The Child with Sickle Cell Disease
A Teaching Manual

Written and Developed by Debra A. Vedro, MSN, RN, CPNP
And Rebecca A. Morrison, MSN, RN, CPNP
Children's Medical Center of Dallas, Dallas, Texas

Illustrated by Mary Ann Zaplac, MA

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Contents

Origin and Distribution of Sickle Cell Disease        Page 3
Genetics                                            Page 4
Sickle Red Blood Cells                               Page 9
Infections                                          Page 10
Splenic Sequestration                               Page 12
Pain                                                Page 14
Chest Syndrome                                      Page 17
Aplastic Crisis                                     Page 18
Strokes                                             Page 19
Gall Stones and Jaundice                            Page 20
Growth and Development                              Page 21
Retinopathy                                         Page 23
Priapism                                            Page 24
Origin and Distribution of Sickle Cell Disease

Sickle cell trait occurred as a natural mutation of the hemoglobin gene. Sickle trait serves as a protective mechanism against malaria. Malaria is a deadly disease found in countries along the equator. People with sickle cell trait are protected with sickle cell anemia and normal hemoglobin are susceptible to it. Over the years people with sickle trait migrated to other continents. Sickle cell disease is seen predominantly in the black population but is also seen in other ethnic groups. These ethnic groups include individuals from parts of Middle East, Central India, and countries bordering the Mediterranean Sea, especially Italy and Greece.
Sickle Cell Anemia

Sickle Cell Anemia (SS) is an inherited blood disorder (autosomal recessive). Approximately one in 400 black babies are born with Sickle Cell Anemia, and about one in 11 have Sickle Cell Trait (AS).

The two hemoglobin types inherited will determine the shape of the red blood cell (RBC). When both parents have Sickle Cell Trait, there is a 1-in-4 chance (25 percent) the baby will have normal hemoglobin (AA), a 50 percent chance the baby will have Sickle Cell Trait (AS), and a 1-in-4 chance (25 percent) the baby will have Sickle Cell Anemia (SS). These chances remain the same with each pregnancy.
Sickle Hemoglobin C Disease

Sickle Hemoglobin C (SC Disease) is a milder form of Sickle Cell Disease. The baby has inherited two (2) abnormal hemoglobins, hemoglobin S and hemoglobin C. Approximately one in 1,000 black babies are born with SC Disease. Hemoglobin C Trait (AC) occurs in about one in 40 black babies. If one parent has AS and the other AC, there is a 1-in-4 chance (25 percent) the baby will inherit AA, AS, AC, or SC Disease.

‘AS and AC are carrier states, not disease conditions.
Sickle Beta Zero Thalassemia (ST0) is clinically similar to Sickle Cell Anemia. STO occurs in approximately one in 10,000 black babies. STO is treated the same as Sickle Cell Anemia.

When one parent has Sickle Trait (AS) and the other parent has Beta Thalassemia Zero Trait (AT) there is a 1-in-4 chance the baby will have normal hemoglobin (AA), Beta Thalassemia Zero Trait (AT), Sickle Beta Zero Thalassemia (STO), or Sickle Trait (AS). These chances remain the same for each pregnancy.

Sickle Beta Plus Thalassemia (ST+) is the mildest form of Sickle Cell Disease. ST-I- occurs in approximately one in 4,000 black babies.

When one parent has Sickle Trait (AS) and the other parent has Beta Thalassemia Plus Trait (AT+), there is a 1-in-4 chance (25 percent) the baby will have normal hemoglobin (AA), Sickle Trait (AS), Beta Thalassemia Plus Trait (AT+), or Sickle Beta Plus Thalassemia (ST+). These chances remain the same for each pregnancy.
Hemoglobin C Disease

Hemoglobin C Disease, a condition found mostly in the black population (approximately one in 6,000), consists of a minor abnormality of the hemoglobin. When both parents have C Trait (AC), there is a 1-in-4 chance (25 percent) that the baby will have normal hemoglobin (AA) or Hemoglobin C Disease (CC), and a 50 percent chance the baby will have Hemoglobin C Trait (AC). These chances remain the same for each pregnancy.
RED BLOOD CELLS

