NURS 6501

Knowledge Check: Module 8
Student Response

This Knowledge Check reviews the topics in Module 8 and is formative in nature. It is worth 20 points where each question is worth 1 point. You are required to submit a sufficient response of at least 2-4 sentences in length for each question.

Scenario 1: Acute Lymphoblastic Leukemia (ALL)

A ten-year-old boy is brought to clinic by his mother who states that the boy has been listless and not eating. She also notes that he has been easily bruising without trauma as he says he is too tired to go out and play. He says his bones hurt sometimes. Mother states the child has had intermittent fevers that respond to acetaminophen. Maternal history negative for pre, intra, or post-partum problems. Child's past medical history negative and he easily reached developmental milestones. Physical exam reveals a thin, very pale child who has bruises on his arms and legs in no particular pattern. The APRN orders complete blood count (CBC), and complete metabolic profile (CMP). The CBC revealed Hemoglobin of 6.9/dl, hematocrit of 19%, and platelet count of 80,000/mm^3. The CMP demonstrated a blood urea nitrogen (BUN) of 34m g/dl and creatinine of 2.9 mg/dl. The APRN recognizes that the patient appears to have acute leukemia and renal failure and immediately refers the patient to the Emergency Room where a pediatric hematologist has been consulted and is waiting for the boy and his mother. The diagnosis of acute lymphoblastic leukemia (ALL) was made after extensive testing.

Question 1 of 2:

What is ALL?

Acute lymphoblastic leukemia (ALL) is a type of cancer of the blood and bone marrow that affects the white blood cells. ALL is the most common childhood cancer of the blood and occurs when the bone marrow cell develops errors in its DNA. The symptoms of LL include enlarged lymph nodes, bruising, fever, bone pain, bleeding from the gums, and frequent infections. For instance, the case scenario patient has several symptoms of ALL, such as easily bruising without trauma, bone pains, and fever.

Question 2 of 2:

How does renal failure occur in some patients with ALL?

Some patients with ALL, as in the case scenario, can develop renal failure as a consequence of several factors, including leukemic infiltration of the kidneys; chemotherapeutic regime related side effects, which result in the tumor lysis syndrome, nephrotoxic, drugs, and septicemias. The tumor lysis syndrome then leads to acute uric acid calcium phosphate nephropathy. In the case scenario patient, the boy got renal failure as a result of leukemic infiltration of the kidneys, as evidenced by the presence of the high blood urea nitrogen and creatinine contents.

Scenario 2: Sickle Cell Disease (SCD)

A 12-year-old female with known sickle cell disease (SCD) present to the Emergency Room in sickle cell crisis. The patient is crying with pain and states this is the third acute episode she has had in the last nine months. Both parents are present and appear very anxious and teary eyed. A diagnosis of acute sickle cell crisis was made. Appropriate therapeutic interventions were initiated by the APRN and the patient's pain level decreased, and she was transferred to the pediatric intensive care unit (PICU) for observation and further management.

Question 1 of 2:

What is the pathophysiology of acute SCD crisis and why is pain the predominate feature of acute crises?

The loss of beta-cell elasticity is central to the pathophysiology of acute SCD. Sickle Cell Disease (SCD) is caused by a mutation in the beta-globin chain of the hemoglobin molecule. The sickle hemoglobin, which results from the mutation, has a singular property of polymerizing when deoxygenated, causing the red blood cells to change their shape to sickle-like shape under certain conditions. Notably, normal red blood cells are somehow elastic and possess a biconcave disc shape that enables the cells to deform to pass through capillaries. However, in sickle cell

diseases, the low oxygen tension promotes red blood cell sickling, and repeated episodes of sickling cause damage to the cell membrane and reduces the cell's elasticity. The cells fail to return to normal shape when normal oxygen tension is restored. As a result, the rigid blood cells are not able o deform as they pass through the narrow capillaries, result in vessel occlusion and ischemia. Pain is the predominant feature of acute crises because sickled-cells get stuck in a blood vessel, blocking the flow of blood. The common triggers of the pain include sudden temperature change, which can make the blood vessels narrow, very strenuous exercise, or excessive exercise, and due to shortage of oxygen. For instance, the case scenario patient had a sickle cell crisis with pain that has lasted for nine months.

