month-old boy has been irritable and has had a decreased appetite for 2 days. Findings include: heart rate, 240 beats/min; respiratory rate, 60 breaths/min; rectal temperature 37.4°C (99.3°); diminished radial and posterior tibial pulses; and good brachial and femoral pulses. Electrocardiography shows tachycardia; QRS complexes are narrow without preceding P waves.

Of the following, the most appropriate INITIAL management is to

- A. administer adenosine intravenously
 - B. administer verapamil intravenously
 - *C. apply an ice-filled plastic bag to the entire face for 5 to 10 seconds CAUT BREATH - VAGAL RESPONSE
 - D. apply an ice-filled plastic bag to the upper face for 60 seconds
 - E. perform synchronized direct current cardioversion
5. You are evaluating a 4-year-old healthy girl at her annual health supervision visit. You note clear breath sounds, strong pulses, a quiet precordium, and a murmur. Your partner noted a murmur at last year's visit.

Of the following, the finding MOST consistent with the diagnosis of an innocent murmur is

- ~~A~~ continuous "machinery" murmur under the left clavicle
 - ~~B~~ harsh systolic murmur at the right upper sternal border VIBRATORY - innocent
 - C. high-pitched systolic murmur in the back between the scapulae
 - D. low-pitched, long, diastolic murmur in the left axilla
 - *E. low-pitched, vibratory systolic murmur at the left sternal border
6. A 6-hour-old term infant has increasing pallor, tachypnea, and respiratory distress. Physical examination reveals an enlarged liver, a gallop rhythm, poor pulses in the upper extremities, and absent pulses in the lower extremities.

In addition to treating this infant for potential sepsis, the most appropriate INITIAL intervention while arranging for transfer to a neonatal intensive care unit is to administer

- A. a dopamine infusion
- B. a loading dose of digitalis
- C. a 25% glucose and water solution
- D. furosemide
- ⓔ prostaglandin E₁

7. An otherwise healthy 4-month-old infant is mildly cyanotic but is in no respiratory distress.

The cyanosis in this patient is MOST likely caused by

- A. aortic valve stenosis
- B. atrial septal defect
- C. patent ductus arteriosus
- D. pulmonary valve stenosis
- * E. tetralogy of Fallot

8. Examination of a 3-hour-old infant reveals dysmorphic features and cyanosis. Both the occiput and facial profile are flat, and the fontanelle is abnormally enlarged. The space between the great and second toe is wide, and there is a palmar crease extending across the left palm. Room air oximetry reveals a saturation of 70%.

Of the following, the MOST likely lesion to be found on echocardiography would be

- * A. atrioventricular septal defect
- B. coarctation of the aorta
- C. hypoplastic left heart
- D. Total anomalous pulmonary venous connection
- E. truncus arteriosus

50% of kids w T21 (+ HD)

↑ risk HIRSHSPRUNG

↑ AML, ALL

↑ HYPOTHYROID

↑ ATLANTO AXIAL
HYPOTONIA

9. An asymptomatic term neonate has intermittent 1-second pauses noted when nurses auscultate the heart rate. Results of physical examination are normal except for the irregularity in heart rhythm.

On electrocardiography, the MOST likely finding associated with the intermittent pauses will be

- A. Mobitz type I second-degree AV block
- B. Mobitz type II second-degree AV block
- * C. nonconducted premature atrial beats
- D. premature ventricular beats
- E. sick sinus syndrome

10. An 11-year-old girl has a 1-week history of dyspnea, malaise, and fatigue. She developed vomiting after 24 hours of feeling abdominal fullness and discomfort. Physical examination reveals a blood pressure of 85/50 mmHg, tachypnea, rales, hepatomegaly, and no cardiac murmur. The heart rate by auscultation is 120 beats/min. Palpation of the pulses reveals a regular rate of 60 beat/min.

Of the following, the MOST likely diagnosis is acute

- A. hepatitis
B. lobar pneumonia
 C. myocarditis
D. pancreatitis
E. pericarditis
- Acute - common cause of sudden death in athletes*
11. You are evaluating a 15-year-old boy in your office. He tells you that he experiences chest pain at times while at home and at school. He describes it as sharp, located in the left chest, exacerbated by deep breathing, and resolving spontaneously. He does not feel palpitations or lightheadedness and has not had syncope. His 54-year-old father and grandfather both have had hypertension and myocardial infarctions. At this time, he has no pain, and results of physical examination are normal.

Of the following, the MOST appropriate approach for this patient at this time is

- A. chest radiography
B. echocardiography
C. electrocardiography
 D. reassurance with clinical follow-up
E. referral to pediatric cardiologist
12. You are evaluating a 16-year-old boy for preparticipation sports screening. The boy states that his older brother was diagnosed with a seizure disorder and died suddenly during high school track practice. He also has a younger sister who has a history of syncope.

Before approving him for sports participation, which of the following tests must be performed?

- A. computed tomography of the head
 B. electrocardiography
C. electroencephalography
D. genetic testing for ion channel abnormalities
E. tilt table test

*prolonged QT syndrome →
β-blockers*

13. A 10-year-old girl had recent pharyngitis with culture-proven group A *Streptococcus*. She was noncompliant with antibiotic therapy. She now presents with fever to 102°F (38.9°C), a heart rate of 120 beats/min, and a respiratory rate of 24 breaths/min. She has no murmurs or gallop rhythm. She has a nonpruritic, macular rash that appears as a serpiginous, erythematous circle surrounding normal skin. She also has an erythematous, warm, swollen left knee and right ankle.

Of the following, the MOST appropriate diagnostic study for this girl is

- A. chest radiography
- *B. echocardiography
- C. left knee and right ankle radiography
- D. rheumatoid factor
- E. skin biopsy of the rash

↓ Jones
↓ Joints
↓
Nodules
Erythema marginatum
Symmetrical edema

14. You are evaluating a 16-year-old girl who complains of the sensation of a racing heart and nervousness. She has lost 3.0 kg in 2 months. On physical examination, her heart rate is 110 beats/min, and she has a fine tremor.

Of the following, the MOST likely diagnosis is

- A. anorexia nervosa (BRADYCARDIA)
- B. anxiety disorder
- *C. hyperthyroidism
- D. postural orthostatic tachycardia syndrome
- E. supraventricular tachycardia

15. A 750-g infant who was born at 27 weeks' gestation was weaned successfully from the ventilator on postnatal day 3. Two days later, the infant has bounding pulses, tachypnea, and a new murmur. Echocardiography confirms the diagnosis of patent ductus arteriosus. Hemoglobin is 13g/dL (130 g/L). Electrolytes, creatinine, and platelets are within normal limits.

Of the following, the MOST appropriate initial management strategy for this infant is to

- *A. intravenous indomethacin (CONTRAINDICATION)
- B. oxygen therapy at an Fio₂ of 1.0
- C. surgical ligation of the ductus arteriosus
- D. transcatheter closure of the ductus arteriosus
- E. transfusion with packed red blood cells

16. A 4-day-old infant had an uncomplicated delivery by elective cesarean section. Tachypnea and mild cyanosis are noted on discharge examination. The second heart sound is single, and precordial activity is increased. There is no murmur. Pulse oximetry documents a saturation of 85% in room air.

Of the following, the MOST likely diagnosis is

- * A. hypoplastic left heart syndrome
- B. pulmonary atresia with intact ventricular septum
- C. tetralogy of Fallot
- D. total anomalous pulmonary venous connection
- E. transposition of the great arteries *SEVERE CYANOSIS*

17. A healthy 8-month-old boy has had intermittent episodes of intense cyanosis of the hands, lower arms, and feet since birth. The infant appears alert and playful during the episodes. At other times, his arms and legs are pink but have a "mottled" appearance. Physical examination reveals no remarkable findings other than mottling of the extremities.

Of the following, the BEST diagnostic test for this child is

- A. ambulatory electrocardiography to detect arrhythmia
- B. co-oximetry to detect methemoglobinemia
- C. echocardiography to detect congenital heart disease
- D. electroencephalography to detect a seizure disorder
- * E. no test because these symptoms are benign

18. When examining a healthy 3-year-old boy during a health supervision visit, you hear a grade 2 systolic ejection murmur most clearly at the upper left sternal border.

Of the following, the MOST likely explanation for this finding is

- * A. a large atrial septal defect
- B. a moderate-sized patent ductus arteriosus
- C. a small ventricular septal defect
- ~~D. moderately severe aortic stenosis~~
- ~~E. severe pulmonic stenosis~~

19. A 14-year-old boy complains of nearly constant chest pain for the past 2 days that worsens when he is lying supine.

Of the following, the MOST likely cause is

- A. *Borrelia* myocarditis
- B. enteroviral pericarditis
- C. idiopathic costochondritis
- D. primary pulmonary hypertension
- E. slipped rib syndrome

20. Of the following, the cardiac lesion that MOST requires endocarditis prophylaxis is

- A. ligated patent ductus arteriosus
- B. repaired coarctation of aorta
- C. repaired secundum atrial septal defect
- D. repaired subaortic ventricular septal defect
- E. unrepaired secundum atrial septal defect

21. You are following a 2-year-old girl who was admitted to the hospital with bronchiolitis. The child had cardiac surgery to repair transposition of the great arteries 6 months ago and recovered uneventfully. Her neurologic examination results are normal. A resident physician asks you about the child's predicted cognitive abilities and school performance.

Of the following, your BEST response is that

- A. electroencephalography is the best predictor of future cognitive function
- B. school performance will depend on the amount of early intervention services she receives
- C. she probably will have difficulty with attention and learning
- D. she will have normal cognitive abilities
- E. there is no information available to predict her cognitive function

22. You are evaluating a 3-day-old infant brought to the emergency department for lethargy. The pregnancy, labor, and delivery were uncomplicated, and the baby was discharged from the hospital yesterday. On physical examination, the heart rate is 180 beats/min, the respiratory rate is 80 breaths/min, and the blood pressure is 50/30 mmHg. The infant is pale and mottled and has cool extremities and weak distal pulses.

Of the following, the MOST likely cardiac diagnosis is

- A. atrioventricular septal defect
- B. critical aortic stenosis
- C. large ventricular septal defect
- D. tetralogy of Fallot
- E. transposition of the great arteries

23. You are called to the newborn nursery to evaluate a 3-hour-old term male who was born after a normal pregnancy and delivery and whose birthweight was 4 kg. He appears comfortable, has mild tachypnea and hyperpnea to 65 breaths/min, and has a heart rate of 160 beats/min. Cyanosis is apparent in his hands, feet, and perioral area. His precordium is quiet, and you hear no murmurs or gallop. His pulses and perfusion are excellent. An oxygen saturation measurement in room air is 68%. When you deliver high-flow oxygen by mask, his oxygen saturation rises to 72%.

Of the following, the MOST likely cause of cyanosis in this newborn is

- A. tetralogy of Fallot
 - B. total anomalous pulmonary venous return
 - C. transposition of the great arteries
 - D. tricuspid atresia
 - E. truncus arteriosus
24. You are evaluating a 16-year-old girl who reports that she has fainted four times in the last 3 months. Two of these episodes occurred while she was standing (once at church and once in line at the school cafeteria). One episode occurred while she was getting out of the shower and one after basketball practice. She does not complain of any exercise fatigue, palpitations, or chest pain.

Of the following, the BEST initial step in her evaluation is

- A. echocardiography
 - B. electroencephalography
 - C. electrocardiography
 - D. head computed tomography
 - E. Holter monitoring
25. A 3-year-old boy presents to the emergency department with a 1-month history of fever, cough, and weight loss. His parents are Cambodian, although he was born in the United States. In talking with his parents through an interpreter, you learn that relatives from Cambodia recently visited the family.

Of the following, the diagnostic test MOST likely to yield the cause of this child's illness is

- A. blood culture
- B. *Mycoplasma* serology on serum
- C. nasopharyngeal swab for viral culture
- D. skin test with purified protein derivative
- E. sweat chloride test

26. You are evaluating a 1-month-old infant who has inspiratory stridor. He is otherwise healthy, but was noted to have stridor at birth that has worsened slightly.

Of the following, the MOST likely cause of the stridor in this infant is

- A. bilateral paralysis of the vocal cords (2)
- B. hypertrophy of the adenoids
- *C. laryngomalacia (1)
- (D) subglottic stenosis (3)
- E. tracheomalacia

27. A 3-month-old boy who has been previously healthy is brought to the emergency department with a 3-day history of rhinorrhea, mild cough, and wheezing. He has been afebrile and has had some difficulty feeding. His pulse oximetry reading is 90% on room air, respiratory rate is 60 breaths/min, and heart rate is 130 beats/min. Chest examination reveals mild subcostal retractions, scattered wheezes, and coarse crackles bilaterally. The rest of the physical examination findings are normal.

Of the following, the pathogen that is MOST likely responsible for his symptoms is

- A. adenovirus
- B. *Chlamydia trachomatis* (WHEEZING) (FEVER) EYE 5-14 DAYS LUNG 1-3 MONTHS
- C. *Haemophilus influenzae*
- *D. Respiratory syncytial virus (APNEA) (F)
- E. *Streptococcus pneumoniae*

28. A term infant is delivered vaginally following an uncomplicated pregnancy and experiences immediate respiratory distress. He has cyanosis, tachypnea, and poor perfusion. The abdomen is scaphoid. Resuscitation includes endotracheal intubation and assisted ventilation. In the neonatal intensive care unit, the nurse reports hearing the heart sounds best to the right of the sternum.

Of the following, the MOST likely anticipated findings on chest radiography are

- *A. bowel in the left hemithorax and atelectatic and small right lung
- B. normal lung volumes but a paucity of pulmonary vascular markings
- C. overinflation with bilateral patchy infiltrates
- D. overinflation with perihilar streaking and fluid in the right minor fissure
- E. underinflation, ground glass densities, and air bronchograms

29. An 18-month-old girl has been having an intermittent nonproductive cough for the past 6 months. Her parents state that the cough wakes the toddler at night a few times a month and occurs when playing vigorously. During a recent upper respiratory tract illness, her cough worsened and occurred daily for 3 weeks. On physical examination, there is no nasal discharge, and the toddler appears healthy.

Of the following, the MOST likely diagnosis is

- A. allergic rhinitis
 - B. asthma
 - C. atypical pneumonia
 - D. gastroesophageal reflux
 - E. sinusitis
30. During the health supervision visit of a 2-year-old boy who is new to your practice, you note a productive cough that his mother says has been present for several months. His weight is below the 5th percentile. Fine crackles are present in his lungs bilaterally, and he has mild clubbing of the fingers. The remainder of his physical examination findings are unremarkable. A chest radiograph reveals poorly defined bronchovascular markings, prompting you to order computed tomograph scan of the chest, which reveals diffuse bronchiectasis.

Of the following, the MOST likely diagnosis is

- A. allergic bronchopulmonary aspergillosis
 - B. cystic fibrosis
 - C. foreign body aspiration
 - D. *Mycobacterium tuberculosis* infection
 - E. primary ciliary dyskinesia
31. A 15-month-old boy has had five episodes of acute otitis media, chronic rhinosinusitis, and two episodes of pneumonia. Chest radiography reveals dextrocardia.

Of the following, the study that is MOST likely to provide a definitive diagnosis is

- A. human immunodeficiency virus testing
- B. mucosal biopsy for ciliary analysis
- C. pH probe to document gastroesophageal reflux
- D. skin testing to document inhalant allergies
- E. sweat chloride test for cystic fibrosis

32. A 3-year-old girl is brought to the emergency department because of stridor that developed over 24 hours and is associated with fever (102.9°F [39.4°C]) and dysphagia. Physical examination reveals a child who is sitting up, drooling, and exhibiting inspiratory stridor. Her immunization status is unknown.

Of the following, the MOST likely finding to support the correct diagnosis is

- A. airway fluoroscopy showing midtracheal expiratory collapse
 - B. anteroposterior neck radiography showing subglottic narrowing
 - C. barium swallow showing a midthoracic indentation
 - D. chest radiography showing irregular tracheal debris
 - *E. lateral neck radiography showing supraglottic edema
33. An 18-month-old girl is brought to your clinic with a 3-day history of difficulty breathing. Physical examination reveals a well-nourished, alert girl who has wheezing localized to the right lower lobe. A review of her chart shows that she had similar complaints and physical examination findings twice last month, which your partner treated with albuterol. The remainder of her past medical history is unremarkable. You obtain a chest radiograph, which appears normal except for right lower lobe atelectasis.

Of the following, the MOST likely diagnosis is

- A. bronchogenic cyst
 - B. cystic fibrosis
 - C. foreign body aspiration
 - D. gastroesophageal reflux
 - E. reactive airway disease
34. An 8-week-old child is being seen for a health supervision visit. His mother reports that he is wheezing and having increasing difficulty while feeding. Physical examination reveals a raised, erythematous birthmark on the chest and biphasic stridor on auscultation.

Of the following, the MOST likely finding would be

- A. a prolapsing epiglottis on nasopharyngoscopy
- *B. a unilateral subglottic mass on fluoroscopy
- C. left mainstem bronchomalacia on bronchoscopy
- D. obstructing adenoids on lateral neck radiography
- E. unilateral air trapping on chest radiography

35. A 16-year-old boy is struck by an automobile while crossing the street and is thrown 15 ft. Upon arrival at the emergency department, he is in severe respiratory distress and complains of chest pain. Findings on physical examination include an ecchymotic area over the right side of the chest and subcutaneous emphysema. Breath sounds are absent on the right side, the trachea is deviated to the left, and the right hemithorax is tympanitic to percussion.

The BEST initial management of this patient is

- A. chest tube thoracostomy
 - B. computed tomography of the abdomen
 - C. computed tomography of the chest
 - D. pericardiocentesis
 - E. plain radiography of the chest
36. A 3-year-old boy has experienced hoarseness for more than 12 months. He has no dyspnea or stridor. Shortly after birth he had ligation of a patent ductus arteriosus.

Of the following, the procedure that will provide the MOST helpful information about this patient is

- A. auscultation of the chest for stridor
 - B. barium esophagography for assessment of a vascular ring
 - C. inspection of the palate for symmetric motion
 - D. flexible airway endoscopy for assessment of vocal cord mobility
 - E. posteroanterior and lateral chest radiography for evaluation of the pulmonary hilum
37. A 5-year-old boy who has had a cough and upper respiratory tract infection for the past week presents with a rapid onset of expiratory stridor and dyspnea with retractions. His temperature is 104°F (40°C). Chest radiography reveals an irregular-appearing tracheal air column.

Of the following, the MOST appropriate treatment is

- A. antimicrobial coverage with ceftriaxone
- B. bronchoscopy to remove purulent debris and antibiotics to cover *Staphylococcus aureus*
- C. bronchoscopy to remove purulent matter and antibiotics to cover *Haemophilus influenzae* type b
- D. emergency tracheotomy
- E. esophagoscopy for biopsy and antibiotics to cover *Haemophilus influenzae* type b

BACTERIAL TRACHEITIS
LAZ CANN - NO DRUGS

38. A 14-year-old girl who has cystic fibrosis has exhibited dyspnea at rest for several weeks. An arterial blood gas analysis is performed while she is receiving 5 L/min of supplemental oxygen.

