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University of Belize Faculty of Nursing and Allied Health NURS 3051- Nursing Care of Infants and Children Congenital Defects Sickle Cell Anemia (SCA) Sickle Cell Anemia (SCA) is one of a group of diseases collectively termed hemaglobinopathies, in which normal adult hemoglobin is partially or completely replaced by abnormal sickle hemoglobin(HgbS). Sickle Cell Anemia includes all of those hereditary disorder, the clinical, hematologic, and pathologic features of which are related to the presence of HbgS. Also know as SS and homozygous sickle cell disease. The most common form of SCD are: Sickle Cell Anemia — The homozygous form of the disease (HgbSS) . Sickle Cell —C Disease a heterozygous variant of SCD, including both HgbS and hemoglobin C (HgbC). Sickle Cell- E (HgbE) Disease - A variant of SCD in which glutamic acid has been substituted for lysine in the number 26 position of the Î²-chain. Sickle Thalassemia Disease — A combination of sickle cell trait and Î²+ (beta plus) refers to the ability to still produce some normal adult hemoglobin. Î’Î¿ (beta zero) indicates that there is no ability to produce normal adult hemoglobin. Of the SCDs, SCA is the most common form in Blacks, followed by Sickle Cell C Disease and Sickle Î²-thalassemia. Another Î²-chain variant, HgbE, is found primarily in people of Southeast Asian origin. People who carry the trait for HgbE are completely asymptomatic, but those who are homozygous exhibit a disease similar to HgbC disease. SCA is found primarily in the Black race, occasionally in Hispanics and infrequently in Whites. Incidence of the disease vary in different geographic locations. Among American Blacks the incidence is 8%. In West Africa the incidence is reported to be as high as 40%. Believed to be a result of of selective protection of trait carriers against malaria caused by Plasmodium falciparium. Mood of Transmission The gene that determines the

production of Hgbs is situated on an autosome. (autosomal recessive inheritance) The expected pattern of transmission from two parents who carry the heterozygous gene HgbAS is a 25% chance of their producing an offspring with SCA. Characteristics of autosomal recessive inheritance Males and females are affected with equal frequency Affected individuals will have unaffected parents who are heterozygous for the trait There is a 1: 4 (25%) chance that any child of two unaffected heterozygous parents will be affected. Unaffected siblings of an affected person have a â…” risk of being a carrier. Affected individuals mated to normal individuals will have normal children, all of whom will be carriers. There is usually no evidence of the trait in previous generations-a negative family history- unless consanguinity is a factor. Basic Defect The basic defect responsible for the sickling effect of erythrocytes is in the globin fraction of hemoglobin, which is composed of 574 amino acids. HgbS differs from HgbA in the substitution of only one amino acid (valine) for another (glutamine) at the sixth position of the Î²-polypeptide chain. Under conditions of dehydration, acidosis, hypoxia, and temperature elevations, the relatively insoluble HgbS changes its molecular structure to form long, slender crystals. These filamentous crystals cause distortion of the cell membrane from a pliable disk to a crescent- or sickle-shaped RBC. The filamentous forms are associated with much greater viscosity than the normal holly leaf structure of HgbA. In most instance the sickling response is reversible under conditions of adequate oxygenation and hydration. However after repeated cycles of sickling and unsicklng, the RBC becomes irreversibly sickled. Although the defect is inherited the sickling phenomenon is not apparent until later in infancy because of the presence of fetal hemoglobin (HgbF). As long as HgbF persists, sickling does not occur