Question 2 of 2:

Discuss the genetic basis for SCD.

SCD occurs when an individual inherits two abnormal copies of the β-globin gene that makes hemoglobin, one from each other. The β-globin gene occurs in chromosome 11. However, various subtypes exist depending on the exact mutation in each hemoglobin gene. The parents of the patient in this scenario each may carry one copy of the gene, but typically do not show the signs and symptoms of the condition.

Scenario 3: Hemophilia

The parents of a 9-month boy bring the infant to the pediatrician's office for evaluation of a swollen right knee and excessive bruising. The parents have noticed that the baby began having bruising about a month ago but thought the bruising was due to the child's attempts to crawl. They became concerned when the baby woke up with a swollen knee. Infant up to date on all immunizations, has not had any medical problems since birth and has met all developmental milestones. Pre-natal, intra-natal, and post-natal history of mother noncontributory. Family history negative for any history of bleeding disorders or other major genetic diseases. Physical exam within normal limits except for obvious bruising on the extremities and right knee. Knee is swollen but no warmth appreciated. Range of motion of knee limited due to the swelling. The

pediatrician suspects the child has hemophilia and orders a full bleeding panel workup which confirms the diagnosis of hemophilia A.

Question 1 of 2:

Explain the genetics of hemophilia

Hemophilia A and B are inherited in an X-linked recessive pattern. Hemophilia is caused by a mutation or change in one of the genes that provide instructions for making the clotting factor proteins required to form a blood clot. The change or mutation can prevent the clotting protein from working as required or from being missing altogether. These genes are situated on the X chromosome, which is one of the two sex chromosomes. For instance, in males, who have only one X chromosome, one altered or mutation of the copy in each cell is sufficient to cause Hemophilia. Females have two X chromosomes and, as a result, need a mutation in both copies of the gene to result in hemophilia. A female with one mutated X chromosome and one healthy X chromosome is a carrier. In the case study, the patient being a boy has a high risk of hemophilia, as evidenced by the symptoms of the condition, such as swollen right knee and excessive bruising.

Question 2 of 2:

Briefly describe the pathophysiology of Hemophilia

The pathophysiology of hemophilia can be explained by the genetic mutation or alteration that occurs as a result of the dysfunction or deficiency of factor VII or by an acquired inhibitor that binds factor VIII. FVIII deficiency, dysfunctional FVIII, or FVIII inhibitors lead to the disruption of the normal intrinsic coagulation cascade, resulting in excessive hemorrhage in response to trauma and, in severe cases, spontaneous hemorrhage. For instance, the case study patient experienced excessive hemorrhage as a result of excessive bruising.

Scenario 4: Myelomeningocele

During a routine 16-week pre-natal ultrasound, spina bifida with myelomeningocele was detected in the fetus. The parents continued the pregnancy and labor was induced at 38 weeks with the birth of a female infant with an obvious defect at Lumbar Level 2. The Apgar Score was 7 and 9. The infant was otherwise healthy. The sac was leaking cerebral spinal fluid and the child was immediately taken to the operating room for coverage of the open sac. The infant remained in the neonatal intensive care unit (NICU) for several weeks then discharged home with the parents after a prescribed treatment plan was developed and the parents were educated on how to care for this infant.

Question 1 of 2:

What is the underlying pathophysiology of myelomeningocele?

Myelomeningocele results from failed closure of the caudal end of the neural tube, resulting in an open lesion or sac that contains dysplastic spinal cord, nerve roots, meninges, vertebral bodies, and skin. In the pathophysiology of myelomeningocele, spina bifida cystica causes a problem when the meningocele, including cord tissue, extends into the cyst. The defect occurs with the spine, and the spinal cord fails to form properly. The defects are known as the neural tube. Neural tube defects are the result of a teratogenic process that causes failed closure and abnormal differentiation of the embryonic neural tube. Neural tube defects occur between the 17th and 30th day of gestation, at a time when the mother may not be aware that she is pregnant and the fetus is estimated to be about the size of a grain of rice. In the case scenario, the patient spina bifida with myelomeningocele was detected in the fetus was detected during the 16-week prenatal ultrasound.