Among the following, the results of the arterial blood gas analysis are MOST likely to show

	<u>pH</u>	<u>Pco₂</u>	<u>PO₂</u>
A.	7.20	20	120
B.	7.20	60	80
C.	7.35	25	80
<input checked="" type="checkbox"/> D.	7.35	60	80
E.	7.50	60	80

39. A 12-year-old girl has been treated with intravenous clindamycin and cefotaxime for the past 72 hours for left lower lobe pneumonia. She continues to have daily temperatures of 103°F (39.4°C).

Of the following, the MOST likely reason for her continued fever is

- A. drug fever
 - B. inappropriate antimicrobial therapy
 - C. insufficient duration of therapy
 - D. the development of an empyema
 - E. viral pneumonia
40. A 12-year-old girl develops low-grade fever and a nonproductive cough following several days of sore throat and malaise. She appears tired, and crackles are audible over the lower right lung field on chest auscultation. Chest radiography reveals patchy alveolar infiltrates in the right lower and middle lobes.

Of the following, the diagnostic test MOST likely to identify the etiology of this illness is

- A. blood culture
- B. serology for *Mycoplasma*
- C. sputum culture
- D. urine antigen detection studies
- E. viral culture of respiratory secretions

41. A 28-day-old infant who weighs 1,100 g has a sudden onset of apnea that resolves with tactile stimulation. In the preceding 2 weeks, the infant had been stable breathing room air, tolerating orogastric feeding, and gaining weight. Physical examination reveals a resting heart rate of 180 beats/min.

Of the following, the MOST likely cause of apnea in this infant is

- A. anemia of prematurity
- B. hypoglycemia
- C. intraventricular hemorrhage
- D. sepsis
- E. symptomatic patent ductus arteriosus

42. A 9-year-old previously healthy child has had a persistent harsh cough for the past 3 months. She has been treated with inhaled albuterol, beclomethasone, and a 7-day course of oral prednisone without any change. The cough is present during the day, but not at night. No one else at home has been ill or coughing.

Among the following, the MOST likely cause of this patient's cough is

- A. asthma
- B. foreign body aspiration
- C. immotile cilia syndrome
- D. psychogenic cough
- E. tuberculosis

43. You are evaluating a 1-month-old boy who has had inspiratory stridor since birth. The stridor is associated with retractions when the infant becomes agitated. Physical examination reveals a weight of 3.4 kg (10th percentile), no expiratory stridor, and a weak cry.

Of the following, the MOST likely cause of the stridor in this infant is

- A. bilateral paralysis of the vocal cords
- B. laryngeal cleft
- C. laryngomalacia
- D. subglottic cyst
- E. tracheomalacia

44. A previously well 5-year-old boy is brought in by his mother because he coughed up blood. He has had some nasal congestion, but the remainder of his medical history is unremarkable. You confirm that the bright red material on the tissue is blood.

Of the following, the MOST likely explanation for this child's coughing up blood is

- A. aspiration of a foreign body
 - B. cystic fibrosis
 - *C. epistaxis
 - D. gastric ulcer
 - E. pulmonary arteriovenous malformation
45. A 15-year-old girl comes to your office complaining of chest and throat tightness when she plays sports and when she is stressed. She has a history of asthma and reports that her medications no longer help her. She is presently receiving high-dose inhaled steroids, a long-acting beta agonist, and an antihistamine and has just completed a 5-day course of prednisone. None of these medications has decreased the incidence or severity of the attacks. Physical examination reveals no wheezing, and her peak flow rate is in the normal range for her age and height.

Of the following, the MOST likely diagnosis is

- A. allergic rhinitis
 - B. asthma
 - C. pneumonia
 - D. sinusitis
 - *E. vocal cord dysfunction
46. A 5-year-old boy presents to the emergency department with an acute exacerbation of asthma. He has had an upper respiratory tract infection for 2 days and a 3-year history of asthma. Initial findings include a respiratory rate of 40 breaths/min, heart rate of 120 beats/min, pulse oximetry reading of 95%, temperature of 99°F (37.2°C), and diffuse wheezing in all lung fields. He is given multiple nebulizer treatments with a beta agonist and an anticholinergic agent as well as oral corticosteroids. On repeat evaluation, his respiratory rate is 27 breaths/min, heart rate is 97 beats/min, and pulse oximetry reading is 92%. He feels much better and looks improved.

Of the following, the MOST likely reason for the decrease in his pulse oximetry reading is

- A. a damaged pulse oximetry probe
- B. a foreign body moved within the airway
- C. bacterial pneumonia
- D. previously unrecognized cardiac disease
- *E. ventilation and perfusion mismatching

47. You are evaluating a 3-week-old infant whose mother states that he coughs when he feeds. You observe a feeding and note stridor, coughing, and wheezing during and immediately after feeding. Physical examination reveals no cardiac murmurs, strong distal pulses, and normal perfusion. He has no stridor at rest.

Of the following, the MOST helpful diagnostic study for this patient is

- A. barium swallow
- B. electrocardiography
- C. laryngoscopy
- D. radiography of the neck
- E. sweat test

VASCULAR RING - Post. INDENTATION ON BARIUM SWALLOW

FACTS – RENAL

Nephrotic syndrome is characterized by significant proteinuria, hypoalbuminemia, edema, and hyperlipidemia. The most common type is minimal change disease, seen in children between the ages of 2 to 8 years and more common in boys than girls (2:1). The initial presentation of the disease (absence of hypertension, hematuria, or hypocomplementemia) and the responsiveness to treatment with prednisone generally are good prognostic signs. Parents are advised to follow a sodium-restricted diet and to restrict fluid intake in the presence of severe oliguria. If severe, the edema can be treated with a combination of albumin and diuretics. The two major complications of nephrotic syndrome include infection (spontaneous bacterial peritonitis with *Streptococcus pneumoniae*) and thromboembolic phenomena (renal vein thrombosis, pulmonary embolism, DVT). FSG (focal segmental glomerulosclerosis) is the cause of 10% of cases of nephrotic syndrome in children. Suspicion for FSG should increase with older age at onset, RBCs in the urine, and lack of response to systemic corticosteroids, with most patients progressing to end-stage renal disease.

Orthostatic proteinuria is an asymptomatic condition, common in adolescence, where children excrete normal or slightly increased amounts of protein in the supine position, but excrete up to a tenfold increase in protein in the upright position. There is no associated hematuria or edema, renal function is normal, findings on biochemical and immunologic evaluations are unremarkable, and results of renal biopsy are normal. The diagnosis is confirmed by testing upright and supine urine collections. Most cases of orthostatic proteinuria will have a negative urine dipstick for protein in the early morning specimen.

Acute post-streptococcal glomerulonephritis is a classic example of the acute nephritic syndrome: the sudden onset of gross hematuria, edema, hypertension, and renal insufficiency. It was the most common cause of gross hematuria in children (IgA nephropathy is now the most common cause). PSGN follows infection of the throat or skin with certain “nephritogenic” strains of group A B-hemolytic strep by 1 to 3 weeks. The most common age of presentation is 7 years with a 2:1 male to female ratio. 70% have hypertension, 50% have transient acute renal failure, and 10 to 20% present with nephrotic syndrome. A urinalysis demonstrates RBCs, frequently in association with RBC casts and proteinuria, and urine is described as cola-colored, smoky, or tea-colored. Blood work reveals an elevated ASO titer in 70% and decreased C3. Anti-hypertensive medicines are indicated to treat the hypertension and complete recovery occurs in more than 95% of children with acute PSGN, but microscopic hematuria may persist for up to 2 years.

Autosomal dominant polycystic kidney disease is usually an adult-onset disorder that has an incidence of 1:1,000 live births. Typical presenting symptoms are hematuria, hypertension, and abdominal pain. Most patients progress to end-stage renal disease by mid-adulthood. The cysts are always bilateral, scattered throughout both enlarged kidneys, and are often discovered in asymptomatic patients whose family members are affected by the disease. Associated abnormalities may include pancreatic and splenic and hepatic cysts, which are commonly asymptomatic, as well as rare occurrences of congenital hepatic fibrosis and heart valve defects. 10% of affected patients experience berry aneurysms of the cerebral circulation that may result in intracranial hemorrhage and death.

Autosomal recessive polycystic kidney disease is often detected in utero by the presence of enlarged kidneys and oligohydramnios. Most affected infants will present with bilateral flank masses. Although some patients who have ARPKD die in infancy from the renal disease (Potter facies – low-set ears/flattened facies/micrognathia, pulmonary hypoplasia), most survive with mild renal insufficiency or hypertension. The majority of patients also have cysts in the liver, and in severe cases may be associated with liver cirrhosis, portal hypertension, and death due to ruptured esophageal varices. When the severity of hepatic manifestations exceeds that of renal involvement, the disorder is called congenital hepatic fibrosis.

IgA nephropathy (Berger disease) is a glomerular disease that occurs most often during the 2nd decade of life and in young adults. It occurs more commonly in males (2-6:1) and is associated with elevated IgA levels and normal complement. The two most common presenting symptoms are episodic, painless gross hematuria (brown rather than red) 1 to 2 days after a URI and persistent microscopic hematuria. Prognosis is variable – 1/3 have mild disease with minimal evidence of progression, 1/3 experience frequent recurrences of gross hematuria with a slow decline in renal function over several decades, and 1/3 have significant proteinuria with hypertension and renal insufficiency that requires renal replacement therapy and transplantation.

Alport syndrome, also termed “familial nephritis”, is a chronic, progressive renal disease that invariably results in end-stage renal failure by the second decade of life. The inheritance is X-linked dominant and most affected patients are males. Most females in families are asymptomatic carriers. Most patients have a positive family history and hematuria even during infancy. Patients often suffer from deafness (high-frequency sensorineural) and ocular abnormalities (anterior lenticonus).

RTA (renal tubular acidosis) is a clinical entity of hyperchloremic normal anion gap metabolic acidosis that is often uncovered during evaluation for growth failure. There are three types:

1. Distal RTA (type 1) – impaired acidification of the urine with inability to secrete hydrogen ions distally, with alkaline pH.
2. Proximal RTA (type 2) – increased urinary bicarbonate wasting from the proximal tubule. Because the distal hydrogen-secreting mechanisms are still intact, these patients can acidify their urine if they are extremely acidotic.
3. Mineralocorticoid deficiency or resistance (type 4) – results from diseases of the adrenal gland which lead to decreased aldosterone or from reflux nephropathy which impairs the function of aldosterone. This form is associated with hyperkalemia.

Abdominal masses in neonates and children are most commonly of genitourinary or retroperitoneal origin: more than 50% of abdominal masses originate in the kidneys, including UPJ obstruction and multicystic dysplastic kidney. In addition to presenting as a mass or as hydronephrosis on prenatal screening, UPJ obstruction may present as a febrile urinary tract infection or macroscopic hematuria following minor trauma, with VUR present in nearly 15% of cases. MCDK is the most common cause of an abdominal mass in the newborn and is found in 1 in 4,000 live births, mostly detected on prenatal ultrasound or with a unilateral flank mass in the neonatal period. The diagnosis should be confirmed with a renal scan, which should demonstrate non-function and a VCUG is mandatory because 15% of patients will have contralateral VUR. Most of the time, MCDK disease can be observed without surgical intervention because the dysplastic kidney involutes over time.

Red urine can be seen in a number of situations, including hematuria, myoglobinuria, hemoglobinuria, or ingestion of food or medications (beets, blackberries, AZO dyes, Chloroquine, Rifampin, and Nitrofurantoin). Urate crystals can cause a red-orange discoloration of the urine and are responsible for the “red-diaper syndrome” that may occur in infants in the first six months of life due to high excretion of uric acid. The strip test for hematuria uses the peroxidase-like activity of hemoglobin/myoglobin to result in a color change. It is important to perform a microscopic exam of the urine after screening with the dipstick to distinguish among hematuria, hemoglobinuria, and myoglobinuria. In the absence of red blood cells, which rules out trauma, myoglobinuria is the most likely diagnosis following a vigorous exercise like football practice or track. This can be confirmed by a qualitative test for myoglobin and measurement of the serum CPK. Treatment is IV hydration and administration of sodium bicarbonate to alkalinize the urine and prevent the precipitation of myoglobin in the tubules.

Idiopathic hypercalcuria (normocalcemic hypercalcuria) occurs in approximately 3 to 6% of all children. The clinical manifestations include calculi, renal colic, hematuria (gross or microscopic), dysuria, frequency, urgency, pyuria, and enuresis. The first sign of hypercalcuria is usually microscopic hematuria with red blood cells in the urine. The initial screening step is a spot urine for calcium and creatinine with a ratio greater than 0.2 suggestive of hypercalcuria. Treatment involves increasing fluid intake sufficient to maintain a high urine volume and possibly thiazide diuretics and alkali in the form of citrate. Furosemide (Lasix) therapy has been associated with the development of hypercalcuria among infants who are receiving the drug for treatment of BPD.

Henoch-Schönlein purpura is a systemic IgA-mediated vasculitis that occurs in males twice as frequently as females between 2 to 8 years of age. A viral syndrome or upper respiratory infection often precedes HSP, with 20% of patients having a recent group A B-hemolytic strep infection. The four distinctive clinical features include: skin manifestations (urticarial erythematous rash that progresses to petechiae and palpable purpuric lesions concentrated on the buttocks and lower extremities), joint manifestations (80% with arthralgia or arthritis, often the knees and ankles), GI manifestations (67% of patients with colicky abdominal pain that may be severe, GI bleeding, intussusception), and renal manifestations (varies from mild hematuria and trace proteinuria to gross hematuria, nephrotic syndrome, and end-stage renal disease). Treatment is based on relief of symptoms, including treatment with NSAIDs and hydration. Steroids may be effective for relief of severe abdominal pain and arthritis. Although HSP can recur in up to 50% of patients, most patients recover within four weeks, and long-term morbidity is dependent only on the severity of nephritis, with less than 1% of cases progressing to end-stage renal disease.

Posterior urethral valves are the most common cause of urethral obstruction in males with incidence ranging from 1:5,000 to 1:8,000. These valves appear as two thin leaflets that run between the floor and roof of the urethra and block the urinary stream during voiding. Characteristic prenatal findings include bilateral hydronephrosis, megaureters, a trabeculated bladder, and oligohydramnios, with severely affected fetuses exhibiting pulmonary hypoplasia and features of Potter sequence. Mothers will frequently describe a weak stream or “dribbling” and a contrast VCUG reveals dilation of the prostatic urethra and a transverse linear filling defect. Despite aggressive medical and surgical intervention, up to 1/3 of infants with posterior

urethral valves will develop end-stage renal failure that requires dialysis and transplantation. Sickle-cell trait is present in about 8% of African-Americans with 35–45 % of their hemoglobin as Hgb S. These patients may present with painless, gross hematuria, with the source of bleeding 5 times more likely to be the left kidney than the right kidney. The pathogenesis of the hematuria is unknown, although it probably results from sickling and vascular obstruction in the medulla. The initial evaluation of a patient who has hematuria associated with sickle-cell trait should include Doppler ultrasonography of the kidneys and bladder to assess anatomy and flow to the kidney.

Diabetes insipidus can result from the absence of anti-diuretic hormone (central DI) or end-organ resistance to the action of the hormone (nephrogenic DI). Tumors of the suprasellar and chiasmatic regions (craniopharyngioma, optic glioma) are common causes. Other causes include Langerhans cell histiocytosis, encephalitis, sarcoidosis, TB, surgical procedures and head trauma. In addition to the polydipsia and polyuria, patients with DI may present with frequent episodes of hypernatremic dehydration during episodes of poor oral intake or gastroenteritis. These patients present with a very elevated serum sodium and a very dilute urine osmolality approaching 100 mOsm/kg H₂O despite intravascular fluid depletion.

Prune belly syndrome (Eagle-Barrett syndrome) most often occurs in males with a characteristic triad of deficient abdominal musculature with wrinkled appearance to the skin, undescended testes, and urinary tract abnormalities probably resulting from severe urethral obstruction in fetal life (often posterior urethral valves). Oligohydramnios and pulmonary hypoplasia are frequent complications in the perinatal period.

Bladder outlet obstruction generally presents with any or all of the following symptoms: reduction in urine output, bladder spasms, incontinence, lower urinary tract infection, abdominal or suprapubic pain, or feeling of abdominal fullness. The etiology of bladder outlet obstruction includes mechanical causes (tumor, posterior urethral valves), neurogenic bladder (due to spinal deformity), or medications (anticholinergics).

The findings of chronic renal failure and rickets are consistent with the diagnosis of renal osteodystrophy. Phosphate retention, which occurs because of deteriorating renal function, is critical to the pathogenesis of renal osteodystrophy. This leads to an increase in the solubility product of calcium and phosphorus with a decrease in serum ionized calcium concentration. In addition, hyperphosphatemia inhibits the activity of a renal tubular enzyme, 1-alpha-hydroxylase, resulting in decreased 1, 25-dihydroxyvitamin D. Calcium malabsorption ensues, with further hypocalcemia, secondary hyperparathyroidism, and increased bone resorption associated with increased levels of alkaline phosphatase.

Hypertension is defined as the persistence of elevated blood pressure above the 95th percentile for age on at least three separate occasions. The blood pressure cuff should completely encircle the upper arm, and the inflatable bladder should cover at least 2/3 of the length of the upper arm. The prevalence of hypertension in children is 1 to 2.5%. In infants and children, hypertension usually is indicative of underlying disease (secondary hypertension). In the newborn, hypertension most often is associated with umbilical artery catheterization and renal artery thrombosis. In early childhood, it is often associated with renal disease (acute post-infectious glomerulonephritis, HUS), coarctation of the aorta, or endocrine disorders. Older children may have hypertension due to renal scarring following recurrent acute pyelonephritis. Renal artery

stenosis may be caused by fibromuscular dysplasia with 5 to 25% of these children having neurofibromatosis. The incidence of essential hypertension increases among adolescents; however, hypertension due to specific medications (oral contraceptives, OTC cold remedies) and drug abuse (cocaine, amphetamines, and diet pills) must be considered in this age group.