because there are no Î²-chains carrying the defect. The newborn has 60% to 80% fetal hemoglobin, but this rapidly decreases during the first year. Sickle Cell Trait Same basic defect as for SCA, but only about 35% 45% of the total hemoglobin is HgbS. The remainder is HgbA. Normally, these individuals are asymptomatic. Although rare, complications have been described, primarily nonpainful, gross hematuria especially in the teenage and adult years. Under conditions of extreme or prolonged deoxygenation, such as riding in unpressurized aircraft or military training, splenic sequestration with profound anemia can often, resulting in death. Pathophysiology and Clinical Manifstations The clinical manifestations of SCA are primarily the result of (1) obstruction caused by the sickled RBCs and (2) increased RBC destruction. -the entanglement and enmeshing of rigid sickle-shaped cells with one another intermittently block the microcirculation, causing vaso-occlusion -the resultant absence of blood to adjacent tissues causes local hypoxia, leading to tissue ischaemia and infarction (cellular death). Spleen- initially enlarges from congestion and engorgement with sickled cells. -repeated insult to splenic sinuses results in infarction - functioning cells gradually replaced with fibrotic tissue, until by age 5 years the spleen is decreased in size and totally replaced by a fibrous mass (functional asplenia) - without the spleen to filter bacteria and to promote the release of large number of phagocytic cells, these individuals are highly susceptible to infection. Liver Altered in form and function Liver failure and necrosis are the result of severe impairment of hepatic blood flow from anemia and capillary obstruction. Hepatomegally common by 1 year and usually persists throughout childhood and early adulthood. -rapid destruction of RBCs results in the development of pigmented gallstones Kidney Abnormalities are a result of congestion of

glomerular capillaries and tubular arterioles with sickle cells and hemosiderin, tissue necrosis, and eventual scarring. Principal results of kidney ischaemia are hematuria, inability to concentrate urine, enuresis and occasionally nephritic syndrome. Bone Bone changes include hyperplasia, and congestion of the bone marrow, resulting in osteoporosis, widening of the medullary spaces and thinning of the corteces. Skeletal deformities result from weakening of the bones- lordosis, kyphosis Chronic hypoxia causes the bones to be susceptible to osteomylitis, frequently from salmonella organisms. Aseptic necrosis of the femoral head from chnonic ischaemia (occasionally). Central Nervous System Primarily vascular from occlusion, ischemia and infarction. -Stroke or cerebrovascular accident (CVA) occurs in 6% to 10% of children with SCD and can result in permanent paralysis or death -A number of neurologic symptoms can herald a minor cerebral insult: Headache, aphasia, weakness, convulsions, visual disturbances or unilateral hemiplegia Loss of vision usually the result of progressive retinopathy and retinal detachment -Probable cognitive impairment Heart Attritubutable to the stress of chronic anemia, can result in decompensation and failure. Cardiomegaly, systolic murmur, septal hypertrophy Anemia, increased hemolysis, hemosiderosis Other Signs and Symptoms -exercise intolerance -anorexia -jaundiced sclera -gallstones -chronic leg ulcers in adolescents and adults- due to decreased circulation caused by vaso-occlusion and tissue ischaemia -growth retardation in both height and weight -delayed sexual maturation and decreased fertility Sickle Cell Crises The most acute symptoms of SCA occur during periods of excacerbation called crises. There are several types of crises: vaso-occlusive, acute splenic sequestrqtion, aplastic, hyper- hemolytic, stroke chest

syndrome and infection. The crises may occur individually or concomitantly with one or more other crises. Vaso-occlusive Crises (VOCs) are the most common and non-life threatening. a result of sickled cells obstructing the blood vessels, causing occlusion, ischaemia and potentially necrosis -VOC symptoms: mild to severe bone pain, acute abdominal pain from visceral hypoxia or gallstones, priapism, and arthralgia -pain is usually migratory, localized or generalized - low-grade fever VOC can result in a variety of skeletal problems. One of the most frequent is the hand-foot syndrome (dactylitis) — occur in children 6 months to 2 years. Caused by infarction of short tubular bones, characterized by pain and swelling of the soft tissue over hands and feet. Usually resolved spontaneously within a couple of weeks. Localized swelling over joints with arthralgia can occur with from erythrostasis with sickle cells. Splenic Sequestration Crises Caused by the spleen’s sequestering (pooling) large quantities of blood, causing a precipitous drop in blood volume and ultimately shock. May be acute or chronic, chronic crisis known as hypersplenism Aplastic Crisis Diminished RBC production, usually triggered by a viral or other infection. When superimposed on existing rapid destruction of RBCs, a profound anemia results. Packed RBC transfusion occasionally required. Megaloblastic Anemia Attributed to an excessive nutritional need for folic acid and/or vitamin B12 during periods of pronounced eythropoesis. Hyperhemolytic Crisis Occurs when there is an accelerated rate of RBC destruction characterized by anemia, jaundice, and reticulocytosis. Other co-existing conditions such as viral infection, transfusion reaction Stroke Sudden and severe complication, often with no related illnesses -sickle cells block the major blood vessels in the brain, resulting in cerebral infarction, causing variable degrees of