Question 2 of 2:

Describe the pathophysiology of hydrocephalus in infants with myelomeningocele.

Hydrocephalus occurs in some infants with myelomeningocele at birth. In the pathophysiology of hydrocephalus, especially in infants with myelomeningocele, the brain tends to be positioned further down into the upper spinal column than normal, which results in a condition known as an Arnold Chiari II malformation. When this condition occurs, the normal flow of fluid out of the brain is obstructed, leading to an excess of cerebrospinal fluid within the brain, which is known as hydrocephalus. For instance, in the case scenario patient, the sac was leaking cerebral spinal fluid, and the child was immediately taken to the operating room for coverage of the open sac, indicating an excess o cerebrospinal fluid within the brain.

Scenario 5: Patent Ductus Arteriosus (PDA)

A preterm infant was delivered at 32 weeks gestation and was taken to the NICU for critical care management. Physical assessment of the chest and heart remarkable for a continuous-machinery type murmur best heard at the left upper sternal border through systole and diastole. The infant had bounding pulses, an active precordium, and a palpable thrill. The infant was diagnosed with a patent ductus arteriosus (PDA).

Question:

Discuss the hemodynamic consequences of a PDA

There are various hemodynamic consequences of a PDA. One of the most common consequences is high blood pressure in the lungs. A PDA causes too much blood to circulate through the heart's main arteries through patent ductus arteriosus, which leads to pulmonary hypertension that can potentially cause permanent damage to the lung. The other hemodynamic consequence and the most dangerous consequence is cerebral under-perfusion due to diastolic reverse-flow and resulting in cerebral hypoxia. Besides, the other consequence is the infection of the blood vessels called bacterial endocarditic, which is also a life-threatening condition. For

instance, in the case scenario, the patient risks developing these hemodynamic consequences if the treatment if not undertaken promptly.

Scenario 6: Lead Poisoning

A 7-year-old male was referred to the school psychologist for disruptive behavior in the classroom. The parents told the psychologist that the boy has been difficult to manage at home as well. His scholastic work has gotten worse over the last 6 months and he is not meeting educational benchmarks. His parents are also worried that he isn't growing like the other kids in the neighborhood. He has been bullied by other children which is contributing to his behaviors. The psychologist suggests that the parents have some blood work done to check for any abnormalities. The complete blood count (CBC) revealed a hypochromic microcytic anemia. Further testing revealed the child had a venous lead level of 21 mcg/dl (normal is < 10 mcg/dl). The child was diagnosed with lead poisoning and it was discovered he lived in public housing that had not finished stripping lead paint from the walls and woodwork.

Question:

How does lead poisoning account for the child's symptoms?

Lead poisoning exposure to high levels of lead as the child may cause anemia, weakness, and kidney problems as well as brain damage. Besides, the high levels of lead in the body can cause stillbirths, developmental delays, neurological changes, and irritability. In the case scenario patient, lead poisoning resulting in delayed development and neurological changes that made the child to experience abnormal growth differently from his peers. Besides, the neurological effects of lead poisoning made the child find it difficult to manage academic work, which has deteriorated over the past six months. The child cannot meet his educational benchmark due to the neurological effects of lead.

Scenario 7: Sudden Infant Death Syndrome (SIDS)

Emergency Medical Services (EMS) was dispatched to a home to evaluate the report of an unresponsive 3-month-old infant. Upon arrival, the EMS found a frantic attempt by the presumed father to resuscitate an infant. The EMS took over and attempted CPR but was unable to

restore pulse or respiration. The infant was transported to the Emergency Room where the physician pronounced the child dead of Sudden Infant Death Syndrome (SIDS). The distraught parents were questioned as to the events surrounding the discovery of the baby. Parents state the child was in good health, had taken a full 6-ounce bottle of formula prior to being put down for the evening. The child had been sleeping through the night prior to this. Parents stated the baby had had some "sniffles" a few days before and was taken to the pediatrician who diagnosed the child with a mild upper respiratory tract viral syndrome. No other pertinent history.

Question:

What is thought to be the underlying pathophysiology of SIDS?