The differential diagnosis of acute scrotal pain in an adolescent male includes epididymitis, orchitis, testicular torsion, incarcerated hernia, and testicular trauma. Epididymitis is infection and inflammation of the epididymis, occurring most commonly in sexually active males secondary to *Neisseriae gonorrhoea* or *Chlamydia trachomatis* – common presentations include urinary frequency, dysuria, urethral discharge, and posterolateral tenderness and swelling of the testes, with decreased pain upon elevation of the scrotum (positive Prehn sign). Orchitis presents very similar to epididymitis with diffuse testicular pain, swelling, and erythema of the adjacent skin – most commonly caused by the mumps virus and may see coexisting parotid swelling. Testicular torsion is the most common and most serious cause of acute painful scrotal swelling – presents with sudden onset of scrotal/inguinal/suprapubic pain with accompanying nausea and vomiting, a swollen tender testicle with absent cremasteric reflex on the affected side – often diagnosed by history and physical alone, although torsion may be confirmed by absent Doppler flow on ultrasound of the scrotum – management includes surgical detorsion which is a urologic emergency that must be performed within 6 hours to reliably preserve testicular function. A varicocele typically presents as a painless left scrotal mass that is located superior to the testes, has the consistency of a “bag of worms,” and disappears when the patient is in the supine position and enlarges with standing and with Valsalva maneuver – it results from a dilation and tortuosity of veins in the pampiniform plexus – management typically includes reassurance only. Testicular cancer is the most common solid tumor in males ages 15 to 35 years – typically presents with a solid, firm, painless testicular mass or generalized testicular swelling – most of these are germ cell tumors with the most common type being a yolk-sac tumor with an elevated serum alpha-fetoprotein – 1/3 of testicular tumors in children are benign – treatment often involves radical orchiectomy and possibly retroperitoneal lymph node dissection – the single most important risk factor for the development of testicular cancer is a prior history of cryptorchid testes.



TEXAS TECH UNIVERSITY HEALTH SCIENCES CENTER
PEDIATRICS

QUESTIONS ON GASTROINTESTINAL AND RENAL

1. A 2-week-old term male infant develops progressive bilious vomiting and poor feeding. You recall that the differential diagnosis of vomiting is age-related.

Of the following, the condition that is MOST likely to cause bilious vomiting in this infant is:

- A. pyloric stenosis 2 weeks
B. gastroesophageal reflux
*C. duodenal atresia 1st coo-1 hr after Always Day 1
D. intussusception 2-24 mo 2 non - 3 yr
E. midgut volvulus < 1 month

2. A mother calls your office because her 2-week-old term infant is passing bloody stools. You instruct her to bring the child to the office immediately for evaluation. The infant has been breastfeeding vigorously. Upon physical examination, the child appears well, but when she passes a stool, it contains both blood and mucus.

Of the following, the MOST likely explanation for the bloody stools is:

- A. Crohn disease
*B. milk protein allergy
C. Meckel diverticulum
*D. necrotizing enterocolitis
E. rectal fissure

3. A 7-year-old boy has had diarrhea for the past 18 months, often unknowingly soiling his underwear. During meals, he complains of abdominal pain and his abdomen becomes distended. He has had no weight loss, fever, or other illnesses. Physical examination reveals abdominal distension with normal bowel sounds. Rectal examination is deferred because of the large amount of stool in the child's underwear.

The MOST likely cause of this child's symptoms is:

- *A. fecal overflow incontinence
B. giardiasis
C. Hirschsprung disease
D. irritable bowel syndrome
E. sexual abuse

4. An 18-month-old male infant presents to the emergency department with a 2-hour history of colicky abdominal pain and vomiting. His mother reports that he is difficult to arouse between these episodes. Physical examination is notable for the absence of palpable bowel in the right lower quadrant and guaiac positive stool.

Of the following, the MOST appropriate diagnostic test is:

- A. abdominal ultrasonography
- B. upper GI series
- C. flat plate radiography of the abdomen
- D. hydrostatic barium enema
- E. pertechnetate radionuclide scan

INTUSSUSCEPTION -
COLICUM
BILIOUS EMESIS

5. A previously healthy 6-year-old girl has episodes of abdominal pain that are periumbilical and sharp in character over the past 4 months. The pain does not interfere with sleep or play. Growth and development are normal. She had had no fever, mouth ulcers, joint complaints or rectal bleeding, but has had diarrhea intermittently. Findings on physical examination are normal. The MOST likely diagnosis is:

- A. acute cholecystitis
- B. chronic appendicitis
- C. Crohn disease
- D. functional abdominal pain
- E. intussusception

6. A 15-year-old previously healthy boy complains of decreased appetite, but he always feels full. He denies taking any medications or drugs. He has had low-grade fevers intermittently over the past year. Weight and height have dropped from the 25th percentile to below the 5th percentile over the past 2 years. Occult blood is present in the stool.

The MOST appropriate diagnostic test to obtain is a(n):

- A. barium enema to visualize the appendix
- B. computed tomographic scan of the abdomen to look for abscess formation
- C. stool smear for white blood cells
- D. ultrasonographic scan of the abdomen
- E. upper gastrointestinal series to visualize the terminal ileum

CROHN'S

7. A 6-week-old boy presents with weight loss and dehydration. He has been vomiting after each feeding for the past 2 weeks. The emesis is forceful and contains only formula. The infant always appears to be hungry. Physical examination reveals an emaciated infant who has dry mucous membranes. A discrete abdominal mass is palpable in the right upper quadrant.

Measurement of serum electrolytes in this infant is MOST likely to reveal:

- A. hypochloremic metabolic acidosis
- B. hypochloremic metabolic alkalosis
- C. normal electrolyte levels
- D. respiratory acidosis with metabolic compensation
- E. respiratory alkalosis

8. A 2-year-old boy presents with fever, abdominal pain and bloody diarrhea. Several days later, he develops pallor and poor urine output. Of the following, the MOST likely explanation for these findings is infection with:

- A. enterohemorrhagic Escherichia coli
- B. enteroinvasive E coli
- C. enterotoxigenic E coli
- D. Salmonella enteritidis
- E. Shigella dysenteriae

HUS

9. A 15-year-old boy has developed icterus during an episode of gastroenteritis. He has had three previous episodes of icterus associated with vomiting and dehydration. He denies pruritus, abdominal pain, fatigue, fever or bleeding episodes. Results of the physical examination and a complete blood count, red blood cell morphology and erythrocyte sedimentation rate are normal.

Of the following, the MOST appropriate management is:

- A. a hematology consultation to exclude hemolysis
- B. a liver biopsy to exclude chronic hepatitis
- C. abdominal ultrasonography to exclude gallstones
- D. hepatitis serologies to exclude infectious viral hepatitis
- E. reassurance that findings are caused by a benign genetic disorder

10. A 14-year-old adolescent is brought to the emergency department for evaluation of hematochezia. Physical examination reveals pallor, tachycardia, and hypotension, but no hepatosplenomegaly or abdominal tenderness.

After obtaining laboratory tests, the MOST appropriate next step in management is:

- A. abdominal ultrasonography
- B. emergent colonoscopy
- C. Meckel scan
- D. placement of a nasogastric tube
- E. upper gastrointestinal radiographic series

11. A 10-year-old boy comes to the emergency department because of vomiting, cramping abdominal pain, abdominal distension and bloody stools. He had three previous episodes of painless rectal bleeding between the ages of 2 and 5 years. You suspect an intussusception and order a barium enema. The study successfully reduces the intussusception and documents no other abnormality.

Of the following, the MOST likely cause of intussusception in this child is:

- A. Meckel diverticulum
- B. mesenteric adenitis
- C. nodular hyperplasia of the small intestine
- D. shigellosis
- E. small bowel lymphoma

12. A 3-year-old child is brought to the emergency department with a fever of 103.1°F (39.5°C) and diarrhea of acute onset. The stool is guaiac-positive and contains leukocytes. There is no history of foreign travel, and the child has not received antibiotics recently.

Of the following, the organism that is MOST likely to be isolated from this child's stool is:

- A. clostridium difficile
- B. Giardia lamblia
- C. rotavirus
- D. salmonella enteritidis
- E. vibrio cholerae

13. A healthy 15-month-old male infant is brought to your office for evaluation of diarrhea. His growth, weight gain, activity, and appetite have been normal. The first stool of the day appears normal, but they become increasingly loose and frequent as the day progresses.

Of the following, your BEST advice to the mother is to

- A. avoid fiber-containing foods
- B. initiate the intake of balanced electrolyte solutions
- C. limit the intake of fluids and fruit juice
- D. restrict the intake of fatty foods
- E. substitute lactose-free for other dairy products

14. You are evaluating a previously healthy 10-year-old child who has fever and persistent vomiting. Initially the emesis was clear, then it became more yellow, and now it contains bright red blood. His brother had acute gastroenteritis 1 week ago. Findings on physical examination, complete blood count, and serum chemistries are normal.

The MOST likely cause of the hematemesis is

- A. esophageal varices
- B. esophagitis
- C. gastric duplication
- D. Mallory-Weiss tear
- E. peptic ulcer disease

15. A 2-year-old boy presents with a 1-month history of intermittent watery diarrhea, abdominal distension, and anorexia. He has had no fever. His medical history is unremarkable, and he is taking no medications. He attends a local child care center.

Of the following, the MOST appropriate test or course of action for diagnosis in this child is

- A. enzyme-linked immunosorbent assay of stool for *Giardia*
- B. measurement of serum immunoglobulin G anti-*Giardia* antibody
- C. small bowel biopsy
- D. string test for *Giardia*
- E. therapeutic trial of metronidazole

16. A 3-year-old boy who has long-standing microcytic hypochromic anemia is brought to the emergency department because of hematemesis. Upon physical examination, the child appears pale, anicteric, diaphoretic and has tachycardia. Abdominal examination reveals splenomegaly without hepatomegaly. Serum transaminase concentrations and the coagulation profile are normal.

Of the following, the MOST likely cause of this child's symptoms is:

- A. acetaminophen toxicity
- B. congenital hepatic fibrosis
- C. hemolytic anemia
- D. portal hypertension
- E. Reye syndrome

17. A term newborn appears jaundiced at 8 hours of age. Her total serum bilirubin concentration is 6.0 mg/dL, and the conjugated fraction is 0.4 mg/dL. She has a vigorous cry, an active suck and normal vital signs.

Of the following, the MOST appropriate plan of action for this infant is to

- A. evaluate thyroid function
- B. examine for hemangiomas
- C. investigate hemolysis
- D. obtain liver function tests
- E. screen for galactosemia

18. The mother of a 4-year-old boy has noted blood on the toilet tissue and a small amount of red blood in the toilet water. She states that the boy's bowel movements have been regular and that he has not experienced pain with their passage. His appetite, activity, and sleeping pattern have been normal, and he has had no abdominal pain.

Of the following, the MOST likely cause of this child's bleeding is:

- A. intussusception
- B. juvenile polyp
- C. Meckel diverticulum
- D. rectal fissure
- E. ulcerative colitis

19. During the health supervision visit for a 3-month-old infant, you observe that he is jaundiced and that his liver is enlarged and firm. Family history is notable for emphysema in his father, paternal grandparents, and paternal uncle, all of whom have never smoked cigarettes.

Of the following, the MOST appropriate test for diagnosing this child's liver disease is

- A. abdominal ultrasonography for choledochal cyst
- B. alpha-1-antitrypsin concentration with genotype
- C. dimethyl iminodiacetic acid scan for biliary atresia
- D. sweat test for cystic fibrosis
- E. urine metabolic screen for tyrosinemia

20. A 5-year-old girl presents after having a brief generalized seizure. Her mother reports that the child has had a 3-day history of fever, tenesmus, and bloody diarrhea. On physical examination, you find a mildly toxic-appearing child who has a temperature of 104°F (40°C) and diffuse abdominal tenderness. The rectal examination produces significant pain. Stool from her rectum is guaiac-positive. You tell her mother that you believe the diarrhea has an infectious cause.

Of the following, the MOST likely pathogen is

- A. *Cryptosporidium* sp
- B. Rotavirus
- C. *Salmonella* sp
- D. *Shigella* sp
- E. *Yersinia* sp

21. A 6-week-old African-American infant has been brought to your office by his grandmother, who thinks he "looks yellow." He has been breastfeeding well and growing along the 50th percentile, but his stools have changed from green to pale yellow in the last 2 weeks. On physical examination, you note mild icterus, normal findings on cardiac evaluation, clear lungs, a liver that is palpable 5 cm below the right costal margin, and a palpable spleen tip. Laboratory studies demonstrate a total bilirubin of 7 mg/dL, direct bilirubin of 3 mg/dL, alanine aminotransferase of 120 U/L, and gamma glutamyl transpeptidase of 700 U/L. Results of abdominal ultrasonography are unremarkable, but the gallbladder is not visualized.

Of the following, the MOST appropriate next management step is

- A. DISIDA (99mTc-disofenin) scan of the liver
- B. echocardiography
- C. hemoglobin electrophoresis
- D. measurement of urine reducing substances
- E. sweat test

BILIARY ATRESIA

22. A 10-year-old boy presents with the acute onset of abdominal pain that was followed shortly thereafter by vomiting. The pain is described as periumbilical. There is no history of trauma. He has had no fever, sore throat, headache, dysuria, or diarrhea. On physical examination, the child is in obvious distress. Other than mild tachycardia, his vital signs are within normal limits. His abdomen is diffusely tender, with localization to the periumbilical area. He also complains of increased abdominal pain when his heel is struck by the examiner's hand. Rectal examination reveals no evidence of occult blood.

Of the following, the MOST likely cause of this child's abdominal pain is

- A. acute pancreatitis
 - B. appendicitis
 - C. intussusception
 - D. pyelonephritis
 - E. small bowel obstruction
23. A 3-year-old child has a history of abdominal distension and rectal prolapse. Rectal examination reveals decreased anal tone and stool at the anal verge. Abdominal radiography shows a distended large intestine and stool in the distal colon and rectum.

Of the following, the MOST likely cause of this child's symptoms is

- A. cystic fibrosis
 - B. functional constipation
 - C. Hirschsprung disease
 - D. hypothyroidism
 - E. intestinal pseudo-obstruction
24. A 3,200-g term male infant presents for his 1-week evaluation. The pregnancy and delivery were uneventful. His parents state that the baby has been difficult to arouse, is feeding poorly, and has been vomiting. On physical examination, the infant weighs 2,800 g, is lethargic and jaundiced, and has a palpable liver 3 cm below the right costal margin. An evaluation for sepsis has been performed and a gram stain of the blood shows gram-negative rods. Additional laboratory results include: total bilirubin, 18 mg/dL; direct bilirubin, 6 mg/dL; alanine aminotransferase, 104 U/L; aspartate aminotransferase, 150 U/L; and positive urine dipstick for protein and reducing substances.

Of the following, the test that would be MOST helpful in the diagnosis of this patient is

- A. Coombs test and maternal anti-Rh titer
- B. examination of a blood smear for red cell morphology
- C. serum amino acids and urine organic acids
- D. serum galactose-1-phosphate uridylyltransferase level
- E. serum hepatitis B surface antigen

Q&A Re

25. A 15-year-old boy presents with red urine following football practice. Findings on physical examination are normal. Urinalysis reveals pH, 6.0; specific gravity, 1.020; blood, +4; protein, trace; and 0 to 2 red blood cells per high power field.

Among the following, the MOST likely explanation for the red color of the urine is

- A. hematuria
- B. ingestion of food coloring
- C. myoglobinuria
- D. presence of urates
- E. trauma

26. You discover a right-sided, palpable abdominal mass in a newborn male who has had an uncomplicated perinatal course. Laboratory evaluation reveals: creatinine, 0.7 mg/dL; normal findings on urinalysis; and platelet count, 350,000/mm³.

Of the following, the MOST likely cause of this mass is

- A. autosomal recessive polycystic kidney disease
- B. midgut volvulus
- C. renal vein thrombosis
- D. multicystic dysplastic kidney
- E. Wilms tumor

27. A 14-year-old girl who never has had menstrual bleeding reports that she has had intermittent lower abdominal pain for the past year. Physical examination reveals a midline lower abdominal mass that is 5 cm in diameter and mildly tender. Sexual maturity rating is 5 for both breast and pubic hair. She states that she has not been sexually active.

The MOST likely cause of this patient's intermittent lower abdominal pain and abdominal mass is

- A. fecal impaction
- B. imperforate hymen
- C. intrauterine pregnancy
- D. müllerian agenesis
- E. tubo-ovarian abscess

28. A 2-week-old male infant is being seen for a routine health supervision visit. Physical examination reveals a distended abdomen, and the bladder can be palpated above the symphysis pubis. The mother has noticed that urine "dribbles" from his urethra.

Of the following, the MOST likely diagnosis is

- * A. posterior urethral valves
- B. prune belly syndrome
- C. urethral diverticula
- D. urethral meatal stenosis
- E. urethral stricture

29. A 9-year-old girl has had intermittent burning on urination and lower abdominal pain for 6 months. Urine cultures obtained at the onset of symptoms and periodically since then have been negative. She appears healthy and has had normal growth. Urinalysis reveals: pH, 6; specific gravity, 1.029; protein, negative; and 20 to 30 red blood cells/high power field without casts.

The most appropriate NEXT study to perform is

- A. another culture of the urine
- * B. determination of the urinary calcium-to-creatinine ratio
- C. measurement of serum complement
- D. measurement of serum immunoglobulin A
- E. renal ultrasonography

30. A 17-year-old sexually active boy has been complaining of increasing scrotal pain, fever, and dysuria for 5 days. Physical examination reveals tenderness and swelling of the left testis, especially posterolaterally; the tenderness decreases when the testis is elevated. Findings on rectal examination are normal. Urinalysis shows many white blood cells.

Among the following, the MOST likely diagnosis is

- A. epididymitis
- B. incarcerated inguinal hernia
- C. prostatitis
- D. testicular torsion
- E. varicocele

31. A 6-year-old boy presents with tea-colored urine. He was diagnosed with an upper respiratory tract infection 2 days ago. His parents report that a similar episode occurred 4 months ago. There is no history of trauma. Findings include: blood pressure, 105/70 mmHg; temperature, 38°C (100.4°F); and absence of rash or joint abnormalities. Serum complement level is normal.

The MOST likely explanation of these findings is

- A. Alport hereditary nephritis
- B. idiopathic hypercalciuria
- C. immunoglobulin A nephropathy (Berger disease)
- D. postinfectious acute glomerulonephritis
- E. thin glomerular basement membrane nephropathy

32. A 6-year-old girl is admitted for evaluation of anasarca. Findings include: serum creatinine, 0.5 mg/dL; albumin, 1.8 g/dL; cholesterol, 300 mg/dL; triglycerides, 350 mg/dL; C3 complement, 100 mg/dL (normal, >80 mg/dL); antinuclear antibody, negative; and urinalysis, 2 to 5 red blood cells per high power field and urine protein, 300 mg/dL.

These findings are MOST consistent with the diagnosis of

- A. membranous nephropathy
 - B. membranoproliferative glomerulonephritis
 - C. minimal change nephrotic syndrome *PROTINURIA, LOW ALBUMIN, EDEMA, CHOLESTEROL PROB.*
 - D. postinfectious acute glomerulonephritis
 - E. systemic lupus erythematosus
33. An asymptomatic 14-year-old boy has normal physical examination results. Routine urinalysis reveals: specific gravity, 1.015; pH, 6.0; negative blood; 2+ protein; 0 to 2 red blood cells; and 0 to 2 white blood cells. A repeat urinalysis 1 week later reveals the same results.