Normal

Sickle Red Blood Cells
Sickle Red Blood Cells

It is the inheritance of the sickle genes that causes red blood cell (RBC) abnormality. All complications of Sickle Cell Disease can be traced to changes in the make-up of the RBC. Normal RBC’s are smooth surfaced, enabling them to change their shape to flow through small blood vessels. Under certain conditions (i.e., acidosis, dehydration, infection, and low oxygen, etc.) RBC’s containing Sickle Hemoglobin become rigid, elongated, and sickle shaped. Some RBCs sickle immediately, while others remain normal for hours before sickling. Most RBCs containing Sickle Hemoglobin can sickle and then unsickle. After repeated cycles of sickling and unsickling, the RBC’s become irreversibly sickled.

In Sickle C Disease, some RBCs are sickle cells. The C Hemoglobin forms slightly misshapened RBCs but they are of normal size and color. The C Hemoglobin tends to reduce the complications caused by the sickled cells.

In Sickle Beta Plus or Zero Thalassemia, a portion of the RBCs are sickle cells. The Thalassemia cells are paler than usual and too small. There is no Hemoglobin A present in Sickle Beta Zero Thalassemia. There is a small amount of Hemoglobin A present in Sickle Beta Plus Thalassemia which tends to minimize the complications caused by the sickled cells.

Blood Flow

Sickled RBC's can become trapped within the blood vessels and thus interfere with normal blood flow. This obstruction can lead to sudden pain anywhere in the body as well as cause damage to body tissues and organs over time.
INFECTIONS

Infection is the major cause of death in children with Sickle Cell Anemia under the age of five years.

The spleen functions as part of the body’s defense against infection by serving as a filter to remove bacteria from the blood stream. The sickle RBC’s damage the spleen by about four months of age so that the spleen does not function normally. This can allow bacteria to grow in the blood stream and cause septicemia, which can be fatal. Children under the age of five years are at highest risk for septicemia.

Streptococcus Pneumoniae, (also called the pneumococcus) and Hemophilus influenzae are the two bacteria most likely to cause septicemia in the child with Sickle Cell Anemia, Ninety percent (90 percent) of the infections occur before the age of three years. Thirty-five percent (35 percent) of children with Sickle Cell Anemia who get pneumococcal sepsis die from the infection.

Signs & Symptoms

- Fever* - 102 F degrees or higher
- Coughing
- Vomiting and or Diarrhea
- Crankiness
- Rapid breathing
- Pale Color
- Unusual sleepiness
- Trouble Breathing

• A fever may be the ONLY initial sign of septicemia

Other potentially serious infectious which are more likely to occur in the child with Sickle Cell Anemia are meningitis, pneumonia and osteomyelitis.

Any infection in the child with Sickle Cell Disease is an emergency. Infection is treatable and complete recovery is possible only if it is recognized and treated early enough. However, even with treatment, permanent disabilities and even death can result.

Penicillin is often prescribed prophylactically twice daily to help fight infection. Septicemia can still occur even if penicillin is taken regularly.
The child with SC Disease or Sickle Beta Pills Thalassemia is not at as high a risk for septicemia as the child with Sickle Cell Anemia or Sickle beta Zero Thalassemia. Penicillin is not always recommended for these children.

Pneumococcal and Hemophilus influenza vaccine should be given to children with Sickle Cell Disease to help boost their immunity.
The spleen is normally a small organ located on the upper left side of the abdomen under the rib cage. When sickle cells are trapped in blood vessels inside and leading out of the spleen, the normal flow of the blood is blocked. Blood stays inside of the spleen instead of flowing through it. This is called sequestration. As a result, the blood count falls and the spleen gels very large and is easy to feel,

If the spleen suddenly enlarges with a significant drop in the blood count, this is a serious and potentially life-threatening problem. When the spleen gradually gets larger over several weeks, the blood count does not change much, so it is not as serious. Any enlargement of the spleen is of concern and must be watched for
changes. Parents should know how their child’s spleen normally feels, so that whenever the child seems sick they can check the spleen to see if it is bigger. If the spleen suddenly becomes larger, the child should be checked by a physician immediately. If the blood count is dangerously low from sequestration, blood transfusion may be necessary. Recurrent episodes are common, and a splenectomy (removal of the spleen) is sometimes required.