In its pathophysiology, respiratory dysfunction, such as upper airway obstruction, plays an important role in the primary mechanism of death in most SIDS babies. The pathogenesis of SIDS involves a persistence of fetal reflex responses into early infancy, during which enhanced inhibitory and depressed excitatory cardiorespiratory reflex responses to local stressors are present, leading to sudden death during sleep in otherwise normal-appearing infants. The defects in these critical life-sustaining neural pathways likely arise during fetal development and, in some cases, are further influenced by prenatal and postnatal exposure to cigarette smoke and alcohol. For instance, the child had an upper respiratory tract viral syndrome, which possibly results in respiratory dysfunction that leads to SIDS.

Scenario 8: Kawasaki Disease

A 4-year-old female is brought to the pediatrician by her mother who states the child has been running a fever to 102.0 F, has "pink eye", and that her tongue looks very bright red and swollen. The mother states the fever has been present for 5 days, noticed the child had developed a rash and that the child's legs look "puffy". No other symptoms noted. Past medical history noncontributory. All immunizations up to date. Physical exam remarkable for current fever of 102.8 F, bilateral conjunctivitis without purulent material, oral mucosa with bright red erythema, dry, with fissuring of the lips. Legs noted to have peripheral edema and are also erythematous. Palmar desquamation noted. There is fine maculopapular rash and + cervical adenopathy. The presumptive diagnosis currently (pending laboratory data) is Kawasaki Disease.

Question 1 of 2:

What is Kawasaki Disease and what is the pathophysiology?

Kawasaki Disease is a condition that causes inflammation in the walls of some blood vessels in the body. The early stages of the symptoms include high fever and peeling skin. In the late stages, there may be inflammation of medium-sized blood vessels. In its pathophysiology, in the earliest stages of the disease, the endothelial cells and the vascular media become edematous, but the internal elastic lamina remains intact. In approximately 7-9 days after the onset of fever, an influx of neutrophils occurs, which is quickly preceded by a proliferation of CD8+ lymphocytes and immunoglobulin A–producing plasma cells. The inflammatory cells secrete various cytokines, interleukins, and matrix metalloproteinase that target the endothelial cells and result in a cascade of events that lead to fragmentation of the internal elastic lamina and vascular damage. In severely affected vessels, the media develops inflammation with necrosis of smooth muscle cells. The internal and external elastic lamina can split, leading to aneurysms and, consequently, Kawasaki Disease. In the case scenario, the child presented with various symptoms of the disease such as fever, rashes on the skin, and puffy legs, as well as with her tongue that looks very bright red and swollen, indicating an inflammation.

Question 2 of 2:

How does Kawasaki Disease cause coronary aneurysms?

Kawasaki Disease is an acute vasculitis of childhood that predominantly affects the coronary arteries by weakening their walls. The weakening of the walls of the arteries occurs as part of the vasculitis leading to coronary aneurysms, which are abnormal dilations of the coronary arteries as evidenced by legs with peripheral edema and erythematous in the case scenario patient, indicating compromised arteries.

Scenario 9: Asthma

A 9-year-old boy was brought to the Urgent Care Center by his parents who state that the child had a sudden onset of difficulty catching his breath, has a new cough and is making a "funny sound" when he breathes. The parents state there is no prior history of this, and the child had not been ill prior to the start of the symptoms. Past medical history noncontributory. No family history of respiratory problems. No known allergies to drugs or food. Physical exam positive for respiratory rate of 26, use of accessory muscles, with suprasternal retractions, heart rate of 132 beats per minute, an audible inspiratory and expiratory wheeze noted, and the pulse oximetry is 89% on room air. After the APRN institutes appropriate urgent treatment, the child's breathing slowly returned to normal, vital signs normalize, and the pulse oximetry increases to 97%. The APRN suspects the child has asthma and tells the parents that they need to bring the child to a pulmonologist for further evaluation and care.

Question:

What is the underlying pathophysiology of asthma?