Of the following, the MOST appropriate next step is to

- A. evaluate him again in 6 months
- B. obtain a first-morning urine specimen
- C. obtain a 24-hour urine collection
- D. obtain serum electrolyte measurements
- E. refer him to a nephrologist

ORTHOSTATIC PROTINURIA

34. The parents of a 3-year-old girl bring her to the emergency department, stating that 1 hour ago she vomited "a cupful" of bright red blood. Physical examination demonstrates a pale, quiet, anicteric child. Pulse is 110 beats/min and blood pressure is 80/40 mmHg. The liver edge is firm and palpable approximately 3 cm below the right costal margin (7 cm span). The spleen is palpable 4 cm below the left costal margin. Past medical history is pertinent for enlarged kidneys that were palpated initially at 1 year of age. At that time, computed tomography showed enlarged kidneys that contained many cysts.

Of the following, the MOST likely underlying cause of the bleeding is

- A. alpha-1-antitrypsin deficiency
- B. autoimmune hepatitis
- C. biliary atresia
- D. cavernous transformation of the portal vein
- E. congenital hepatic fibrosis

35. A 7-year-old child is admitted with cola-colored urine and hypertension. She had an upper respiratory tract infection 2 weeks ago. Findings include: periorbital edema; blood pressure, 150/100 mmHg; creatinine, 1.2 mg/dL; C3 complement, 20 mg/d (normal, >80 mg/dL); antinuclear antibody titer, negative; and red blood cells and casts in urine are too numerous to count.

Of the following, the MOST likely diagnosis is

- A. Goodpasture syndrome
- B. immunoglobulin A nephropathy
- C. postinfectious acute glomerulonephritis
- D. systemic lupus erythematosus
- E. Wegener granulomatosis

36. An 18-year-old boy comes to your office complaining that he feels a heavy, dragging sensation in his left groin following exercise. He is not sexually active, and there is no history of trauma. Findings on physical examination include sexual maturity rating (Tanner) stage 5 genital development and a noticeable fullness above the left testis, which decreases when the patient lies down.

Of the following, the MOST likely explanation for these findings is a(n)

- A. hydrocele
- B. inguinal hernia
- C. spermatocele
- D. testicular tumor
- E. varicocele

37. During a 2-week health supervision visit, a mother expresses concern about her son's poor feeding and small penis. On physical examination, you confirm micropenis and note hypotonia.

Of the following, the MOST likely diagnosis is

- A. Kallman syndrome
- B. Klinefelter syndrome
- C. Down syndrome
- D. Prader-Willi syndrome
- E. septo-optic dysplasia

H₃O

Hypotonia
Hypomotile
Hypogonadism

38. A 10-year-old boy presents for his annual preparticipation sports physical examination. His father has renal failure. On physical examination, the boy's blood pressure is 145/100 mmHg and pulse is 90 beats/min. Urinalysis reveals 10 to 20 red blood cells per high power field, and the serum creatinine level is 0.7 mg/dL. Renal ultrasonography shows enlarged kidneys, with three renal cysts in each.

Of the following, the MOST likely diagnosis in this child is

- A. autosomal dominant polycystic kidney disease
- B. autosomal recessive polycystic kidney disease
- C. juvenile nephrophthisis
- D. medullary sponge kidney
- E. multicystic dysplastic kidney disease

39. A 15-year-old boy who recently emigrated from Mexico complains of a painful left testicle for 2 days associated with fever. He denies a history of sexual activity. He is noted on physical examination to have Sexual Maturity Rating (Tanner) stage 4 genitalia, with a diffusely tender left testis. The overlying scrotal skin is mildly erythematous. There is no penile discharge. Results of Doppler ultrasonography of the testes are normal.

Of the following, the next MOST appropriate study to obtain is

- A. complete blood count
- B. culture for *Neisseria gonorrhoeae*
- C. erythrocyte sedimentation rate
- D. mumps serology
- E. polymerase chain reaction for *Chlamydia trachomatis*

40. One month ago, a 10-year-old boy presented to you with edema of the face, hands, and feet. Urinalysis revealed 20 to 25 red blood cells and 4+ protein. You diagnosed nephrotic syndrome and placed him on a recommended regimen of oral steroids. His face now exhibits cushingoid features, but his edema has not subsided. Urinalysis today reveals 4+ protein and moderate blood.

Of the following, the MOST likely diagnosis at this time is

- A. focal segmental glomerulosclerosis
 B. Henoch-Schönlein purpura
 C. immunoglobulin A nephropathy
 D. minimal-change nephrotic syndrome
 E. postinfectious acute glomerulonephritis
41. A 17-year-old boy presents to the school health center after feeling a testicular mass while performing a monthly self-examination.

Of the following, males at HIGHEST risk for testicular cancer are those who have had

- A. abdominal radiation
 B. anabolic steroids use
 C. cryptorchidism
 D. gynecomastia
 E. pesticide exposure
42. A 3-month-old boy is admitted to the hospital for evaluation of failure to thrive. His birthweight was at the 50th percentile and length at the 75th percentile. Currently his weight is below the 5th percentile and length is at the 25th percentile. His vital signs and physical examination results are otherwise normal. He appears well hydrated. Measurement of serum electrolytes reveals: sodium, 139 mEq/L; potassium, 4.7 mEq/L; chloride, 114 mEq/L; bicarbonate 12 mEq/L; blood urea nitrogen, 8 mg/dL; and creatinine, 0.3 mg/dL. A consulting nephrologist recommends measurement of urine pH (which is 8).

Of the following, the MOST likely cause of this child's acidosis is

- A. inborn error of metabolism
 B. lactic acidosis
 C. type I (distal renal tubular) acidosis
 D. type II (proximal renal tubular) acidosis
 E. cystic fibrosis

43. The mother of a 7-year-old girl is concerned because her daughter has intermittent daytime and nighttime wetting. She was toilet trained by the age of 3.5 years, but the enuresis has been present for several years. The child is developing along the 75th percentile for height and weight. The mother has withheld liquids after 6:00 PM each night, with subsequent slight improvement during the night but continued wetting during the day. Results of a urinalysis and urine culture are negative, and renal ultrasonography and serum electrolyte measurements are normal.

Of the following, the BEST course of management is to

- A. perform voiding cystourethrography
- B. prescribe oxybutynin
- C. prescribe prophylactic antibiotics
- D. refer for psychotherapy
- E. wait 6 months and re-evaluate

44. During the evaluation of a 5-year-old boy for poor speech development, you discover sensorineural hearing loss. His past medical history includes several episodes of otitis media between 2 months and 2 years of age. Several family members have kidney disease. The boy has microscopic hematuria.

Of the following, the MOST likely mode of inheritance of his disease is

- A. autosomal dominant
 - B. autosomal recessive
 - C. random mutation
 - D. x-linked dominant
 - E. x-linked recessive
- ALPHEG*

45. A 7-year-old girl presents with a rash on the lower extremities that extends to the buttocks. She also reports swollen and aching knees and ankles and intermittent abdominal pain. Her vital signs are normal, and the physical examination reveals swollen and tender joints and abdominal tenderness. She has multiple purpura on her lower extremities. Her complete blood count and serum electrolyte concentrations are normal.

Based on her presentation, the BEST course of action is to

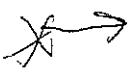
- A. check an ANA
- B. check a random urine specimen for protein and creatinine
- C. schedule a renal biopsy
- D. schedule renal ultrasonography
- E. start therapy with azathioprine

HUS
PURPURA
ARTHRITIS
ABD PAIN
KIDNEY

46. A 5-year-old boy is hospitalized following a motor vehicle accident. Evaluation reveals a depressed occipital skull fracture and displaced femur fracture. He develops polyuria postoperatively. You decide to measure serum and urine sodium (Na) as well as urine osmolality (Osm).

Of the following, the findings that are MOST consistent with the diagnosis of diabetes insipidus in this boy are

- A. serum Na, 120 mEq/L; urine Na, 10 mEq/L; urine Osm, 600 mOsm/kg H₂O
- B. serum Na, 120 mEq/L; urine Na, 50 mEq/L; urine Osm, 300 mOsm/kg H₂O
- C. serum Na, 140 mEq/L; urine Na, 50 mEq/L; urine Osm, 300 mOsm/kg H₂O
- D. serum Na, 160 mEq/L; urine Na, 10 mEq/L; urine Osm, 600 mOsm/kg H₂O
- E. serum Na, 160 mEq/L; urine Na, 10 mEq/L; urine Osm, 100 mOsm/kg H₂O



FACTS – INFECTIOUS DISEASE AND NEUROLOGY

Infant botulism is bulbar weakness and paralysis that develops in infants during the first year of life secondary to ingestion of *Clostridium botulinum* spores and absorption of botulinum toxin. The source is infected foods, such as contaminated honey, or spores unearthed from the ground. The toxin prevents the presynaptic release of acetylcholine. Onset of symptoms is 12 to 48 hours after ingestion of spores, with constipation as the classic first symptom. Neurologic symptoms follow, including weak cry and suck, loss of previously obtained motor milestones, ophthalmoplegia and hyporeflexia, with a symmetric and descending paralysis. Diagnosis is based on suggestive history, neurologic exam, identification of toxin or bacteria in the stool, and EMG showing brief, small-amplitude muscle potentials with an incremental response during high-frequency stimulation. Treatment is supportive with NG feeds, assisted ventilation as needed, and botulism immune globulin. Antibiotics are contraindicated and may worsen the clinical course. The outlook is excellent, and complete recovery is expected over several weeks or months.

Tetanus is caused by a neurotoxin from the anaerobic bacterium *Clostridium tetani* and is characterized by muscle spasms that are often worsened by noise or other external stimuli, progressing from mild to severe over a one-week period and subsiding over a period of weeks if the patient survives. The clinical manifestations of tetanus include localized tetanus (local muscle spasm in areas contiguous with the infected wound), cephalic tetanus (dysfunction of the cranial nerves in association with a wound on the head or neck), and generalized tetanus (full body involvement). Neonatal tetanus remains prevalent in areas of the world where mothers have not been vaccinated and local umbilical cord care is poor. The diagnosis is made clinically, and treatment includes the use of human tetanus immune globulin. In addition, all infected wounds should be cleaned properly and debrided.

Lyme disease is a reactive inflammatory disorder of the skin, heart, CNS, and connective tissue, caused by a spirochetal infection with *Borrelia burgdorferi* and transmitted via tick bite. High-risk areas reflect the natural habitat of ticks of the Ixodes species (infected deer tick) which are the vectors for Lyme disease transmission, especially prevalent in woodlands and fields in the New England states. Early disease includes erythema migrans (the classic rash that develops in 2/3 of patients and is described as annular and “target-like” with variable degrees of central clearing), constitutional symptoms like fever/headache/myalgias/fatigue/arthritis/lymphadenopathy, neurologic findings such as aseptic meningitis/bilateral facial nerve palsy/encephalitis, and carditis that usually presents as heart block. The hallmark of late disease is arthritis. Laboratory diagnosis involves serologic testing with ELISA and Western blot assay. Early localized disease or late disease with arthritis is typically treated with either doxycycline (for children >8 years of age) or amoxicillin. Carditis and meningitis require IV ceftriaxone or penicillin. Most children have an excellent prognosis. Rocky Mountain spotted fever is caused by *Rickettsia rickettsii*, a Gram-negative intracellular coccobacillus that is transmitted by the bite of a tick. Incidence is highest in school-age children and occurs during the spring and summer, most often in the Southeastern regions of the United States. Clinical features include fever, myalgias, a petechial rash that begins on the extremities (ankles and wrists) and moves in a caudal and centripetal direction,

hepatosplenomegaly and jaundice, CNS symptoms such as headache, and hypotension. Significant laboratory findings include thrombocytopenia and hyponatremia. Diagnosis is made clinically but should be confirmed with serologic tests for rickettsia. Treatment includes oral or IV doxycycline and supportive care – antibiotics are usually started empirically, given the possibility of significant morbidity or mortality in untreated infection.

Pertussis is an acute respiratory infection also known as “whooping cough.” Caused by *Bordetella pertussis*, infants younger than 6 months of age are most at risk for severe disease, and adolescents and adults whose immunity has waned are the major source of pertussis infection of unimmunized or under-immunized children. This highly contagious disease is characterized by three stages: catarrhal stage (lasts 1 to 2 weeks with URI symptoms), paroxysmal stage (lasts 2 to 4 weeks with the hallmark fits of forceful coughing that are often followed by a whoop or inspiratory gasp in older children and rarely in young infants, post-tussive vomiting is common and young infants may have cyanosis, apnea, and choking during the paroxysms), and convalescent phase (lasts weeks to months and is a recovery stage in which the paroxysmal cough continues but becomes less frequent and severe). Diagnosis is suspected based on clinical features and an elevated white blood cell count with lymphocytosis, but is confirmed based on a pertussis PCR. Management includes hospitalization of young infants, supportive care and oxygen as needed, antibiotics given to all patients and contacts to prevent the spread of infection (azithromycin or erythromycin is used). Respiratory isolation is needed until antibiotics have been given for at least 5 days.

Histoplasmosis is a common granulomatous infection caused by the dimorphic fungus *Histoplasma capsulatum*. Most common in the Ohio, Mississippi, and Missouri River Valleys in the United States, histoplasmosis comes from soil contaminated by animal droppings, especially birds and bats, and active disturbance of that environment is necessary for dissemination of the spores. Acute pulmonary histoplasmosis is the most common form of the disease, and if symptomatic, presents as an influenza-like illness with abrupt onset of fever, malaise, headache, myalgia, and nonproductive cough, with chest x-ray revealing patchy pneumonic infiltrates and hilar adenopathy. *Coccidioides immitis* is also a dimorphic fungus that causes primary pulmonary infection with possible lymphohematogenous dissemination. Coccidiomycosis occurs primarily in the Southwestern United States, being endemic in Southern California, Arizona, New Mexico, and Western and Southern Texas. 60% of infections are subclinical, with 40% presenting with a flu-like illness, occasionally with erythema nodosum. Treatment is not required in most patients.

Mammalian bites typically occur in boys during the spring and summer months. Younger children are typically bitten on the head and neck, whereas older children are bitten predominantly on the extremities. Cat bites are more likely to be infected than dog bites because of the increased incidence of puncture wounds. Victims of animal bites have a high risk of infection due to *Pasteurella multocida* with pain, redness, and swelling within the first 1 to 2 days after the bite. Treatment includes copious wound irrigation and antibiotics such as amoxicillin-clavulanic acid, with tetanus prophylaxis if indicated. Human bites are typically located on the trunk or face in young children, or, if occurring during a fist fight, located at the metacarpophalangeal joint. Infection rate is high, with *Eikenella corrodens* being the most common. Management includes copious wound irrigation and antibiotics.

Croup (laryngotracheobronchitis) is inflammation and edema of the subglottic larynx, trachea, and bronchi, most often caused by parainfluenza viruses. This typically occurs in children 3 months – 3 years and begins in the late fall and winter with an upper respiratory infection prodrome for 2 to 3 days, followed by inspiratory stridor, fever, barking cough, and a hoarse voice, which typically lasts 3 to 7 days, with respiratory distress and stridor worsening at night and with agitation. AP radiograph of the neck demonstrates the “steep sign” of subglottic narrowing. Treatment includes supportive care with cool mist and fluids, intramuscular dexamethasone (corticosteroids) for children with stridor at rest, and possible racemic epinephrine for children with significant respiratory distress. Bronchiolitis is inflammation of the bronchioles, predominantly affecting children younger than 2 years of age. Epidemics occur from November to April and risk of infection is increased with daycare attendance, multiple siblings, exposure to tobacco smoke, and lack of breast-feeding. More significant disease occurs in patients with chronic lung disease, CHD, history of prematurity, immunodeficiency, and in infants younger than 3 months. RSV is the most common cause and clinical features include rhinorrhea, nasal congestion, fever, and cough progressing to tachypnea, fine rales, wheezing, and evidence of respiratory distress. Hypoxemia and apnea may occur. Chest x-ray reveals hyperinflation with air trapping, patchy infiltrates, and atelectasis. Improvement is noted within 2 weeks. Treatment is primarily supportive with bulb suctioning, hydration, and oxygen. Prophylaxis with RSV monoclonal antibody is given to infants with a history of prematurity.

Diarrheal disease and resulting dehydration are among the most common causes of childhood morbidity and mortality worldwide. Rotavirus is the most common infectious agent causing gastroenteritis. This RNA virus is usually seen in the winter months and is spread by the fecal-oral route, having an incubation period of 1-3 days. Patients may be asymptomatic or may have significant vomiting, diarrhea, and dehydration, often self-limited and lasting 4-7 days. Diagnosis can be made by a stool ELISA with absent WBCs. Treatment is supportive with particular attention to fluid management and early institution of feedings to prevent gut atrophy. Some children may develop transient lactose intolerance. Norwalk virus is an RNA virus that is linked to outbreaks of gastroenteritis in all age groups, particularly in closed populations (daycare centers, schools, cruise ships – typically vomiting is more prominent and there is a shorter duration of symptoms). Enterotoxigenic *E. coli* (ETEC) is the major cause of traveler’s diarrhea, generally a noninvasive watery diarrhea without stool WBCs. Bacterial causes typically present with blood and leukocytes in the stool: enterohemorrhagic *E. coli* (0157:87) is responsible for hemolytic uremic syndrome; *Shigella sonnei* is associated with seizures secondary to neurotoxin released; Salmonella species is associated with exposure to poultry/milk/eggs/reptile exposure; *Campylobacter jejuni* is the most common cause of bacterial bloody diarrhea in the U.S. and can lead to Guillain-Barré syndrome; *Yersinia enterocolitica* can lead to a mesenteric adenitis that mimics acute appendicitis; *Clostridium difficile* leads to colitis following antibiotic usage.

Subacute lymphadenitis is typically caused by one of two infectious agents. Cat-scratch disease, caused by the Gram-negative bacteria *Bartonella henselae*, presents with regional lymphadenopathy (especially in the axillary, cervical, or inguinal region), distal to and after a cat or kitten scratch. The initial scratch results in a papule along the line of the scratch, followed by lymphadenopathy one to two weeks later, with the lymph node being erythematous, warm, and tender, with fever occurring in 1/3 of patients. Other findings may include Parinaud

oculoglandular syndrome (conjunctivitis and preauricular lymphadenitis) and hepatitis. Diagnosis is made by elevated serum IgM antibody to *Bartonella henselae*, and treatment is usually supportive, with antibiotics reserved for patients with systemic disease or immunodeficiencies (azithromycin, Bactrim, and Ciprofloxin – surgery is not indicated and may result in persistent fistulous tracts). The most difficult differential distinction in this clinical situation is atypical nontuberculosis mycobacterial infection caused by *Mycobacterium avium-intracellulare*. NTM usually is acquired from soil, dust, water, and contaminated food and presents as a local lymphadenitis without systemic illness, with the eventual development of nodal necrosis and overlying skin inflammation, most often in the submandibular/submaxillary and parotid regions. A mildly positive PPD suggests the diagnosis, but culture of the node is necessary for confirmation following the preferred treatment of complete surgical excision.