Babies and young children with Sickle Cell Anemia are at greatest risk of splenic sequestration. After age five years, the spleen becomes smaller and in most cases it cannot enlarge any more. Children with Sickle C Disease usually experience this complication after the age of five years.
PAIN

Painful episodes are common complications in children with Sickle Cell Disease. When the sickled cells are unable to flow through small blood vessels they obstruct blood flow causing vascular occlusion (vaso-occlusion). Vaso-occlusion reduces blood flow to an area of the body resulting in pain. This can occur anywhere in the body, including fingers, arms, legs, ribs, abdomen, and organs such as the spleen, brain, and eyes.

During infancy, vaso-occlusive crises (VOC’s) are generally manifested as dactylitis or hand-foot syndrome. This is characterized by soft tissue swelling, warmth and/or pain in the hands and/or feet due to ischemia (decreased oxygen) in
these small bones. Dactylitis can be recurrent but usually does not occur after two or three years of age. The most common sites of pain in children over two years of age are the long bones, joints, back, and abdomen.

VOC’s can vary in duration, intensity, location, and time between episodes. They can be mild, moderate, or severe in terms of pain. Sometimes swelling is seen in the area of pain. VOC’s may be preceded by a fever, dehydration, trauma, swimming, exposure to cold and/or emotional stress and unknown factors. Infection may occur at the same time.

**MANAGEMENT OF PAIN**

Painful episodes can often be treated and managed at home with regular acetaminophen (Tylenol), ibuprofen, or acetaminophen with codeine and hydration (an extra two to four ounces of water or juice every hour). Rubbing or application of heat with a heating pad or hot water bottle to the painful area may also alleviate discomfort.
Sometimes pain is unresponsive to home therapy. During those times, the child should go to the emergency room for intravenous (IV) hydration and pain medication. IV morphine is usually used. Most of the time this is adequate treatment so that home therapy will then be effective.

In very severe painful crises, the child may need to be admitted to the hospital for IV therapy. Sometimes the child can be taught to give his or her own pain medication by vein. This is called patient controlled analgesia, or PCA.
CHEST SYNDROME

Chest syndrome is a common cause of hospitalization in children with sickle cell disease. It is clinically similar to pneumonia. Chest syndrome can be fatal in the child with sickle cell disease.

Chest syndrome is the result of sickling in the lungs. It is believed that sickled cells clump up in the small blood vessels in the lungs or move there from some place else in the body. This may be triggered by a lung infection like pneumonia. Chest syndrome may develop right before, during, or after an episode of pain in the abdomen or bones.

Signs & Symptoms

- Chest pain
- Fast breathing and/or retractions
- Congested ‘pneumonia-like cough
- Abdominal pain
- Fever
- Trouble breathing

Treatment

Analgesia, hydration, antibiotics and oxygen are used. Sometimes a blood transfusion is necessary if the blood count or if the chest syndrome is severe.
An aplastic crisis results from an infection caused by Parvovirus B19. It causes production of RBC’s to be shut down for about 10 days. This means that RBC’s are not being made during this period. Because the RBC’s in children with Sickle Cell Anemia live only 10 to 15 days (compared to 120 days in children who do not have Sickle Cell Anemia), the blood count (hemoglobin and hematocrit) drop very rapidly to a dangerously low level during the infection.

Aplastic crisis usually occurs in children under the age of 16 years. It occurs in the general population but can only be noticed in those people with chronic hemolytic anemia (e.g. Sickle Cell Anemia). Recurrences of aplastic crisis are rare.
Treatment

Most often a blood transfusion is given to raise the blood count until the body can start making its own RBC’s again. Brothers and sisters with Sickle Cell Anemia should have their blood count checked since Parvovirus B19 is very contagious and they may be at risk of an aplastic crisis too.