The pathophysiology of asthma is sophisticated and incorporates airway inflammation, intermittent airflow obstruction, and bronchial hyperresponsiveness. The mechanism of inflammation in asthma may be acute, subacute, or chronic, and the presence of airway edema and mucus secretion also contributes to airflow obstruction and bronchial reactivity. Airflow obstruction can be caused by a variety of changes, including acute bronchoconstriction, airway edema, chronic mucous plug formation, and airway remodeling. Airway obstruction causes increased resistance to airflow and decreased expiratory flow rates. These changes lead to a decreased ability to expel air and may result in hyperinflation. During bronchial hyperresponsiveness, Hyperinflation compensates for the airflow obstruction, but this compensation is limited when the tidal volume approaches the volume of the pulmonary dead space; the result is alveolar hypoventilation. Uneven changes in airflow resistance, the resulting uneven distribution of air, and alterations in circulation from increased intra-alveolar pressure due to hyperinflation all lead to ventilation-perfusion mismatch. The actions of the three

mechanisms lead to airway inflammation and asthma symptoms, as noted in the case scenario patient.

Scenario 10: Cystic Fibrosis (CF)

A 24-year-old female with known cystic fibrosis (CF) has been admitted to the hospital for evaluation for possible lung transplant. She was diagnosed with CF when she was 9 months old and has had multiple hospitalizations for pneumonia, respiratory failure, and small bowel obstructions. She currently is oxygen dependent and has been told by her physicians that she has end stage pulmonary disease secondary to CF. The only recourse for her currently is lung transplant.

Question 1 of 2:

What is cystic fibrosis and discuss the pathophysiology

Cystic fibrosis is a hereditary disease that affects the lungs and digestive system. The body produces thick and sticky mucus that can clog the lungs and obstruct the pancreas. The pathophysiology of CF results from a mutation in the CF transmembrane conductance regulator (CFTR) gene. The CFTR protein produced by this gene regulates the movement of chloride and sodium ions across epithelial cell membranes. When mutations occur in one or both copies of the gene, ion transport is defective. It results in a buildup of thick mucus throughout the body, leading to respiratory insufficiency, along with many other systemic obstructions and abnormalities. A combination of decreased mucociliary clearance and an altered ion transport allows for bacterial colonization of the respiratory tract, most commonly Pseudomonas, Haemophilus influenza, and Staphylococcus aureus. These pathogens cause an overwhelming inflammatory response. Ultimately, chronic infection and this repetitive inflammatory response can lead to airway destruction, as evidenced by the respiratory failure in the case scenario patient.

Question 2 of 2:

What is the reason people with CF are often malnourished?

People with CF are often malnourished due to maldigestion and malabsorption of food. Besides, patients with C require more calories than other individuals because they often demand a lot of energy due to coughing, increased work to breath due to chest problems, and repeated infections. For instance, in the case scenario patient, the maldigestion and malabsorption are evidenced by small bowel obstructions.

Scenario 11: Idiopathic Scoliosis

A 14-year old girl who was trying out for cheerleading underwent a physical examination by the APRN who notices that the girl had uneven hip height, asymmetry of the shoulder height, shoulder and scapular prominence and rib prominence. The rest of the physical exam was normal and the APRN referred the girl to an orthopaedist for evaluation for possible scoliosis. Radiographs in the orthopaedic office confirms the diagnosis of idiopathic scoliosis. The spinal curve was measured at 26 degrees and it was recommended that the girl be fit for a low-profile back brace.

Question:

What is thought to be the pathophysiology of idiopathic scoliosis?

Even though the pathophysiology of idiopathic scoliosis is not clearly understood, it is believed to be caused by various mechanisms such as asymmetric bone growth dysregulation, susceptibility of bones to deformation, abnormal passive spinal system maintenance and disturbed active spinal system maintenance. Symptoms include uneven shoulders with one shoulder blade appearing more prominent than the other, uneven waist, one hip higher than the other. The patient in the case scenario had all these symptoms.

Scenario 12: Hemolytic Uremic Syndrome (HUS)

- A 2-year-old boy was brought to Urgent Care by his parents who state the boy has been having large amounts of diarrhea, been very irritable and very pale. The parents noticed there was blood in the diarrhea and when the boy's legs became swollen, they sought care. Past medical history noncontributory and all immunizations up to date. Social history noncontributory and the child is in day care 5 days a week. No known exposure to other sick children and the only new event the parents could think of is the day care workers took the children to a local petting zoo about a week ago. Physical exam revealed a pale, ill appearing child with swollen legs, tender abdomen, and petechia on the legs and abdomen. The APRN suspects the child may have been exposed to a bacterium at the petting zoo and arranges for the patient to be transferred to the Emergency Room. There the child was found to be in renal failure, have hypertension and was diagnosed with hemolytic uremic syndrome (HUS). The symptoms include bloody diarrhea, abdominal pain, pale skin, irritability, fatigue, fever, unexplained bruises or bleeding, decreased urination.