Infectious mononucleosis, usually caused by Epstein-Barr virus, is often asymptomatic in young children, but develops typical signs and symptoms in older children – fever, malaise and fatigue, pharyngitis (typically exudative, resembling GABHS pharyngitis), posterior cervical lymphadenopathy, and hepatosplenomegaly. A CBC may demonstrate atypical lymphocytes, a monospot may be positive (detects the presence of heterophile antibody and is less sensitive in children under 4 years of age), and EBV antibody titers may be positive. Treatment is typically supportive, although corticosteroids may be used for severe pharyngitis. Complications can include neurologic problems (cranial nerve palsies and encephalitis), severe pharyngitis that may cause upper airway obstruction, splenic rupture, malignancy (EBV has been isolated from nasopharyngeal carcinoma and Burkitt's lymphoma), and amoxicillin-associated rash (diffuse, pruritic maculopapular rash in patients misdiagnosed with GABHS pharyngitis and historically associated with ampicillin).

Measles, also known as rubeola, or 10-day measles, is a highly infectious virus that presents with the classic prodrome of the three Cs (Cough, Conjunctivitis and Coryza). The enanthem is characterized by Koplik spots, small gray papules on an erythematous base located on the buccal mucosa (these are pathognomonic of measles and are present before the generalized exanthem). Exanthem is characterized as an erythematous maculopapular eruption that begins around the neck and ears and spreads down the chest and upper extremities during the subsequent 24 hours, later becomes confluent, and is accompanied by high fever. Although bacterial pneumonia is the most common complication and the most common cause of mortality, other complications include otitis media, laryngotracheitis, encephalomyelitis, and subacute sclerosing panencephalitis. Diagnosis is based on clinical features and confirmation of measles by serologic testing. Management includes supportive care, possible use of vitamin A, and immunoglobulin for post exposure prophylaxis in high-risk individuals who are exposed to measles. Rubella, also known as German measles or 3-day measles, is a highly infectious virus that is often mild and asymptomatic. If symptomatic, painful lymphadenopathy of the suboccipital, posterior auricular, and cervical nodes precedes the non-pruritic, maculopapular, confluent exanthem. It begins on the face, spreads to the trunk and extremities, and lasts for 3 days, usually with minimal fever. Major complication is congenital rubella syndrome, which occurs after primary maternal infection during the first trimester with fetal anomalies occurring in 30-50% of infected fetuses, presenting clinically with thrombocytopenia, hepatosplenomegaly, jaundice, purpura (“blueberry muffin rash”), and a triad of congenital cataracts, sensorineural hearing loss, and a PDA.

Mycoplasma pneumoniae is one of the most common causes of pneumonia in older children and adolescents, presenting with low-grade fever, chills, nonproductive cough, headache, pharyngitis, and malaise. The cough may last 3-4 weeks and lung examination reveals widespread rales. Although positive cold agglutinins are suggestive and chest x-ray findings may show bilateral diffuse infiltrates, definitive diagnosis is made by elevation of serum IgM titers for mycoplasma. Management includes oral erythromycin or azithromycin. *Chlamydia pneumoniae* is another common cause of pneumonia in the 5-20 year-old age group, presenting with pharyngitis, hoarseness, fever, cervical adenopathy, and productive cough, and treatment is likewise with a macrolide.

Roseola (exanthema subitum or 6th disease) is a febrile illness occurring almost exclusively in children younger than 3 years of age and is caused by HHV6. Roseola begins with a high fever, averaging 103° F, for 2 to 5 days, followed by a pink macular rash that develops soon after a rather abrupt defervescence. Approximately 10-15% of affected infants may have seizures during the febrile phase of roseola. Treatment is supportive.

Parvovirus B19 is the cause of erythema infectiosum (fifth disease). The most common findings in symptomatic disease are mild fever and a distinctive rash, characterized by intense erythema of the cheeks ("slapped cheek" appearance) and a symmetric, maculopapular lacy reticulated rash that may develop on the trunk and spread to the extremities. Three other clinical manifestations are common with parvovirus B19 – arthralgia and arthritis (small joints of the hands and occasionally the ankles, knees, and wrists, most often in adolescent or adult females); nonimmune hydrops fetalis; and a transient aplastic crisis in a patient with a chronic hemolytic anemia (sickle cell disease, G6PD deficiency, autoimmune hemolytic anemia ...).

Adenovirus can cause several clinical syndromes in children, ranging from severe pneumonia in infants to upper respiratory tract infections in older children that often are accompanied by conjunctivitis, pharyngitis, and occasionally gastroenteritis. There is a slight increased incidence of adenoviral infections from late winter through early summer, and epidemics have been associated with contaminated swimming pools. Although adenovirus generally is thought of as the primary cause of keratoconjunctivitis, it also is a frequent cause of respiratory symptoms including pharyngoconjunctival fever (pharyngitis, conjunctivitis, high fever). Treatment is supportive.

Toxic shock syndrome is a toxin-mediated illness characterized by fever, shock, desquamating skin rash, and multiorgan dysfunction. *Staphylococcus aureus* is the most common organism associated with TSS (especially in young women using tampons), although an increase in GABHS-associated TSS has been reported. Clinical features include fever, hypotension, diffuse macular erythroderma (appears similar to sunburn), desquamation, multisystem involvement (GI, myalgias, conjunctivitis, pyuria, thrombocytopenia, CNS), and a positive blood culture for *Staphylococcus aureus*. Treatment includes supportive measures to reverse shock, anti-staphylococcal antibiotics, and removal of the nidus of infection (tampon) if present. IVIG may have some benefit.

Congenital TORCH infections typically cause mild infection with few symptoms in the mother, but may have serious effects on the baby, including but not limited to: congenital heart disease, hearing impairment, mental retardation, low blood counts (anemia and thrombocytopenia), hepatosplenomegaly, pneumonia, microcephaly, jaundice, visual problems, and a "blueberry muffin" skin rash. CMV is by far the most common congenital infection with frequent hearing loss and periventricular calcifications (however, 90-95% are asymptomatic). Congenital toxoplasmosis tends to produce more ophthalmologic findings, diffuse cerebral calcifications, and communicating hydrocephalus. Congenital rubella often leads to the triad of deafness (sensorineural), cataracts, and a pDA.

Epileptic syndromes are characterized by a specific age of onset, seizure characteristic, and EEG abnormality. Benign rolandic epilepsy (benign centrotemporal epilepsy) is the most common partial epilepsy during childhood and commonly presents at 3 to 13 years of age, more common in males, with an autosomal dominant variable penetrance inheritance. Seizures typically occur in early morning hours when patients are asleep with oral-buccal manifestations (moaning, grunting, pooling of saliva – seizure may spread to face and arm, then generalize into tonic-clonic seizures). The EEG shows biphasic spike and wave disturbances in the mid-temporal and central regions. Many patients may not require anticonvulsant prophylaxis, but if needed, valproic acid and carbamazepine are good choices. Outcome is excellent, and seizures remit spontaneously during adolescence with no adverse effects on development or cognition. Absence epilepsy of childhood presents between 5 and 9 years of age with a female-to-male predominance of 3:2, inherited in an autosomal dominant fashion with age-dependent penetrance. Typical presentation includes 5-to-20-second lapses in consciousness many times per day, often accompanied by automatisms, such as eye blinking and incomprehensible utterances (loss of posture, urinary incontinence, and a postictal state do not occur). EEG reveals a generalized 3-Hz/second spike and wave discharge arising from both hemispheres, and treatment includes ethosuximide (first-line drug) or valproic acid. Outlook is very good with seizures usually resolving by adolescence without cognitive impairment. Infantile spasms (West syndrome) typically occur in children 3-8 months of age and are characterized by brief, myoclonic jerks lasting 1-2 seconds each, occurring in clusters of 5-10 seizures spread over 3-5 minutes (also known as jackknife seizures or salaam seizures). Although tuberous sclerosis is the most commonly identified cause, other causes include PKU, HIE injury, IVH, meningitis, and encephalitis. EEG shows the characteristic hypsarrhythmia pattern, and management includes ACTH, valproic acid, and vigabatrin (most effective for tuberous sclerosis). Outlook is poor with most children developing moderate to severe mental retardation. Juvenile myoclonic epilepsy (myoclonic epilepsy of Janz) presents with myoclonic jerks between 12 and 16 years of age, especially prevalent upon awakening and sometimes causing an adolescent to inadvertently throw objects during the morning routine. The generalized tonic-clonic seizures that also occur in this disorder are of greater concern. EEG reveals a 4 to 6 per second generalized spike or polyspike and wave activity. Typically responds well to valproic acid, but life-long treatment is usually required.

Meningitis is inflammation of the meninges and is classified as bacterial or aseptic. Risk factors for bacterial meningitis include young age (most common during the first month of life), immunodeficiency (asplenia, humoral-mediated immunodeficiency, and terminal complement deficiency), and anatomic defects (basilar skull fracture, VP shunt). Infants often present with

minimal and nonspecific signs and symptoms (poor feeds, irritability, lethargy, respiratory distress, often with a bulging fontanelle). Older children often present with fever and signs suggestive of meningeal irritation, including alteration in LOC, nuchal rigidity, seizures, photophobia, emesis, and headache. Lumbar puncture demonstrates pleocytosis with a predominance of neutrophils, low CSF glucose, increased protein, and positive Gram-stain and culture. Common bacterial causes from 0-2 months include GBS/*Streptococcus agalactiae* > *E. coli* > *Listeria monocytogenes* – treat with ampicillin plus an aminoglycoside or 3rd-generation cephalosporin (IV acyclovir for possible HSV infection should be considered for ill neonates, especially those presenting with apnea, seizures, or cutaneous vesicles). Older infants and children (>2 months) get infections with *Streptococcus pneumoniae* > *Haemophilus influenzae* type b > *Neisseria meningitidis* – treatment with 3rd-generation cephalosporin +/- vancomycin if meningitis is highly suspected. Complications include hearing loss (the most common complication), global brain injury in 5-10% of patients, SIADH, seizures, hydrocephalus, brain abscess, cranial nerve palsy, learning disability, and focal neurologic deficits. Viral aseptic meningitis causes a CSF lymphocytic pleocytosis with normal CSF glucose and normal to minimally elevated CSF protein – enteroviruses are the most common cause of viral meningitis, often in the summer and fall, and may have a co-existing macular rash; *Mycobacterium tuberculosis* presents initially with nonspecific lethargy or irritability, but during the second week of illness, progresses rapidly with findings that include cranial nerve deficits/altered LOC/coma/paraplegia/eventual death if untreated, often with very high CSF protein and low glucose; HSV meningitis typically presents with fever, mental status changes, and focal seizures or focal status encephalitis, with periodic lateralized epileptiform discharges/PLEDS on EEG, often with increased numbers of red blood cells in the CSF and enhancement of the temporal lobes on MRI. The neonatal form of herpes presents at 1-3 weeks of life with three different presentations: SEM disease – localized to the skin, eyes, and mouth with discrete vesicles on an erythematous base; disseminated disease – generalized and involves the liver, adrenals, lungs, and often the CNS; and localized CNS disease that may be seen with the use of fetal scalp electrodes.

Migraine headaches are the most common cause of headaches in children and adolescents, occurring in up to 5% of school-age children. Before puberty, incidence is higher in males; after puberty, incidence is much higher in females. Typical clinical features include a prolonged, throbbing, unilateral headache that starts in the supraorbital area and often radiates to the occiput. In young children, the headache is often bifrontal. Other symptoms include nausea and vomiting, visual disturbances including blurred vision/scotomata/jagged streaks of light, photophobia or phonophobia, minimal relief from OTC meds, symptoms improved by sleep, and normal neurologic exam. Treatment includes rest, elimination of known triggers, abortive treatment with sumatriptan (a selective 5-HT agonist), and prophylactic treatment with propranolol (drug of choice). Tension headaches are bifrontal or diffuse, dull, aching headaches that are often associated with muscle contraction. They are unusual during childhood and extremely rare in children younger than 7 years of age. The pain increases in intensity during the day and isometric contraction of the temporalis, masseter, or trapezius muscle often accompanies the headache. No vomiting, visual changes, or paresthesias occur. Treatment includes reassurance and pain control, with stress and anxiety reduction to help provide long-term relief. Cluster headaches, also rare during childhood, are characterized by unilateral frontal or facial pain, accompanied by

conjunctival erythema, lacrimation, and nasal congestion. These headaches usually last less than 30 minutes but may recur several times in a day and then not occur again for weeks or months. Treatment includes abortive therapy with oxygen or sumatriptan. Headaches from brain tumors (or other causes of increased intracranial pressure) are often worse during sleep or upon awakening, with symptoms commonly subsiding during the day as venous return from the head improves with upright posture.

Cerebral palsy is a group of static (nonprogressive) encephalopathies caused by injury to the developing brain in which motor function is primarily affected. Intelligence may be normal, but injury to the brain that causes cerebral palsy often leads to other neurologic effects, including seizures, cognitive defects, MR, learning disabilities, sensory loss, and visual and auditory deficits. Risk factors for CP include prematurity (the most common risk), TORCH infections, traumatic delivery, low Apgar score at 15 minutes, HIE, IVH, and kernicterus. The basis of diagnosis is repeated neurodevelopmental examination showing increasing tone or spasticity, hypotonia, asymmetric reflexes or movement disorder, or abnormal patterns in the disappearance of primitive reflexes or emergence of postural responses. Spastic diplegia is a form of CP that is marked by bilateral spasticity of the lower extremities with very little deficit in the upper extremities – scissoring may be present when the child is suspended vertically, there may be a history of early rolling, and the child may walk on tiptoes – this injury pattern arises commonly in preterm infants from ischemia to the vascular border zone around the cerebral ventricles and resultant periventricular leukomalacia. Spastic hemiplegia is a unilateral spastic motor weakness with upper extremity involvement typically greater than lower extremities – early hand preference, grasping always on the same side, and fisting or absent pincer grasp on the other side – often caused by perinatal vascular insults/postnatal trauma/CNS malformations. Spastic quadriplegia involves motor weakness of the head, neck, and all four limbs, often with seizures/scoliosis/GER/speech problems – due to CNS infections/trauma/malformations/HIE. Extrapyramidal (nonspastic) cerebral palsy results in athetoid movements with problems modulating control of the face, neck, trunk, and limbs – marked hypotonia is present – often seen in full-term infants with HIE or with kernicterus leading to basal ganglia damage.

Guillain-Barré syndrome (acute inflammatory demyelinating polyneuropathy) is a demyelinating polyneuritis characterized by ascending weakness, areflexia, and normal sensation, most commonly associated with gastroenteritis due to *Campylobacter jejuni*. Ascending, symmetric paralysis with ataxia may progress to respiratory arrest and cranial nerve involvement occurs in 50% of patients. Lumbar puncture shows albuminocytologic dissociation (increased CSF protein in the absence of an elevated cell count) and there is decreased nerve conduction velocity. Because of the risk of respiratory muscle paralysis, treatment should be initiated as soon as possible, usually with IVIG (preferred treatment for children) or plasmapheresis. Complete recovery is the rule in children, but physical therapy may be necessary for several weeks or longer to aid in recovery.

Rett syndrome, a neurodegenerative disorder that occurs exclusively in females, presents with normal development until 6 months-1 year of age, when regression of language and motor skills and acquired microcephaly become apparent. An ataxic gait or fine tremor is an early neurologic finding, but the hallmark is repetitive hand-wringing movements and a loss of purposeful and spontaneous use of the hands, which may not appear until 2-3 years of age.

Transverse myelitis is characterized by the abrupt onset of progressive weakness and sensory disturbances in the lower extremities, often preceded by a recent history of a viral infection. This acute demyelinating disorder of the spinal cord occurs over hours or a few days and presents initially with low back or abdominal pain and paresthesias of the legs. The leg muscles are weak and flaccid and a sensory level is present, usually in the midthoracic region. Pain, temperature, and light touch sensation are affected, with the eventual development of sphincter disturbances and hyperreflexia. The diagnosis is confirmed by results of spinal MRI, and spontaneous recovery occurs over a period of weeks or months and is complete in 60% of cases.

Pseudotumor cerebri is a clinical syndrome that mimics brain tumors and is characterized by increased ICP with a normal CSF cell count and protein content and normal ventricular size, anatomy, and position. Although over 90% of cases are idiopathic, the most commonly reported causes are: drugs (tetracycline, excess vitamin A, corticosteroids, nalidixic acid, nitrofurantoin), endocrine disorders (hyperthyroidism, Cushing's syndrome, hypoparathyroidism), and thrombosis of the dural venous sinuses. Occurring most often in adolescent, obese females, the most frequent symptoms are headache and diplopia secondary to paralysis of the abducens nerve. Papilledema with an enlarged blind spot is the most consistent sign in a child beyond infancy. Although mainly a self-limited condition, optic atrophy and blindness can be significant complications. If repeated follow-ups document decreased visual acuity, an initial lumbar tap that follows a CT or MRI scan is both diagnostic and therapeutic. Several additional lumbar taps and the removal of sufficient CSF to reduce the opening pressure by 50% occasionally lead to resolution of the process. Acetazolamide and corticosteroids have been effective for some patients.

Duchenne and Becker muscular dystrophies are progressive, X-linked myopathies characterized by myofiber degeneration with a deletion in the dystrophin gene. Onset of symptoms is between 2 and 5 years of age with a slow, progressive weakness affecting the legs first. In DMD, children lose the ability to walk by 10 years of age, and in BMD, patients lose the ability to walk by 20 or more years of age. Other findings include pseudohypertrophy of the calves, a positive Gowers' sign, and cardiac involvement including cardiomyopathies. Diagnosis is made by elevated CK levels, abnormal EMG or muscle biopsy, and DNA testing. There is no cure but oral steroids can improve strength transiently when the disease is in the early stages. In DMD, patients are wheelchair-dependent by 10 years of age and often die in their late teens from respiratory failure.

Febrile seizures – any seizure that is accompanied by fever owing to a non-CNS cause in patients from 6 months to 6 years of age, fairly common and occurring in 3% of all children. A simple febrile seizure lasts less than 15 minutes and is generalized. A complex febrile seizure lasts more than 15 minutes, has focal features, or recurs within 24 hours. The diagnosis of a febrile seizure is based on history, a normal neurologic examination, and the exclusion of any CNS infection. Approximately 30% of patients with one febrile seizure will have a recurrence, but recurrence risk decreases with increasing patient age. Most first-time or occasional febrile seizures are not treated with anticonvulsants. The risk factors for development of epilepsy as a complication of febrile seizures include a positive family history of epilepsy, a prolonged or atypical febrile seizure (complex), delayed developmental milestones, a pre-existing neurologic disease, and abnormal neurologic findings.

Neonatal seizures are defined as seizures that occur during the first month after birth (principally during the first few days). The causes include: hypoxic-ischemic encephalopathy (most common cause in first three days), trauma, hemorrhage, infection (TORCH infections), cerebral dysgenesis, and metabolic disorders (amino acid defects, organic acid defects, urea cycle defects, electrolyte abnormalities). Benign familial neonatal seizures are an autosomal dominant disorder characterized by 10 to 20 seizures per day commencing around the third postnatal day that are usually self-resolving. Pyridoxine dependency leads to generalized clonic seizures soon after birth that are refractory to fosphenytoin and phenobarbital. Seizures related to this condition often commence in utero (often mistaken as fetal hiccups). Treatment involves immediate pyridoxine (vitamin B₆).