STROKE

A stroke is a sudden and severe complication of sickle cell anemia. The most common cause of a stroke in children with sickle cell anemia is cerebral infarction (blockage of the oxygen supply to the brain by sickled cells). Strokes occur in six to 12 percent of individuals with sickle cell anemia, more commonly between three and 10 years of age.

A stroke may occur with a painful episode or an infection, but in most cases there are no related illnesses. Although recovery from a stroke may be complete in some cases, frequently the stroke can cause brain damage, paralysis, convulsions, coma and even death.

Repeat strokes occur in at least 60 percent of the children who have already suffered one stroke. A repeat stroke causes greater brain damage and increases the risk of death. To prevent recurrent strokes, blood transfusions are often given at four or five week intervals. It is not known how long these transfusions must be given to prevent another stroke from occurring.
GALLSTONES AND JAUNDICE

Gallstones in children with sickle cell disease are the result of elevated bilirubin excretion due to the increased hemolysis. Gallstones are found in about 30 to 50 percent of children with sickle cell anemia. They may be symptomatic or asymptomatic. Most physicians monitor the child with asymptomatic gallstones and do not recommend a cholecystectomy (removal of the gallbladder) until symptoms occur. Elective cholecystectomy may be indicated when gallstones are symptomatic (chronic right upper quadrant pain, nausea, vomiting, and fullness after meals).

The complications of gallstones can include passage of stones causing colic, common bile duct obstruction, cholecystitis, and rarely, pancreatitis.

Often the eyes of children with sickle cell anemia may appear yellow or jaundiced. This is due to the accumulation of a waste product (called bilirubin) from the increased RBC hemolysis associated with sickle cell disease. It is a benign complication.
GROWTH AND DEVELOPMENT - FEMALES

Females with sickle cell anemia maintain a lower average height and weight than those females with normal hemoglobin. This lower than average height and weight continues until late adolescence.

Puberty is usually delayed by several years. Menarche (beginning of the menstrual period) is also delayed. It is important to reassure the adolescent that she will eventually catch up with her peers.
GROWTH AND DEVELOPMENT - MALES

Males with sickle cell anemia maintain a lower average height and weight than those males with normal hemoglobin. This lower than average height and weight continues until late adolescence.

Puberty is usually delayed by several years. It is important to reassure the adolescent that he will eventually catch up with his peers.
Retinopathy can occur in children with sickle cell disease. Sickle cells can damage blood vessels in the retina and vision can be affected. Retinopathy is more common in adolescents with Sickle Hemoglobin C Disease. It is recommended that after the age of 10 years, children with sickle cell disease have periodic ophthalmology examinations.
PRIAPISM

Priapism is a persistent, unwanted erection of the penis that is often extremely painful. Priapism may present in the following ways:

**Stuttering**

There may be repeated, reversible, painful erections occurring over several hours (the penis becomes erect, the erection goes away, then becomes erect again, etc.). There are no problems with sexual functioning.

**Prolonged**

There may be a prolonged, painful erection that does not go away for more than several hours. This can last up to several days or weeks. This type of priapism needs attention by a doctor. Severe priapism can lead to partial or complete impotence. Sometimes a blood transfusion and liberal analgesics are given during the episode.

**Persistent**

There maybe a persistent penile erection that may last for weeks to years. This type of priapism is usually painless. It usually develops after a long episode of prolonged or stuttering priapism (as described above), Sexual functioning is often impaired.

Specific causes are unknown. Acute episodes often begin during sleep or following sexual activity, but frequently there is no identifiable event or cause. There is no current therapy to prevent episodes of priapism. There is no way to predict who will develop priapism and impotence. Those individuals experiencing repeated episodes are encouraged to avoid long periods of bladder distention, dehydration, and extended sexual activities.