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Sickle Cell Anemia

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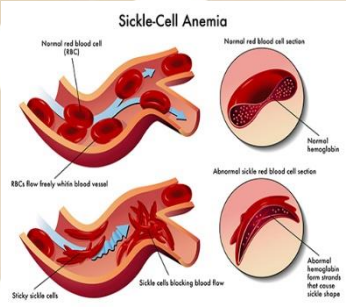
Introduction

Sickle Cell Anemia (SCA) is a group of inherited red blood cell (RBC) disorders caused by a single mutation on the β-globin gene which causes the synthesis of an abnormal hemoglobin S (HbS) (Connes et al., 20118). A healthy red blood cell has a round shape that allows it to easily move through small blood vessels to carry oxygen to supply to all parts of the body (CDC, 2017). In SCA individuals, the sickle cell assumes a C-shape called "sickle" (CDC, 2017). This sickle shape gets them to stuck and clog the blood flow when moving through the small blood vessels, this lead to the typical symptoms of pain and infection, and complication such as chest syndrome and stoke (CDC, 2017).

African countries carry the highest incident of individuals with the SCA (Centers for Disease Control and Prevention (CDC), 2017). In the United State, approximately 1 in 12 African-Americans carry the trait for SCA, and 1 of every 350 African-American infants born have the disorder (CDC, 2017). However, the disease is also familiar with Hispanics, Indians, and individuals with Asian backgrounds (CDC, 2017).

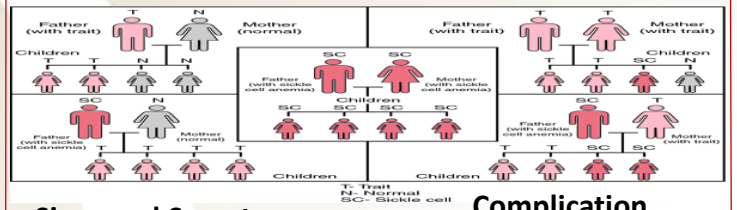


Topic Statement

SCA is a public concern. The cost of hospital stay during crisis and management of complication of SCA was estimated at $488 billion (CDC, 2017). With early and effective interventions, there could be a decrease in hospitalizations for cost-effectiveness and also increase life expectancy and overall quality of care delivered to individuals in this population. Understanding the pathophysiology underlining SCA will enable health care providers to effectively manage symptoms, prevent complication and treat pain to improve quality of life in this patients population.

Pathophysiological Process

- SCA is a genetic disorder inherited from a mother, father, or both parents that are present from birth. SCA is as a result of a single mutation on a hemoglobin-beta gene which causes the synthesis of an abnormal hemoglobin S (HbS) (Connes et al., 20118). Hemoglobin (Hb) is a molecule in RBC responsible for the transportation of oxygen hence during deoxygenation, HbS polymerizes and causes sickle cell deformity which also initiates a pathological cascade that results in injury to the red sickle cell, hemolysis, and vaso-occlusion of RBC (Connes et al., 2018).
- The sickle-shaped RBC causes a blockage that slows or stops the flow of blood(Connes et al., 2018). When this happens, oxygen cannot reach targeted tissues. Deoxygenation can cause attacks of sudden and severe pain called "pain crises" (Connes et al., 2018) Due to the deformed shape, cells can also stick to vessel walls, and HbS induces RBC membrane damage leading to an imbalance of calcium, potassium, and water exchange into and out of the cell (Waltz & Connes, 2014). Accumulation of Calcium within the cell allows for the efflux of potassium and water from the cell, which leads to dehydration and Vaso-occlusive crisis (Waltz & Connes, 2014). The vasso-occlusion causes an obstruction and reduce blood flow to the vital organs leading to ischemia, necrosis, and pain. Repeated episodes of deoxygenation and oxygenation weaken the RBC cell membranes (Smith, 2014). The average life span of a healthy RBC is 90 to 120 days. However, weakened RBCs are hemolyzed and removed within 10 to 20 days resulting in increased demand for RBC production that produces the symptoms of feeling fatigued and lethargy (Waltz & Connes, 2014).
- SCA has an autosomal recessive inheritance pattern, which means an individual must inherit two copies of a recessive allele from both parents in order to have the condition: individuals that carry one dominant and one recessive allele are carriers (CDC, 2017). Sickle cell carriers usually live a healthy life and do not exhibit any of the signs of the disease; however, they can pass the trait on to their offspring (CDC, 2017).
- The risk for septicemia and meningitis is prevalent in about 10% of children with SCA during the first three years of life; bacterial infections are the most common cause of death at any age (Matthie & Jenerette, 2015). The swelling and inflammation of the hands and other infections are caused by poor blood circulation due to blocked vessels (Matthie & Jenerette, 2015). Severe abdominal pain is caused by splenic sequestration that's blood pooling within the spleen, which causes the spleen to become enlarged (splenomegaly) (Matthie & Jenerette, 2015). Breathing complications (chest syndrome) results from lack of oxygen being transported from the lungs to other areas of the body and retention of carbon dioxide (Smith, 2014). Deoxygenation and high levels of carbon dioxide also cause other complications of the heart, brain, and muscles as these organs rely on oxygen for homeostasis (Smith, 2014).



Signs and Symptoms

During crisis, signs and symptoms may include

- Pallor
- Pain crisis
- Fatigue
- Lethargy
- Anemia
- Jaundice
- irritability
- swelling of hands, feet and joints.

(Matthie, & Jenerette, 2015).