Epidural hematoma is bleeding between the inner table of the skull and the dura, most often associated with tearing of the middle meningeal artery. Following a blow to the head, there is an initial loss of consciousness, followed by a lucid interval, and followed once again by an alteration in the level of consciousness. This "waxing and waning" is typical and is associated with signs and symptoms of increased intracranial pressure. Diagnosis is by head CT which shows a lenticular density, and management is immediate surgical drainage. Prognosis is generally good if surgery can be performed rapidly. A subdural hematoma is blood beneath the dura, associated with tearing of the bridging meningeal veins by direct trauma or shaking. It is more common than epidural hematoma and is seen most commonly in infancy, with clinical features including seizures and signs and symptoms of increased ICP – symptoms develop more slowly than with an epidural bleed. Diagnosis is by head CT which shows a crescentic density, management includes neurosurgical consultation and usually surgical drainage, and prognosis may be poor if the underlying brain is also injured.

Acute spinal cord injury is most often caused by motor vehicle crashes and sports injuries in the adolescent population. Two-thirds of sports-related injuries are the result of diving accidents, often in natural bodies of water, but up to 1/3 occur in swimming pools. Axial loading or hyperflexion of the vertebral column is the usual mechanism of injury and generally there is no associated head trauma. Common presentations may include numbness in the hands and feet, paralysis below the level of the injury, and absent patellar reflexes. Treatment includes the use of high-dose methylprednisolone and immediate neurosurgical consultation.

Adrenoleukodystrophy is a progressive demyelination of the CNS associated with adrenal cortical failure. It is transmitted by X-linked inheritance and affects males. The onset typically occurs between 5-10 years of age and initially presents with an alteration in behavior ranging from a withdrawn state to aggressive outbursts. Poor school performance follows invariably and may lead parents to seek psychological services. Neurologic deterioration is then relentlessly progressive and includes disturbances of gait and coordination, loss of vision and hearing, and ultimate deterioration to a vegetative state, with seizures as a late manifestation. Physical exam will reveal hyperpigmentation and hyperreflexia of the extremities.

Benign paroxysmal vertigo is characterized by abrupt, brief episodes of vertigo with ataxia in children ages 1 to 3 years. The child may appear frightened, have pallor, and may indicate feeling dizzy. Rapid eye movements or horizontal nystagmus is characteristic, and this condition is believed to be a migraine variant.

Bell palsy is an idiopathic or postviral swelling of the facial nerve and its canal. Because this is a lower motor neuron process, one entire side of the face is affected with definite muscular weakness. Loss of tearing in one eye, hyperacusis, or alteration in taste can also occur. Recovery is spontaneous and complete in up to 90% of cases. Treatment involves minimizing corneal exposure with usage of an ocular lubricant at night, with some consideration given to oral prednisone use.



TEXAS TECH UNIVERSITY HEALTH SCIENCES CENTER
PEDIATRICS

QUESTIONS ON INFECTIOUS DISEASE AND NEUROLOGY

1. You are working in a refugee camp when a mother brings in her 8-day-old boy. The mother states he started becoming irritable 2 days ago, and now any loud noise appears to cause him pain, as evidenced by muscle tightening and back arching causing his head to nearly touch his feet. Physical examination reveals only a dried packing on his umbilical cord, as is the local custom. He appears normal until he is stimulated by touch or a loud noise, and then he begins to cry, stiffens, and arches his back. The stiffness continues until he calms down.

Of the following, the MOST likely diagnosis is

- A. bacterial meningitis
- B. botulism
- C. generalized seizure
- D. tetanus
- E. viral encephalitis

2. You are evaluating a 7-year-old boy who has sickle cell disease for a 5-day history of fever, rhinorrhea, headache, and worsening fatigue. Physical examination reveals a tired-appearing, pale boy who has a temperature of 101.5°F (38.6°C); heart rate of 150 beats/min; very pale conjunctivae and mucous membranes; and a faint diffuse, erythematous, lacy rash that is most prominent on his cheeks and trunk. Laboratory tests show a white blood cell count of $12 \times 10^3/\text{mcL}$ ($12 \times 10^9/\text{L}$), with 50% neutrophils, 45% lymphocytes, and 5% monocytes; hemoglobin of 4 g/dL (40 g/L); hematocrit of 16%; and reticulocyte count of less than 1%.

Of the following, the MOST likely cause for this patient's present illness is infection with

- A. coxsackievirus
 - B. cytomegalovirus
 - C. Epstein-Barr virus
 - D. parainfluenza virus
 - E. parvovirus B19
- NEW IMMUNE HYDROPS FETALIS, SLAPPED CHEEK, ARTHRITIS/ARTHRALGIAL
STOPS RETICULOCYTE COUNT → KILLS BABY RBC'S

3. A 16-year-old boy has had low-grade fever, dry cough, and malaise for 1 week. Medical history is unremarkable. Physical examination reveals bibasilar rales. Scattered interstitial infiltrates are seen on a chest radiograph. Results of a complete blood count are normal.

Of the following, the MOST likely cause of this boy's illness is

- A. adenovirus
- B. *Chlamydia pneumoniae* LOW GRADE FEVER
- C. *Chlamydia trachomatis*
- D. influenza virus
- E. respiratory syncytial virus

4. An 18-day-old term infant develops fever, lethargy, and focal seizures. Findings include: an ill-appearing infant with out exanthem, hepatomegaly, or jaundice. Analysis of cerebrospinal fluid reveals white blood cells, $115/\text{mm}^3$, 45% neutrophils; 55% lymphocytes; red blood cells, 40 mm^3 ; glucose, 45 mg/dL; protein 200 mg/dL; and negative Gram stain.

TEMPORAL LOBE

In addition to ampicillin and cefotaxime, the MOST appropriate treatment to begin at this time is

- * A. acyclovir
 B. amphotericin B
C. dexamethasone
D. metronidazole
E. vancomycin
5. An 8-month-old boy presents to your clinic with a 3-day history of a temperature to 103°F (39.4°C). Last night, his mother noted that he became stiff and had rhythmic motions of both arms that lasted about 20 seconds. She stated he seemed "kind of out of it" during this episode. He was febrile at the time, and she did not know if this had been seizure activity or just shivering due to his high temperature. This morning, he was without fever but had developed a rash. On physical examination, the boy is afebrile and does not appear toxic. Results of the physical examination are normal, with the exception of a maculopapular rash over his trunk and extremities.

Of the following, the MOST likely cause of this patient's illness is

- A. adenovirus
B. enterovirus
 C. human herpesvirus type 6 FEVER 3 DAY THEN RASH (ROSEOLA = HHV 6)
 D. rubella
 E. rubeola
6. During the month of July you have seen several young female adolescents whose complaints include rhinitis, cough, and scratchy throat. Almost all have had yellowish eye discharge with few other findings. Information obtained from the families indicates that almost all of the girls spent some time at the same summer camp prior to developing the symptoms.

Of the following, the MOST likely etiologic agent is

- A. adenovirus
B. *Bordetella pertussis*
C. *Chlamydia trachomatis*
D. respiratory syncytial virus
E. *Streptococcus pneumoniae*

7. A 3-month-old baby develops constipation followed by poor feeding with weak suck. After several days, she displays ptosis. On physical examination, the baby is afebrile and has loss of head control, with mild weakness of the upper extremities. Deep tendon reflexes are diminished.

After stabilization, the MOST appropriate diagnostic study to obtain is

- A. blood lead level
- B. genetic testing for spinal muscular atrophy
- C. serum assay for botulinum toxin 12-48 HOURS AFTERWARDS → DESCENDING PARALYSIS
- D. urine porphobilinogen
- E. urine screen for heavy metals

8. The MOST common clinical manifestation of an atypical mycobacterial infection in an otherwise healthy child is

- A. cervical lymphadenitis & *BARTONELLA HENSELEA*
- B. osteomyelitis
- C. pneumonia
- D. soft-tissue abscess
- E. urinary tract infection

9. Four days after attending a 2-week Boy Scout camp in southern Rhode Island, a 12-year-old boy develops fever, malaise, and a single circular red lesion.

Of the following, the MOST appropriate antibiotic for treatment is

- A. cephalexin
- B. clarithromycin
- C. dicloxacillin
- D. doxycycline LYMES DISEASE - ERYTHEMA CHROMICUM MIGRANS, BEBES PALSY, ASEPTIC MENINGITIS, HEART BLOCK, ARTHRITIS & ARTHRALGIAS
- E. trimethoprim-sulfamethoxazole

10. You are evaluating a 1-year-old boy who has a 2-day history of rhinorrhea, pharyngitis, and low-grade fever. Today he developed a barking cough with inspiratory stridor and a temperature of 101°F (38.3°C). On physical examination, he has a hoarse voice, coryza, a mildly inflamed pharynx, and a slightly increased respiratory rate (35 breaths/min).

Of the following, the MOST likely cause of his illness is

- A. adenovirus
- B. coronavirus
- C. enterovirus
- D. respiratory syncytial virus
- E. parainfluenza virus

11. A child is bitten on the hand by a neighbor's dog. Within 24 hours there is erythema, pain, and swelling at the site of the bite. The child is taken to the emergency department where cultures are taken of sanguinopurulent drainage from the wound.

Of the following, the MOST likely organism infecting the wound is

- A. *Eikenella corrodens* (HUMAN BITE)
 - B. *Francisella tularensis*
 - C. *Pasteurella multocida* QUICK FEVER, REDNESS TX: AUGMENTIN
 - D. *Staphylococcus aureus*
 - E. *Streptococcus pyogenes*
12. A 16-year-old previously healthy boy presents with a 2-week history of intermittent elevated temperatures to 102°F (39°C), headache, malaise, fatigue, myalgias, and a progressively worsening nonproductive cough. During history-taking, he reports that recently he explored several caves while hiking in a forest preserve in Ohio. Physical examination reveals a tired-appearing adolescent who has a temperature of 102.4°F (39.1°C), a dry cough, and diffuse intermittent rhonchi on chest auscultation. Laboratory findings include a white blood cell count of $12 \times 10^3/\text{mcL}$ ($12 \times 10^9/\text{L}$), with 60% neutrophils, 2% band forms, and 38% lymphocytes. Chest radiograph shows patchy left upper and left lower lobe opacities and hilar adenopathy.

Of the following, the MOST likely pathogen causing this patient's condition is

- A. *Aspergillus fumigatus*
 - B. *Coccidioides immitis*
 - C. *Histoplasma capsulatum*
 - D. *Rhizopus* sp
 - E. *Sporothrix schenckii*
13. A 17-year-old boy who had a 2-day history of fever, sore throat, headache, and malaise was treated with amoxicillin for streptococcal pharyngitis. Six days following the initiation of therapy, he returns because of persistent symptoms and a generalized eruption composed of small erythematous macules.

Of the following, the eruption is MOST likely due to

- A. erythema infectiosum
- B. infectious mononucleosis
- C. measles
- D. penicillin allergy
- E. scarlet fever

14. A 17-year-old girl presents to the emergency department with a history of being "light-headed." Upon further questioning, you discover that she has had a temperature for the preceding 2 days to 104°F (40°C) and has complained of malaise, headache, sore throat, and muscle tenderness. She decided to come to the emergency department when she started developing a rash on her face, chest and legs today. She describes the rash as looking like a sunburn. Her past medical history is unremarkable and she is on the fourth day of her menstrual cycle. Physical examination reveals an ill-appearing adolescent who has a temperature of 103°F (39.4°C), a heart rate of 132 beats/min, and a blood pressure of 70/50 mmHg. She has erythema on her face and chest, peripheral cyanosis and conjunctival hyperemia, and her major muscle groups are tender to palpation.

Of the following, the BEST initial treatment approach to this patient is

- A. antimicrobial therapy
- B. corticosteroids
- C. fluid replacement
- D. intravenous immunoglobulin
- E. plasmapheresis

15. A 3-year-old girl who has nephrotic syndrome presents with a 1-day history of fever and abdominal pain. On physical examination, you find a toxic-appearing child who has a temperature of 104°F (40°C), a heart rate of 120 beats/min, a respiratory rate of 32 breaths/min, and a blood pressure of 75/55 mmHg. Upon palpation of her abdomen, you discover ascites and diffuse tenderness with rebound. You suspect peritonitis.

Of the following, the MOST likely organism to cause this patient's condition is

- A. *Candida albicans*
 - B. *Escherichia coli*
 - C. *Proteus mirabilis*
 - D. *Staphylococcus aureus*
 - E. *Streptococcus pneumoniae*
- ↑ RISK OF THROMBOEMBOLISM
- ↑ RISK OF PERITONITIS BY STREP PNEUM
- PNEUMONIA = NEPHROTIC SYNDROME

16. Pertussis has been confirmed in a 6-week-old infant who was hospitalized for apnea and severe paroxysms of coughing. His 3-year-old brother has received his primary series of pertussis immunizations.

Of the following, the MOST appropriate management of the 3-year-old boy is to

- A. administer cephalexin
- B. administer clindamycin
- C. administer erythromycin
- D. administer penicillin
- E. observe without any antibiotic therapy

17. A 12-year-old Mexican girl whose family recent immigrated to the United States presents with a 3-day history of fever, cough, rhinorrhea, and red eyes. Findings include: temperature, 40.5°C (104.9°F); mild photophobia; profuse rhinorrhea; and a confluent maculopapular rash over the face, neck, trunk, and upper arms. The mother states that the girl's temperature increased when the rash appeared.

The MOST likely diagnosis is

- A. Kawasaki disease
 - B. measles *RUBEOLA 10 DAY MEASLES*
 - C. roseola *FEVER X 3 DAYS*
 - D. rubella
 - E. scarlet fever
18. A 9-month-old boy presents to a hospital emergency department in February with a 1-day history of fever to 38.5°C (101°F); 8 to 10 voluminous, watery stools; and several episodes of vomiting. Over the past 12 hours, he has voided only once. Physical examination reveals a lethargic boy who has dry mucous membranes and slow capillary refill.

Of the following, the MOST appropriate diagnostic study to perform on a stool sample is

- A. antigen testing for rotavirus *WINTER, β BLOOD*
 - B. culture for enteroviruses
 - C. culture for *Salmonella* species
 - D. examination for ova and parasites
 - E. testing for *Clostridium difficile*
19. A 6-year-old child presents with a 5-week history of multiple right-sided preauricular and superior cervical lymph nodes that are enlarging. Conjunctivitis of the right eye was noted at the onset of the adenopathy, but it is no longer present.

Among the following, the organism MOST likely to cause these findings is

- A. *Streptococcus pyogenes*
 - B. *Bartonella henselae* *PANICULITIS OCULI GLANDULAR SYNDROME*
 - C. *Francisella tularensis*
 - D. *Mycobacterium avium-intracellulare* *SX RENOVAL*
 - E. *Staphylococcus aureus*
20. A newborn has hepatosplenomegaly, purpuric rash, jaundice, thrombocytopenia, and microcephaly. Computed tomography of the head demonstrates periventricular cerebral calcifications.

Of the following, the MOST appropriate diagnostic testing for this infant includes

- A. maternal human immunodeficiency virus serology
- B. urine culture for CMV
- C. serologic testing of mother and infant for *Toxoplasma*
- D. VDRL on infant and maternal sera
- E. Viral culture of swabs of infant's throat and conjunctivae

CMV vs Toxo vs RUBEOLA
 ↓
 DIFFUSE CALCIFICATION
 PERIVENTRICULAR CALCIFICATION

21. Early in June, a 7-year-old girl is brought to your office for evaluation of several days of high fever, headache, and one day of rash. Her arms and legs are covered with purpuric macules and papules that have begun to spread to her trunk. You obtain appropriate cultures and diagnostic studies and admit her to the hospital.

Of the following, the MOST likely diagnosis to explain these findings is

- A. coxsackie A virus infection
- B. gonococcal sepsis
- C. Henoch-Schönlein purpura
- D. leptospirosis
- E. Rocky Mountain spotted fever ARMS TO TRUNK, HYPONaTREMIC, THROMBOCYTOPENIA
Tx: DOXYCYCLINE

22. In July, a fully immunized 13-year-old girl who has not traveled out of her hometown in Colorado during the past year awakens with a generalized tonic-clonic seizure. She has fever, a few loose stools, and a faint macular rash. Evaluation of the cerebrospinal fluid suggests viral meningoencephalitis.

Of the following, the MOST likely etiology of this girl's findings is

- A. enterovirus
- B. herpesvirus type 2
- C. *Mycoplasma pneumoniae*
- D. Rubeola
- E. Western equine encephalitis

- NEURO
23. The parents of a 4-year-old girl report that several nights each week she comes into their bedroom. While standing next to their bed, she exhibits right facial jerking and difficulty swallowing and talking. A cousin has absence seizures.

Of the following, the MOST likely diagnosis is

- A. absence seizures
- B. benign rolandic epilepsy BENIGN CENTROTEMPORAL EPILEPSY - PARTIAL, MIDDLE OF NIGHT, 3-13yo
GOES AWAY BY ITSELF
- C. conversion reaction
- D. night terrors
- E. panic attacks

24. A 22-month-old girl is nonverbal. She sat alone at 7 months and walked by 13 months, but now exhibits a wide-based stance, no longer ambulates, and will not pick up or manipulate toys. Findings include: height and weight at the 50th percentile; head circumference below the 5th percentile, with no increase over the past 8 months; normal fundi; and no organomegaly.

Of the following, the MOST likely diagnosis is

- A. adrenoleukodystrophy
- B. cerebral palsy
- C. GM₂ gangliosidosis (Tay-Sachs disease)
- D. hypothyroidism
- E. Rett syndrome

25. A 9-year-old boy is referred for evaluation of seizures that began at 2 years of age and mental retardation. His parents also report that he has developed aggressive behavior toward his siblings. Physical examination reveals several hypopigmented macules on the trunk.

Of the following, the additional physical finding that will be MOST helpful in confirming diagnosis is

- A. adenoma sebaceum INFANTILE SPASMS (ALSO IN PREBIRTHAL EID)
- B. café au lait spots
- C. linear epidermal nevus on scalp
- D. multiple lentigines
- E. nevus flammeus on the face
26. A 6-year-old boy is brought in for evaluation because his family is worried that his gait has become "clumsy." Physical examination reveals a bright boy who has normal deep tendon reflexes but who has difficulty rising from the floor. He needs to support his trunk by "climbing up" his lower limbs to assume an erect stance.

Of the following, these findings are MOST suggestive of

- A. Duchenne muscular dystrophy
- B. Guillain-Barré syndrome
- C. myasthenia gravis
- D. myotonic dystrophy
- E. transverse myelitis
27. A father brings his 6-year-old daughter to you because her teacher has observed multiple, daily episodes in which the child stares and is unresponsive to verbal cues. The teacher also has noted facial twitching with some of these several-second events. Results of physical examination of the girl are normal.