Question:

What is the pathophysiology of HUS?

The pathophysiology of HUS involves the binding of Shiga-toxin to the globotriaosylceramide receptor on the surface of the glomerular endothelium. This action includes a cascade of signaling events leading to apoptosis and binding of leukocytes to endothelial cells. The Shiga-toxin-activated endothelial cells then become thrombogenic by a mechanism that induces the release of cytokines and chemokines that are implicated in platelet activation. Additionally, the binding action of Shiga-toxin inactivates a metalloproteinase called ADAMTS13, the deficiency of which causes the closely related TTP. Once ADAMTS13 is disabled, multimers of von Willebrand Factor (vWF) form and initiate platelet activation, leading to the microthrombus formation. As a result, the arterioles and capillaries of the body become obstructed by the resulting complexes of activated platelets, which have adhered to the

endothelium via large multimeric vWF. Through a mechanism known as microangiopathic hemolysis, the growing thrombi lodged in smaller vessels destroy red blood cells (RBCs) as they squeeze through the narrowed blood vessels, forming schistocytes, or fragments of sheared RBCs. For instance, the case scenario patient showed symptoms of HUS, such as diarrhea and very irritable and pale skin.

Scenario 13: Pituitary Dwarfism

The parents of a 3-year-old boy bring the child to the pediatrician with concerns that their child seems "small for his age". The parents state that the boy has always been small but did not worry until the child went to day care and they noticed other children of the same age were much bigger. They also note that his teeth were very late in coming in. Normal prenatal, perinatal and postnatal history and no medical history on either side of family regarding issues with growth and development. Physical exam is normal except for short limbs and small teeth. The pediatrician suspects the child has pituitary dwarfism. A complete laboratory and radiographic work up confirmed the diagnosis.

Question:

What is the pathophysiology of pituitary dwarfism?

The pathophysiology of pituitary dwarfism can be explained by a deficiency of pituitary growth hormone, which is the main endocrine form of dwarfism and results from various factors such as hereditary tumors, infections, or infarction of the pituitary which induce dwarfism. The symptoms include below-average growth, an immature appearance, a chubby body build, a prominent forehead, and an underdeveloped bridge of the nose. For instance, the case scenario patient seems small for his age and had other slow growth of the teeth.

Scenario 14: Osteogenesis Imperfecta (OI)

A 4-year-old boy was brought to the Emergency Room by his parents with a suspected femur fracture. The parents state the child was playing on the couch when he rolled off and cried out in pain. There were no other injuries noted. Review of the child's chart revealed this was the

4th Emergency Room visit in the last 15 months for fractures after low impact injury. The parents were suspected of child abuse and Child and Protective Services were consulted. The APRN assessing the child noted that the child had unusually thin and translucent skin, poor dentition, and blue sclera. The APRN suspects the child may have osteogenesis imperfecta (OI). Laboratory results revealed an elevated serum alkaline phosphatase and the diagnosis OI was made based on the clinical picture and elevated alkaline phosphatase.

Question:

What is the pathophysiology of OI?

Osteogenesis Imperfecta (OI) is a complex group of disorders characterized by excessive fragility of bone and pathologic fractures. The pathophysiology of OI is not clearly understood, but most cases of OI are due to defects in the expression or structure of Type I collagen, the principal collagen of bone leading to structurally abnormal collagen chains, in which the majority are single amino acid substitutions for glycine residues. The common symptoms include pain, bone fracture, abnormal bone formation, blue sclerae, bow leggedness, bruise, corns and calluses, deformity, hearing loss, macrocephaly, scoliosis, short stature, or stiffness. For instance, the case scenario patient had suspected femur fracture and felt pain after rolling off the coach, indicating major symptoms of IO.