neurologic impairment Repeat strokes cause progressively greater brain damage in 47% to 93% of children Chest Syndrome Clinically similar to pneumonia, associated with chest pain, fever, pneumonia-like cough, and associated anemia. Believed to be a result of a VOC or infection that cause sickling in the small blood vessels of the lungs, with occlusion, stasis, and anemia. Repeated episodes of chest syndrome may cause restrictive lung disease and pulmonary hypertension Overwhelming Infection Major cause of death in children under the age of 5 years with SCD. Streptococcus pneumonia and Hemophilus influenzae type b most common organisms. Result of defective splenic function. Repeated insult on splenic sinuses by sickled cells result in impaired filtration and function, allowing the development of septicemia and possible death. Diagnostic Evaluation - Stained Blood Smear — may reveal a few sickled RBCs -Sickle Turbidity Test (Sickledex) — A reliable screening method -Hemoglobin Electrophoresis- fingerprinting of the protein —accurate, rapid and specific for detecting the homozygous and heterozygous forms of the disease Therapeutic Management The aim of therapy is to (1) prevent the sickling phenomenon and (2) to treat the medical emergency of sickle cell crises. Three types of treatment available: supportive and symptomatic, specific and curative. - Research investigating hydroxyurea and erythropoietin, which may increase the concentration of fetal hemoglobin and ultimately reduce complications. - Bone Marrow Transplantation also being investigated - Main general objectives; 1. bed rest to minimize energy expenditure and oxygen utilization 2. hydration for hemodilution through oral and IV therapy 3. electrolyte replacement, since hypoxia results in metabolic acidosis which promotes sickling 4. analgesics for the severe pain of vaso-occlusive crises 5. blood

replacement to treat anemia 6. antibiotics to treat any existing infection 7. Hib and meningoccal vaccines recommended because of susceptibility to infections Oxygen therapy may help in severe crises especially with cardiac failure Splenectomy may be a life saving measure in children with recurrent splenic sequestration. Autosplenectony has several advantages since the spleen is the major site of sickling, sequestration and destruction of RBCs. Prognosis Varies Greatest risk in children under 5 years of age Majority of death caused by overwhelming infection As child grows older the crises become less severe and less frequent SCA is chronic illness with a potentially terminal outcome Nursing Considerations Assessment -Nurses involved in screening for SCA -Assessment of child in Sickle Cell Crises Nursing Diagnoses Derived from observation and assessment: 1. Risk for injury related to abnormal hemoglobin, decreased ambient oxygen, dehydration 2. Pain related to tissue anoxia (vaso-occlusive crisis) 3. Altered family processes related to a child with potentially life-threatening disease Planning The primary goals for the child with SCD and the family are: 1. Child will experience minimal effects of sickling. 2. Family and child (when appropriate) will receive adequate education regarding the sickling phenomenon, possible consequences, and genetic counseling. 3. Child and family will adjust to a lifelong, potentially fatal hereditary disease. Implementation Minimize Tissue Deoxygenation Promote Hydration Minimize Crises Promote Supportive Therapies During Crisis Decrease Surgical Risk Encourage Screening and Genetic Counseling Support Family Explain the Disease Evaluation The effectiveness of nursing intervention is determined by continual reassessment and evaluation of care based on the following observations: 1. Observe child for any evidence of sickling; monitor

preventive therapies and strategies 2. Interview family regarding genetic counseling 3. Interview family regarding their understanding of the disease, the sickling phenomenon and its consequences 4. Interview and observe child and family regarding the way in which the disease has affected their lives. REFERENCE Whaley, L., & Wong, D. (2000). Nursing Care of Infants and Children (6th ed.) St. Louis, MO: Mosby. Pp 1666 -1676.