Among the following, the MOST appropriate medication for this child is

- A. atomoxetine
- B. carbamazepine
- C. clonidine
- D. ethosuximide
- E. methylphenidate
28. A 6-year-old boy presents with a 5-day history of worsening ataxia following an episode of gastroenteritis 1 week ago. On physical examination, the child has mild facial diparesis and areflexia. His gait is ataxic, and the child has weakness of the ankle flexor and extensor muscles.

Of the following, the test MOST likely to lead to the correct diagnosis is

- A. computed tomography of the head
- B. lumbar puncture BREAK DOWN OF MYELIN \Rightarrow \uparrow PROTEIN
- C. measurement of serum vitamin E level
- D. magnetic resonance imaging of the spine
- E. urine toxicology screen

29. You are evaluating a 15-year-old girl who has had fever, lethargy, confusion, and unremitting headaches for 3 days. Examination of CSF after an atraumatic lumbar puncture reveals 130 leukocytes/mm³, 420 red blood cells/mm³, and a negative Gram stain. MRI shows increased signal in the left temporal lobe. EEG documents focal discharges.

Of the following, the MOST likely cause of encephalitis in this patient is

- A. enterovirus
 - B. herpes simplex virus type 1
 - C. human immunodeficiency virus
 - D. postinfectious encephalomyelitis
 - E. rubeola
30. A 12-year-old child is brought to the emergency department after diving head first into a shallow pool. He reports severe neck pain, paresthesias in the hands, and numbness in the feet. He has no volitional movement in his legs, and his patellar reflexes are absent. His blood pressure is 110/70 mmHg. The nearest neurosurgeon is 2 hours away by ambulance, and you arrange for transfer.

Prior to transfer, the MOST appropriate treatment is administration of

- A. a saline infusion of 20 mL/kg
 - B. ceftriaxone
 - C. high-dose methylprednisolone
 - D. fosphenytoin
 - E. ranitidine
31. A term newborn experiences generalized tonic-clonic seizures 1 hour after birth. The vaginal delivery was unremarkable, and Apgar scores were 9 at both 1 and 5 minutes. The pregnancy was notable for the mother reporting that the "baby had hiccups." Except for the ongoing seizures, physical examination results are normal, and the baby boy is afebrile. Despite full dosing of intravenous fosphenytoin and phenobarbital, the seizures continue.

Of the following, the MOST likely cause of the child's seizures is

- A. Aicardi syndrome
- B. benign familial neonatal seizures
- C. hypoxic-ischemic encephalopathy
- D. nonketotic hyperglycinemia
- E. pyridoxine dependency *B6 - given some*

32. A 10-year-old boy presents after falling from a second floor window onto the pavement 30 minutes ago. There was a 3-minute loss of consciousness initially, but he has been alert and talking to his mother during the ambulance ride to the hospital. On physical examination, the child has tenderness and a hematoma on the right parietal region of his head. Results of an initial complete neurologic examination are normal, but on subsequent examination 15 minutes later, the boy exhibits marked lethargy and slurred speech.

Of the following, the MOST likely explanation for his current symptoms is

- A. cerebral contusion
- B. concussion
- C. epidural hematoma
- D. subarachnoid hematoma
- E. subdural hematoma

33. A 14-year-old girl has experienced brief, shock-like jerks of her arms and has sometimes inadvertently thrown objects in her hand. This morning she had a generalized tonic-clonic seizure after awakening. Subsequent electroencephalography shows 4 to 5 cycle per second generalized spike and wave discharges with normal background activity.

The statement you are MOST likely to include in your discussion with the family is that

- A. absence epilepsy frequently resolves after 1 year
 - B. ketogenic diet is the treatment of choice
 - C. lifelong treatment with valproic acid is likely necessary
 - D. no treatment is indicated
 - E. rolandic seizures never should be treated with carbamazepine
 - F. REFER TO NEURO
- JUVENILE MYOCLONIC EPILEPSY - WHICH GOES AWAY

34. A 7-year-old girl has had weakness of the right side of her face for 4 days following a systemic viral infection 2 weeks ago. She denies any hearing difficulty or hyperacusis and claims that tearing and taste are normal. Physical examination reveals weakness of the upper and lower face. Deep tendon reflexes are normal.

Of the following, the MOST appropriate next step is

- A. application of an ocular lubricant at night
- B. initiation of amoxicillin
- C. magnetic resonance imaging of the brain
- D. nerve conduction velocities and electromyography
- E. prescription of oral acyclovir

35. A 15-year-old obese girl who has cystic acne has experienced a frontal headache for 1 week. She reports that the only drug she takes is isotretinoin. Last night she presented to the emergency department for headache. Computed tomography of the head was obtained and was normal; she was given meperidine and discharged home. She presents now to your office for follow-up. The girl has papilledema, but her physical examination findings are otherwise normal.

Of the following, the MOST appropriate next step in the evaluation of this patient is

- A → A. lumbar puncture *HIGH OPENING PRESSURE*
 B. magnetic resonance imaging of the brain with gadolinium contrast *PSEUDOTUMOR CEREBRI*
C. neurosurgery consultation
D. ophthalmology consultation
E. urine toxicology screen

36. A 7-year-old girl who has incompletely repaired tetralogy of Fallot complains of frontal headache and vomiting without diarrhea for 2 days. On physical examination, she has a temperature of 102.3°F (39.1°C) and appears lethargic. She does not have papilledema, but has some difficulty finding words to name objects.

Of the following, the MOST appropriate next step in the evaluation of this child is

- A. computed tomography of the head with contrast
B. echocardiography
C. electroencephalography
D. lumbar puncture
E. stool culture for *Shigella*

37. A 2-year-old girl is rushed by ambulance to the emergency department for sudden-onset ataxia. Her parents have yet to arrive. On physical examination, the girl is afebrile, yet diaphoretic, with some nystagmus on far lateral gaze. Her ataxia has resolved. The remainder of physical examination findings are normal.

Of the following, the MOST likely diagnosis for this child is

- A. basilar migraine
 B. benign paroxysmal vertigo
C. cerebellar hemorrhage
D. phenytoin intoxication
 E. seizure

38. You have been treating a 2-year-old girl for pneumococcal meningitis for the past 5 days.

Of the following, the MOST likely complication of her disease is

- A. brain abscess
B. cerebral infarct
C. cranial nerve palsy
 D. hearing impairment
E. sagittal sinus thrombosis

39. A 3-year-old boy is brought to your office with his eyes fixed in an upward gaze. He is alert and frightened, but cooperative. Vital signs are normal. You suspect a drug reaction.

While awaiting the results of a toxin screen, these findings are MOST likely to be alleviated by the administration of

- A. charcoal by nasogastric tube
 - B. diazepam intramuscularly
 - C. diphenhydramine intravenously
 - D. syrup of ipecac by mouth
 - E. promethazine rectally
40. A 3-month-old infant has had hypotonia and motor delay since birth. The delivery was complicated by shoulder dystocia. Apgar scores were 3 and 7 at 1 and 5 minutes, respectively. The infant required bag and mask ventilatory support for a short time in the delivery room.

Of the following, the finding that would be MOST suggestive of a lower motor neuron cause for this infant's findings is

- A. a persistent palmar grasp
 - B. absent deep tendon reflexes
 - C. an aggravated supporting-standing response
 - D. persistent tonic neck reflex
 - E. positive ankle clonus
41. A 4-month-old girl presents with a 2-day history of a temperature of 103°F (39.4°C), irritability, decreased oral intake, and vomiting. She has not received any of her immunizations. On physical examination, she appears lethargic and has a bulging fontanelle. You suspect meningitis and perform a lumbar puncture (LP). Results of the LP are: white blood cell count, 800/mm³ (88% neutrophils, 12% monocytes); red blood cell count, 2/mm³; glucose, 15 mg/dL (0.83 mmol/L); and protein, 150 mg/dL.

Of the following, the MOST likely cause for this patient's meningitis is

- A. enteroviral
 - B. herpetic
 - C. listerial
 - D. pneumococcal
 - E. tubercular
42. A 9-month-old girl has a generalized febrile seizure associated with symptoms of an upper respiratory tract infection and a temperature of 39°C (102.2°F).

Of the following, the factor that is MOST likely to increase her chance of epilepsy (recurrent nonfebrile seizures) is having

- A. a febrile seizure after 2 years of age
- B. a rash consistent with roseola
- C. a seizure that lasts 10 minutes *normal < 15 min*
- D. cerebral palsy
- E. three febrile seizures in 1 year

43. A 5-year-old boy acutely refuses to walk and complains of back pain. On physical examination, he is febrile, has exaggerated reflexes and flaccidity of the legs, and a distended bladder. Sensory examination is difficult to interpret. Radiography of the spine is normal.

Of the following, the MOST likely cause of these findings is

- A. vertebral diskitis
- B. Guillain-Barré syndrome
- C. sacroiliitis
- D. transverse myelitis *Demyelination of spinal cord @ certain level*
- E. vertebral osteomyelitis

44. You are conducting a health supervision visit for a 5-month-old infant who had been in the neonatal intensive care unit. She feeds well and is thriving. The baby was born at 29 weeks' gestation, and head ultrasonography at 6 weeks of age had documented periventricular leukomalacia. Physical examination reveals brisk reflexes, ankle clonus, and bilateral Babinski sign.

Of the following, the type of cerebral palsy for which this infant is MOST at risk is

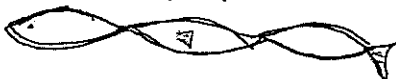
- A. ataxic
- B. athetoid
- C. spastic diplegic
- D. spastic hemiplegic
- E. spastic quadriplegic

45. A mother brings her 5-month-old daughter to your clinic for evaluation of colic. While interviewing the parent, you observe the child repeatedly flex her upper and lower extremities all at once. The child is developmentally abnormal.

The most likely diagnosis is

- A. colic
- B. gastroesophageal reflux
- C. infantile spasm
- D. intussusception
- E. constipation

245 997920



① ~~RTA~~ ORGANIC ACIDEMIA (ANION GAP)

OMIM ONLINE MENDELIAN INHERITANCE IN MAN

124R6W260369100107

FACTS – GROWTH, DEVELOPMENT, AND IMMUNIZATIONS

An infant typically doubles his birthweight by 5 months and triples his birthweight by 1 year of age. An infant increases his length 50% by 1 year of age, doubles his length by 4 years, and triples it by age 13. Most 4-year-old children are about 40 inches tall and 40 lb. After losing 5-10% of birthweight in the first few days of life, an infant should gain about 1 oz (30 grams) per day in the first few months of life. From early toddlerhood through the pre-adolescent age, the normal annual growth velocity will be at least 5 cm (2 inches) per year.

Growth failure can be the result of several etiologies:

- Children who have nutritional causes of poor growth (inadequate caloric intake or malabsorption) usually present with decreases in weight initially, with subsequent fall-off in height velocity if poor nutrition persists.
- Children who have endocrine disorders (hypothyroidism, Cushing disease, growth hormone deficiency) typically present with isolated short stature because height is affected most and weight is spared.
- Children who have chromosomal abnormalities usually exhibit growth retardation in head circumference as well as length and weight, with developmental delays and other physical abnormalities.

Macrocephaly means a large head and is defined as a head circumference that is 2 standard deviations above the mean. Signs of increased intracranial pressure suggesting hydrocephalus or an intracranial mass include irritability or somnolence, loss of appetite, vomiting, a bulging fontanelle, strabismus, impairment of upward gaze, and increased tendon reflexes and hypertonicity. Progressive hydrocephalus with increased intracranial pressure is the most common pathologic cause of a rapidly enlarging head. However, if there are no signs of increased intracranial pressure, the child's development is normal, and measurement of the parent's head circumference is large, the infant's large head usually is a result of benign familial macrocephaly.

Microcephaly is defined as a head circumference that measures more than 3 standard deviations below the mean for age and gender, most often the result of a small brain, as the skull generally grows in response to brain growth. Microcephaly can be categorized as primary (genetic – usually have small head circumferences at birth) or secondary (acquired – bacterial meningitis would be one such case).

Developmental milestones are extremely important because they reflect neurologic maturation and social and sensory development in the growing child. Development, with regard to gross motor, fine motor, social, and speech, progresses sequentially in the healthy child:

- 2 months – smiles socially, coos, follows object past midline
- 4 months – holds head erect with no head lag, raises body using arms from prone position, may start rolling prone to supine, brings hands to midline, grabs a rattle, tracks and follows objects visually to 180 degrees, can vocalize through differentiated crying, laughs
- 6 months – sits with or without support, reaches for objects, transfers objects from hand to hand, rolls bilaterally, tries to obtain small objects with a raking movement, babbles
- 9 months – crawls, pulls to stand, cruises, immaturely picks up small objects using a thumb and index finger, feeds self with finger foods, searches for hidden objects (object constancy), stranger anxiety, enjoys social games like peek-a-boo and pat-a-cake, responds to own name, waves bye-bye, nonspecific ma-ma and da-da.
- 12 months - walks alone, uses mature pincer grasp, follows a single command with a gesture, drinks from a cup with assistance, cooperates with dressing, uses one to two words other than specific da-da/ma-ma.
- 15 months - builds tower of two blocks, scribbles with crayons, walks backwards, follows a one-step command without a gesture, uses four to six words
- 18 months - runs, walks upstairs with one hand held, climbs up onto an adult chair, eats with a spoon, can stack three to four blocks, copies parent in tasks, points to body parts on command, uses 10 to 20 words
- 2 years - walks up and down steps without help, runs fast, jumps, kicks a ball, throws a ball with overhand motion, removes clothes, draws a vertical line, builds a tower of six to eight blocks, uses greater than 50 word vocabulary and speaks in two-word sentences, uses personal pronouns like “I, you, me”

Development proceeds in a cephalic to caudal as well as a proximal to distal direction. The final goal of every step of developmental progression is a move from dependence to independence. It is important to correct for prematurity when assessing the growth and development of children who are younger than age 2.

Shapes – a 2-year-old can copy a vertical line. A 3-year-old can copy a circle. A 4-year-old can copy a cross. A 4-1/2-year-old can copy a square. A 5-year-old can copy a triangle. A 6-year-old can copy a diamond.

Immunizations are one of the most important components of well child care and are the cornerstone of pediatric preventative care. There are two types of immunizations – active and passive.

- Active immunization involves induction of long-term immunity through exposure to live attenuated or killed (inactivated) infectious agents.
 - Live vaccines are more likely to induce long-lasting immunity, but carry the risk of vaccine-associated disease. As a result, these should generally be avoided in patients with compromised immunity (cancer patients, HIV, primary immunodeficiencies). Examples of live vaccines include MMR, varicella, rotavirus, OPV, yellow fever, smallpox, oral typhoid, and TB (BCG vaccine).
 - Non-live vaccines are not infectious and tend to induce immunity for shorter periods, thus requiring booster immunizations. Examples include DTaP, hepatitis A and B, IPV, Hib, influenza, pneumococcal, meningococcal.
- Passive immunization involves delivery of preformed antibodies to individuals who have no active immunity against a particular disease but who have either been exposed to or are at high risk for exposure to the infectious agent. Examples include:

Varicella zoster immune globulin (VZIG) – should be given to immunocompromised children within 96 hours of exposure.

Immunocompromised is defined as patients receiving chemotherapy, HIV, other primary immunodeficiencies, organ transplant patients, newborns of mother who had the onset of chickenpox within the five days prior to or 48 hours after delivery, premature NICU babies, or susceptible pregnant women. Significant exposures would include household contact, face-to-face indoor play, close hospital exposures, or onset of varicella in the mother of a newborn from five days before to two days after delivery.

Hepatitis B immune globulin (HBIG) – should be given to all infants born to mothers who are hepatitis B surface antigen positive, in addition to hepatitis B vaccine, because the incidence of chronic hepatitis B infection occurs in as many as 90% of infants infected perinatally.

Hepatitis A immune globulin – all previously unimmunized persons with close personal contact with a hepatitis A case, such as household and sexual contacts, should receive hepatitis A IG within two weeks after last exposure. When HAV infection is identified in an employee or child enrolled in a child-care center in which all children are toilet-trained, immunoglobulin is recommended for previously unimmunized employees in contact with the index case and for unimmunized children in the same room as the index case. When the infection is identified in an employee or child in a child-care center in which children are not toilet-trained, immunoglobulin is recommended for all previously unimmunized employees and children in the facility. School-room exposure generally does not pose an appreciable risk of infection and IG administration is not indicated when a single case occurs.

- Rabies immune globulin – because rabies virus causes an acute, fatal, encephalomyelitis in humans, any bite from a wild animal (bats, raccoons, skunks, foxes, coyotes) should be followed by immediate postexposure prophylaxis with HRIG in conjunction with five doses of rabies vaccine.
- Tetanus immune globulin – recommended only for contaminated wounds (dirt, feces, soil, saliva) in patients who have received fewer than three doses of tetanus toxoid or in whom immunization status is unknown, or in any tetanus-prone wound in a patient with HIV (*Clostridium tetani* is a spore-forming, anaerobic, gram-positive rod that is found worldwide in soil and human and animal feces. It produces a potent exotoxin that causes severe generalized muscle spasms. Because immunization with tetanus toxoid does not confer lifelong immunity, booster doses are recommended at 10-year intervals. The need for additional booster doses of tetanus is determined by the nature of the wound and the patient's prior immunization history. Those who have received at least three doses of tetanus toxoid, with the last being within the preceding 10 years, do not require additional protection for a clean, minor wound. For wounds that are contaminated, however, an additional dose of tetanus toxoid should be administered unless the last immunization was within 5 years.)
- RSV immune globulin – used for immunoprophylaxis in infants and children less than 2 years of age with chronic lung disease who have required medical therapy (oxygen, diuretics, steroids, bronchodilators) within the six months before the RSV season, especially useful in premature babies.
- Measles immune globulin – can prevent or modify the disease if given within six days of exposure to susceptible household contacts, pregnant women, children less than 1 year of age, and immunocompromised individuals.

Postexposure chemoprophylaxis - can help prevent the spread of certain contagious diseases:

- Pertussis – recommended for all households and other close contacts, including child-care contacts, with oral Erythromycin for 14 days. Physicians who prescribe Erythromycin to newborns should inform the parents about the potential risk of developing pyloric stenosis.
- Meningococcus – close contacts of all persons who have invasive meningococcal disease, whether sporadic or in an outbreak, are at high risk for infection and should receive chemoprophylaxis within 24 hours of the diagnosis, regardless of vaccine status. Close contact include all household contacts, child-care and nursery-school contacts during the previous seven days, persons who have had direct contact with the patient's oral secretions, and persons who frequently eat or sleep in the same dwelling as the index patient. However, classroom contacts of students who have meningococcal disease are considered casual contacts and the use of prophylactic antibiotics is not recommended. Treatment includes Rifampin (drug of choice), Ciprofloxacin, and/or Ceftriaxone.

Haemophilus influenzae – chemoprophylaxis with Rifampin is recommended for all household contacts with at least one contact younger than 4 years of age who is unimmunized or incompletely immunized, all members of a household if a child is younger than 12 months of age, all occupants of a household with an immunocompromised child, and nursery and child-care contacts irrespective of age when two or more cases of invasive disease have occurred within 60 days.