Complication

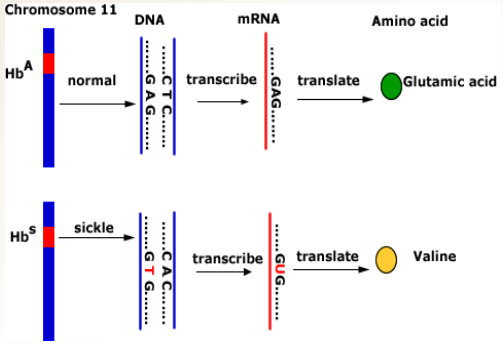
The most serious, acute complications of SCA include:

- cardiovascular collapse due to acute sequestration crisis
- cerebrovascular accident
- acute respiratory distress syndrome due to acute chest syndrome
- aplastic crisis that progresses to high-output heart failure
- pneumonia and infections that progress to septicemia

The most serious, chronic complications of SCA include organ failure of the heart, liver, and kidney and loss of vision that may lead to complete blindness.

Underlying Pathophysiology

- Every healthy adult hemoglobins consist of two alpha chains with 141 amino acids in sequence complemented non-alpha chains of beta, gamma or delta (Bellas, 2018). The non-alpha chain instead composes of 146 amino acids in the sequence (Bellas, 2018).
- Sickling of RBC happens when a healthy alpha chain of amino acid of Hemoglobin is substituted with the beta chain (Bellas, 2018). This result in the abnormal synthesis of one of the amino acid chains on chromosome 11(Bellas, 2018).
- Every beta-globin chain has a sequenced group of amino acids indicating glutamine in the sixth position from the terminal end (Bellas, 2018). In SCA, the abnormal beta-globin chain is a result of a substitution of glutamine amino acid by valine indicated on the sixth position of the beta chain which results in abnormal HbS present in the person's red blood cells (Bellas, 2018).
- The inheritance of a single abnormal amino acid makes an individual a carrier with sickle cell trait which means that the person inherits one copy of alpha (α) chains (HBA) and one copy of beta (β) chains (HbS) (Bellas, 2018). Receiving two copies of mutated two beta (β) chains (HbS) inheritance results in sickle cell disease (SCD) with severe signs and symptoms that alter the person's quality of life and life span (Bellas, 2018).



Significance of Pathophysiology

- Understanding the etiology and the pathophysiology underlining the SCA will enable health care providers to effectively manage symptoms and treat pain to improve the quality of life in this patients population.
- Primary prevention involves genetic counseling and general public knowledge about the pathophysiological process of the disease.
- Early detection through embryonic and newborn screening is necessary to reduce morbidity.
- Management of SCD is mainly prevention of complications and aggressive treatment of hydration during a crisis as well as pain management. For children, prophylactic antibiotics and pneumococcal vaccines are encouraged as well as stroke prevention techniques.
- that bone marrow transplant is the only evidenced base cure for sickle cell anemia and other experimental trails such as gene therapy, nitric oxide, and drugs that helps to boost hemoglobin production and reduce viscosity such as GMI 1070 are underway (Wun et al., 2014).

Implication for Nursing Care

Nurses play a significant role in the management of signs and symptoms and treatment of individuals with SCA. Since treatment is aimed at relieving of symptoms and prevention of complication educating patients on taking their prescribed medications such as antibiotics, vitamins, and other pain-relieving medicines to prevent a crisis is essential (Smith, 2014). Nurses ensure to teach patients effective measures to prevent crisis such as drinking more fluids, staying warm, avoiding large crowds, avoiding smoking, and second-hand smoking (Matthie & Jenerette, 2015). The nurse teaches the parents to understand effective measures to control pain. For the teen and adult, the nurse educates patients to choose safe physical activities that can improve pain management yet reduce stress on the joint to avoid further complications (Waltz & Connes, 2014). During a crisis, nurses ensure to prevent infection, and carefully assess patients for early signs of complications that indicate damage to any organ, and promptly report critical labs and negative diagnostic test for specific treatment including possible surgery and blood transfusions (Matthie & Jenerette, 2015).

Knowing the pathophysiology of the disease, the nurse will treat pain with analgesics and hydration promptly. The nurse educates and advocate for the patients and family about their treatment options The nurse ensures that patients stay current on their childhood vaccinations, pneumococcal vaccine, and the annual flu shot for prevention of infection and other complications (Onimoe & Smarzo, 2017). The nurse will educate patients about pain medication tolerance, how to manage stress and explore other pain relief techniques such as heat, therapy, acupuncture, and when to contact the healthcare provider (Onimoe & Smarzo, 2017). The nurse will provide resources for community support groups and organizations that will help patients with other specialty coping mechanisms

Conclusion

SCA is an inherited blood disorder where RBC takes an abnormal C-shape which impedes blood flow in vessels (Onimoe & Smarzo, 2017). This obstruction of blood blow causes Vasso-occlusion that result in acute illness and progressive multiple organ damage. Pain "crisis" is the hallmark symptoms of SCA (Onimoe & Smarzo, 2017). SCA is caused by a mutation in the hemoglobin beta gene situated on chromosome 11 (Bellas, 2018). SCA is a recessive genetic disorder where two copies of mutated genes have to be inherited from both parents for one to portray the symptoms (Bellas, 2018). Individuals who inherit a copy of the mutated gene are referred to be the sickle cell trait carrier who may live symptoms free in their lifespan (Bellas, 2018).The average life expectancy for this population has increased as a result of improved management of symptoms and bone marrow replacement (Gardner, 2018). Health care providers ensure to educate patients on the importance of following medication regime to reduce aggregation of symptoms to prevent complication (Waltz & Connes, 2014). Both family and patient education are essential to reduce prevalence and complication of SCA.

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