Influenza – chemoprophylaxis for influenza A and B for high-risk children (asthma, heart disease, HIV, renal disease) likely to be exposed to individuals infected with influenza, unimmunized individuals in close contact with or providing care to high-risk individuals, immunodeficient individuals, and individuals at high risk with contraindication to the flu vaccine.

Immunizations

Live viral vaccines can be given simultaneously at different sites, otherwise >1 month apart

MMR causes transient anergy to TB protein, so can't trust PPD for 2 months after MMR

Severely immunocompromised patients should not receive live viral vaccines

Contraindications: DTaP (encephalopathy within 7 days of previous dose, precaution with seizure disorder), MMR or Varicella (pregnancy or severely immunocompromised), Influenza (history of anaphylaxis to eggs), IPV/MMR/Varicella (history of anaphylaxis to Neomycin), for all vaccines (moderate or severe illness regardless of fever)

IVIG – if given, should delay live viral vaccines by 6-12 months

MMR and Varicella (live vaccines) are given at the 1 year visit

MMR can cause a high fever and rash 1 to 2 weeks after the shot

TEXAS TECH UNIVERSITY HEALTH SCIENCES CENTER
PEDIATRICS

QUESTIONS ON GROWTH, DEVELOPMENT, IMMUNIZATIONS
AND PREVENTATIVE PEDIATRICS

1. You are seeing a 4-year-old boy, who is new to your practice, for a preschool evaluation. His birth records reveal that his weight, length, and head circumference were all at the 50th percentile. He now weighs 18 kg (10th percentile) and has a height of 85 cm (<5th percentile). Findings on physical examination are normal.

The MOST likely cause of this boy's poor growth is

- A. a chromosomal abnormality
- B. an endocrine disorder → HEIGHT MORE THAN WEIGHT
- C. inadequate caloric intake
- D. increased fluid intake
- E. intrauterine growth retardation

2. The head size of a 6-month-old boy is at the 95th percentile. At birth, his head size was at the 50th percentile. His overall development has been normal.

Of the following, the MOST helpful method for distinguishing between benign macrocephaly and hydrocephaly is to

- A. examine the fundi
- B. measure fontanelle size
- C. measure the parents' head size
- D. obtain plain radiography of the skull
- E. plot the head circumference on a growth chart

35 cm @ Birth
45 @ 1 yr
55 @ ADULT

3. In deciding if a child's growth is normal, important information can be obtained by plotting the rate of growth (growth velocity) on a growth curve chart.

In the prepubertal boy, the average growth rate (in cm/y) is CLOSEST to

- A. 2
- B. 5
- C. 8
- D. 11
- E. 14

4. A 15-month-old girl who was placed in foster care recently has had poor growth over the past 6 months. Her weight is markedly below the 5th percentile for her age and has plateaued since the age of 9 months. Length and head circumference are at the 25th and 50th percentiles, respectively. Development is normal, although she still uses a bottle rather than a cup.

The findings in this patient are MOST likely due to

- A. a chromosomal abnormality
B. an endocrine disorder
C. an intrauterine insult
 D. Constitutional growth failure (Delayed puberty)
E. inadequate intake of calories
- ↓ weight, bottle, foster care
5. You are examining a 4-month-old boy who is brought in by his foster mother for his first health supervision visit. His weight, length, and head circumference were at the 50th percentile at birth. You now note a decrease in the head circumference from the 50th to the 10th percentile, although his weight and height remain at the 50th percentile. The infant was delivered at term. The mother reports that the boy had an infection at 3 weeks of age.
- AM BABY 2 mo u fever gets LP

Of the following, the MOST likely cause of his microcephaly is

- A. familial microcephaly
~~B. maternal diabetes~~
~~C. maternal hypertension~~
 D. meningitis
~~E. poor nutrition~~
6. You are examining a girl at her 1-year health supervision visit. Her weight, length, and head circumference all were at the 10th percentile at birth. There were no pregnancy, labor, delivery, or nursery complications. Physical examination reveals her weight, length, and head circumference are at the 5th percentile.

Of the following, this child's growth parameters MOST likely represent

- A. a chromosomal abnormality
B. a malabsorptive disorder
 C. an endocrine disorder
~~D. inadequate caloric intake~~
 E. normal growth

7. A 12-year-old girl is at the 5th percentile for height. She is an otherwise healthy child. The bone age is greater than two standard deviations below the chronologic age.

Of the following, the MOST likely cause of this patient's short stature is

- A. constitutional growth delay *MOST LIKELY*
- B. genetic short stature
- C. growth hormone deficiency (*MIDLINE DEFECT*) (*ANTERIOR PITUITARY*) (*CENTRAL TESTIS*)
- D. hypothyroidism
- E. ulcerative colitis

8. During a routine health supervision visit, you pull an infant to a sitting position. She has no head lag and maintains the sitting position with her arms propped forward on the table. She is able to reach for the otoscope and transfers objects from hand to hand.

Of the following, these motor skills are MOST likely to emerge at age

- A. 4 months
- B. 6 months
- C. 8 months
- D. 10 months
- E. 12 months

9. Two months ago an infant girl began to smile in response to her mother. She now laughs out loud and initiates social interaction.

Of the following, her present age MOST likely is

- A. 2 months
- B. 4 months
- C. 6 months
- D. 8 months
- E. 10 months

10. A boy speaks six specific words in addition to "mama" and "dada." He is able to follow one-step commands without a gesture.

His age is CLOSEST to

- A. 12 months
- B. 15 months *✓*
- C. 18 months
- D. 21 months
- E. 24 months

11. A healthy 2-month-old infant was born at 32 weeks' gestation. She has grown well since birth. On physical examination of this infant, the MOST likely finding is

- A. ability to fixate on a face and follow it briefly
- B. ability to reach and grasp a rattle
- C. ability to watch an object and follow it to midline
- D. absence of the Moro reflex
- E. babbling and cooing vocalizations

12. You are precepting a group of medical students during a structured observation at a local child care center.

Of the following motor milestones, the one that is MOST typical of a 24-month-old child is

- A. building a tower of two cubes
- B. copying a circle
- C. scribbling
- D. throwing a ball overhand
- E. walking backwards

13. You see a 9-month-old girl for a health supervision examination. She has been healthy, and growth parameters are at the 50th percentile. She can cruise, feed herself with her fingers, and recognize her own name.

Of the following, she MOST likely also can

- A. build a tower of two cubes
- B. follow a command with a gesture
- C. independently place her arms in a shirt when she is being dressed
- D. release a cube into a cup
- E. say "mama" and "dada" as sounds

14. In answer to your questions during a health supervision examination, a mother reports that her son dresses himself and brushes his teeth without help. When you ask him, he copies a circle and cross, draws a simple figure of a person, walks up and down steps, hops on one foot, and balances on one foot for 3 seconds.

This boy's developmental age is CLOSEST to

- A. 3 years
- B. 4 years
- C. 5 years
- D. 6 years
- E. 7 years

15. You see an 18-month-old girl for a health supervision examination. She has been healthy, and her growth parameters are at the 50th percentile. On physical examination, she stacks a tower of two cubes, runs, and walks up stairs with her hand held.

Of the following she MOST likely also can

- A. copy a straight line
- B. feed herself with a spoon
- C. identify pictures in a book
- D. jump off the floor with both feet
- E. wash her own hands

16. Of the following fine motor milestones, the one that is MOST characteristic of a 12-month-old child is the ability to

- A. build a tower of four blocks
- B. release a raisin into a bottle
- C. scribble spontaneously
- D. transfer objects between hands
- E. use a neat pincer grasp to attain a raisin or pellet

17. A 12-year-old boy cut his leg on an old ax that was in a storage shed. Review of his medical record reveals that he received five doses of diphtheria and tetanus toxoids and whole-cell pertussis vaccine absorbed (DTP); the last dose was administered when he was 4 years old. You clean and disinfect the wound.

Of the following, your BEST management would be to administer

- A. DTP
- B. DTP and tetanus immune globulin (TIG)
- C. adult tetanus toxoid and diphtheria toxoid (Td)
- D. Td and TIG
- E. No additional doses of vaccine or immune globulin

18. A child care worker responsible for the infants' room presents with a 3-day history of fever, jaundice, and nausea. She is diagnosed as having hepatitis A virus (HAV) infection. No one else in the child care center appears ill.

Of the following, the infection control measure that would be MOST helpful in stopping the spread of the HAV in this situation is

- A. administration of immune globulin (IG) to all children and workers in the child care center
- B. administration of IG to all children and workers who had direct contact with the index case
- C. careful handwashing for all workers
- D. no hiring of new employees or acceptance of new children for 1 month
- E. serologic testing of all workers and administration of IG to those who are seronegative

19. An 8-year-old boy was bitten 5 days ago by an unknown cat in the woods. The Health Department cannot locate the cat. His parents are concerned about administering rabies shots to him because he has a history of severe egg allergies. His bite wound has scabbed over and does not appear infected.

Among the following, the MOST appropriate management of this patient is to administer

- A. human diploid cell rabies vaccine (HDCV)
- B. no postexposure prophylaxis because of egg allergies
- C. Rabies immune globulin (RIG) and if skin testing is negative, administer HDCV
- D. RIG and HDCV
- E. Rabies vaccine absorbed (RVA)

20. A pediatric resident has a persistent cough of several weeks' duration accompanied by a 5-lb weight loss and night sweats. A Mantoux test containing 5 tuberculin units of a purified protein derivative is placed and results in 15 mm of induration. A chest radiograph reveals a left upper lobe infiltrate. Appropriate antituberculous therapy is initiated.

The MOST important criterion for determining when the resident can return to work is when

- A. a chest radiograph shows no evidence of disease
- B. a single sputum culture is negative for *Mycobacterium tuberculosis*
- C. 6 months of antituberculous therapy has been completed
- D. The cough has resolved
- E. Three consecutive sputum smears for acid-fast bacilli are negative

21. A 5-year-old child who has acute lymphoblastic leukemia and is receiving chemotherapy was exposed to varicella 2 days ago. He has no history of prior varicella infection or vaccination.

Of the following, the MOST appropriate management is to prescribe

- A. acyclovir intravenously
- B. acyclovir orally
- C. immunoglobulin G intravenously
- D. varicella vaccine
- E. zoster immune globulin intramuscularly

22. You are assessing a newborn for discharge from the nursery and are considering the infant's risk of hepatitis B infection.

Of the following, the situation that places the infant at HIGHEST risk of acquiring chronic hepatitis B infection is

- A. having a clotting disorder
- B. having a mother who is positive for hepatitis B e antigen
- C. living in an institution for the developmentally disabled
- D. living in the household of a hepatitis B carrier
- E. living with grandparents who emigrated from an area highly endemic for hepatitis B

23. Of the following children, the one for whom intravenous immune globulin is MOST indicated is a

- A. 2-month-old who has pertussis
- B. 3-year-old who has Kawasaki disease
- C. 4-year-old who is traveling to India
- D. 5-year-old who has asymptomatic human immunodeficiency virus infection
- E. 9-year-old who has arthritis and Lyme disease

24. Your colleague recently was prescribed isoniazid therapy due to a tuberculin skin test conversion accompanied by negative results on chest radiography.

Of the following, the BEST policy for the clinic is to

- A. allow your colleague to work with no restrictions
- B. inform all patients of your colleague's infection
- C. initiate isoniazid therapy in all office contacts
- D. prohibit your colleague from providing patient care for 3 months
- E. require your colleague to wear a mask for the first 2 weeks of therapy while providing patient care

25. The mother of a child who is infected with human immunodeficiency virus (HIV) would like to enroll her child in a local child care center.

Of the following, the circumstance that is MOST likely to exclude the child who has HIV infection from attending a child care center is

- A. a child who exhibits aggressive behavior such as biting and scratching
- B. a child who has a history of occasional nose bleeds
- C. a child who is not yet toilet trained
- D. no circumstances of exclusion
- E. the parent(s) or guardian who does not want to disclose the HIV status of the child

26. You are speaking to the mother of a child who attends a junior high school where one of the students was diagnosed with meningococcal disease 24 hours ago. Her child does not have any classes with the index patient and, except for passing him in the hall during lunch 3 days ago, has had no other contact with the patient. The child's mother is frantic because the school sent home a notice asking parents to bring their children to the public health department or their private physician to receive antibiotic prophylaxis.

Of the following, the MOST appropriate advice for this parent is that her child

- 7
- A. does not require antibiotic prophylaxis and does not need to be seen
 - B. does not require antibiotic prophylaxis but needs to be evaluated to determine if she is developing symptoms of meningococcal disease
 - C. needs to be seen to obtain nasopharyngeal cultures for meningococcal organisms and if the cultures are positive, may require antibiotic prophylaxis
 - D. requires antibiotic prophylaxis and should be seen immediately
 - E. should be seen immediately to determine if she needs to be hospitalized and treated for possible meningococcal disease

27. For all otherwise healthy children, you would MOST likely recommend the use of prophylactic antibiotics for

- A. child care contacts of a child who has an invasive *Haemophilus influenzae* type b infection
- B. child care contacts of a child who has pneumococcal meningitis
- C. child care contacts of an infant who has pertussis
- D. classroom contacts of a child who has scarlet fever
- E. classroom contacts of an adolescent who has meningococcal meningitis

28. A 1-year-old boy who acquired human immunodeficiency virus infection perinatally presents for a routine health supervision visit. He receives highly active antiretroviral therapy with three drugs, and he is currently asymptomatic. However, he has a history of treatment for *Pneumocystis carinii* pneumonia.

Of the following, the vaccine that is CONTRAINDICATED for this boy is

- A. conjugated *Haemophilus influenzae* type b
- B. diphtheria-tetanus toxoids with acellular pertussis
- C. injectable poliovirus
- D. split-virus influenza
- E. varicella

29. As part of the examination of a 5-year-old girl during a health supervision visit, you review her medical history.

Of the following, the condition that is the STRONGEST indication for routine annual influenza immunization is

- A. all healthy children
- B. asthma
- C. asymptomatic heart disease
- D. attendance at child care
- E. frequent otitis media

30. A 3-year-old child who has a history of recurrent otitis media with effusion (OME) in infancy is brought to the clinic. His mother is afraid that he has a hearing loss because he does not talk as much as his brother did at the same age. He speaks in three-word sentences, and you can understand fewer than 50% of his words. Results of his physical examination including the ears are normal.

Of the following, the MOST appropriate statement regarding this child's condition is that

- A. even mild conductive hearing loss could affect his later school performance without frank speech delay
 - B. OME does not cause conductive hearing loss severe enough to cause speech delay
 - C. performing hearing screening solely in response to parental concern is not recommended
 - D. testing air and bone conduction thresholds in the office will help you rule out hearing loss
 - E. the absence of middle ear fluid rules out conductive hearing loss
31. During a health supervision visit of a 6-year-old child, you ask the mother if there are any guns in the home. She states that her husband is a hunter, but he keeps his shotgun in his pickup truck.

Of the following, the BEST anticipatory guidance with regard to firearm safety is to tell the mother to

- A. enroll herself and her child in gun safety classes
- B. ensure that she specifically asks if other guns are in the home
- C. ensure that there are gun safety locks on the shotgun
- D. insist that the gun be stored in a locked gun cabinet or safe with ammunition locked separately
- E. teach the child to use the gun properly at the earliest possible age

32. The mother of a 4-month-old infant is planning a winter trip to the tropics with her infant and asks about the use of sunscreens and the safe amount of sun exposure for the infant.

Of the following, the MOST appropriate advice for the infant is to

- A. apply waterproof sunscreen with a UVB SPF of 30 or greater at least every 30 minutes
- B. avoid mid-day sun and apply sunscreen with a UVB SPF of 15 or greater
- C. avoid all but incidental sun exposure because of decreased sweating and the risk of heat stroke
- D. comply strictly with the use of physical sun blocks such as zinc oxide and titanium dioxide paste
- E. use only special sun-protective clothing

33. You are discussing the epidemiology of childhood drowning in the United States in a public health class at the community college.

Of the following, a TRUE statement about drowning deaths among children younger than 5 years of age is that

- A. drowning is the leading cause of death due to injury
- B. for every one drowning victim there are five near-drownings
- C. pool alarms have eliminated the need for fencing
- D. residential pools are the most common drowning site
- E. the ratio of male-to-female drowning deaths is equal

3:1

34. You are examining a 2-month-old infant who does not yet roll over. Results of physical examination and developmental assessments are normal. The parents are concerned about home safety issues.

Of the following, the MOST appropriate time to discuss the storage of poisonous substances with this infant's parents is

- A. today
- B. at the 4-month visit
- C. at the 6-month visit
- D. at the 9-month visit
- E. at the 12-month visit

35. A 2-year-old child has a venous blood lead level of 45 mcg/dL. He lives in a 50-year-old house. There is no history of pica.

The MOST likely source of this child's lead poisoning is

- A. folk remedies
- B. garden soil
- C. household dust
- D. lead-glazed pottery
- E. lead plumbing

36. You are seeing a previously healthy 12-year-old boy for a health supervision visit. All findings on physical examination are normal except for a blood pressure of 148/90 mmHg.

Of the following, the most appropriate INITIAL management step is to obtain

- A. a 24-hour blood pressure recording
 - B. a urinalysis and urine culture
 - C. renal sonography
 - D. serum concentrations of electrolytes, blood urea nitrogen, and creatinine
 - E. three blood pressure measurements at monthly intervals
37. As part of a kindergarten visit, a pediatric resident sits at a table and draws with the children.

Of the following, the MOST advanced fine motor skill he typically should observe in this group of 5-year-olds is their ability to copy a

- A. circle
 - B. cross
 - C. diamond
 - D. square
 - E. vertical line
38. A 2-month-old breastfed female infant is brought to the emergency department for a low-grade fever (temperature 100° F [37.8° C]). Findings on physical examination are normal. A complete blood count reveals a white blood cell count of $8 \times 10^3/\text{mcL}$ ($8 \times 10^9/\text{L}$), hemoglobin of 9.5 g/dL (95 g/L), and platelet count of $230 \times 10^3/\text{mcL}$ ($230 \times 10^9/\text{L}$). The mean cell volume is 90 fl.

Of the following, the BEST next step in the management of this patient's anemia is to

- A. add a daily multivitamin with iron
 - B. begin oral supplementation of folic acid
 - C. obtain a hemoglobin electrophoresis
 - D. obtain a reticulocyte count
 - E. provide reassurance to the mother
39. A 2-week-old infant whose birthweight was 3.23 kg now weighs 3.0 kg. The mother is breastfeeding and reports good milk production. The infant nurses every 2 to 3 hours and has eight wet diapers per day. Findings on physical examination are unremarkable.

Of the following, the BEST advice for this mother is to

- A. continue to breastfeed and return to the office in 1 week to recheck the infant's weight
- B. hospitalize the infant for evaluation of failure to thrive
- C. return to the office for the 2-month health supervision visit
- D. stop breastfeeding and change to formula
- E. supplement breastfeeding with formula

