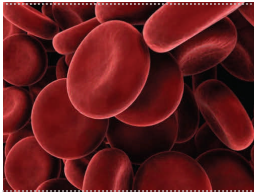


## Parents' Guide to Sickle Cell Disease (Hb SS Disease)

You have just learned that your baby has sickle cell disease (hemoglobin SS). The information below will help answer some of your questions. However, it should not take the place of an informed discussion with your baby's doctor (primary care provider).



### What is Hemoglobin?

Hemoglobin is a main ingredient in the red blood cells. Hemoglobin helps red blood cells carry oxygen from the lungs to all parts of the body and gives blood its red color. There are many hemoglobin types (this is not the same as a blood type). Hemoglobin A is the main type of hemoglobin present in the normal red blood cell. The presence of hemoglobin in red blood cells makes them smooth and shaped with a large rounded depression on each side of the cell, known as a biconcave disk shape (see picture on upper left). This shape allows normal red blood cells to be very flexible, moving easily through the blood vessels to deliver oxygen to the body.



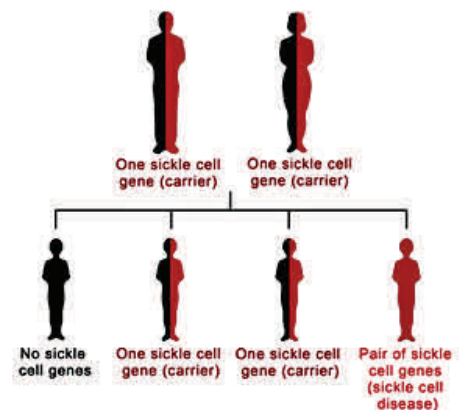
### What is Sickle Cell Disease?

Hemoglobin S causes the red blood cells to become sickle-shaped and is responsible for creating sickle cell disease (see picture on left). Children *inherit* this disease from their parents as a recessive genetic disorder. This means one sickle cell gene (hemoglobin S) is passed from *both* mom and dad to the baby causing sickle cell disease, which has two sickle cell genes (hemoglobin SS). When both parents have one hemoglobin S gene and one normal hemoglobin A gene, there is a 1 in 4 or 25% chance with each pregnancy that a child will inherit two hemoglobin S genes, one from each parent, and have sickle cell disease. These are the possible outcomes with each pregnancy (see picture below).

- 25% (1 in 4) chance of having a child with hemoglobin A (normal) (A/A)
- 50% (2 in 4) chance of having a child with sickle cell trait\* (A/S)
- 25% (1 in 4) chance of having a child with hemoglobin SS disease (S/S)

*\*People with sickle cell trait do not have sickle cell disease. They cannot develop these disease later in life. They can pass the trait on to their children.*

Sickle cell disease (hemoglobin SS disease) is found in males and females equally and occurs in all races. The disease is most common among people of African, Caribbean, Asian, Southeast Asian, Mediterranean, and Central and South American descent. All newborn babies in Utah are tested for sickle cell disease, regardless of their race or ethnic background. Sickle cell disease is a lifelong condition and is not contagious.



### What Problems can Sickle Cell Disease Cause?

In certain situations the hemoglobin SS may sickle. Sickling is a condition in which the red blood cells become hard and stick to and clog up small blood vessels. Those with sickle cell disease may have periods of feeling well and times of sudden intense pain that can be anywhere in the body. Damaged (sickled) red blood cells cannot deliver oxygen to the body and over time may damage the body's tissues and organs.

Sickled red blood cells are not very flexible in moving through the blood vessels and may rupture. They have a life span that is shorter than normal red blood cells. (Normal blood cells live about 120 days). This leads to anemia (low red blood cell count) and decreases the ability of red blood cells to hold onto oxygen.

The remains of ruptured red blood cells in the blood produce bilirubin, which can cause jaundice (yellow skin). Children with sickle cell disease sometimes have serious health problems, if not treated, that can lead to death. These problems are: *infection of the blood* (septicemia) and *sudden enlargement of the spleen with a drop in the blood cell count due to trapping of a large amount of blood in the spleen* (acute splenic sequestration). Other serious problems include: sudden pain, swelling of hands and feet, fever, increased infections, anemia, chest pain and trouble breathing, pneumonia, stroke, blood in urine, leg ulcers, gallstones, vision problems, organ damage, kidney failure, painful erections, and problems during pregnancy.

### ***What can be done to Treat Sickle Cell Disease?***

If your child who has been diagnosed with sickle cell disease (SS) is having any of the symptoms listed above, you should first consult your child's doctor for instructions.

#### **■ Pain Management**

Recommended home treatment includes applying a heating pad to the painful area and medicines such as acetaminophen (Tylenol®) or ibuprofen (Advil® and Motrin®). Drinking plenty of liquids and rest can help. Once the pain goes away children are usually active again. If the pain is not better after taking medicine at home, your child may need treatment with stronger medicine in the emergency room or to be admitted to the hospital for treatment.

#### **■ Penicillin**

Very serious infections of the blood sometimes occurs in infants and young children with sickle cell disease. Penicillin prescribed by your child's doctor taken two times a day can help decrease these infections in your child. Keeping your child's immunization up-to-date will significantly decrease the change of getting severe infections.

### ***What are the Most Important Things to Remember ?***

- Work closely with your child's doctor and hematologist (a doctor who specializes in blood disorders). Make sure your child has regular checkups and call your child's doctor with questions.
- Your child should avoid extreme hot and cold temperatures. He/she should avoid becoming exhausted, get plenty of rest, and drink lots of water to reduce the chances of having pain.
- Check your child's spleen as advised by the hematologist. Your child's doctor will show you where the spleen is and what feels normal. If the spleen suddenly feels larger, your child should be seen as soon as possible by his or her doctor.
- Your child will need an immediate medical evaluation for a fever of 38.5° C (101° F) or greater, difficulty breathing or chest pain. Take your child to a facility that provides emergency care.
- If your child has no energy and looks very pale he or she should be evaluated by a doctor.
- Be sure your child receives *all* childhood immunizations when they are scheduled and any additional immunizations recommended by your child's doctor.
- Call your child's doctor if you have questions and have your child seen if you have medical concerns.

### ***What Additional Treatment are Available?***

Blood transfusions and medications to decrease or prevent the formation of sickle-shaped red blood cells may be used. Sickle cell disease in extreme severity may be treated by a bone marrow transplant from a compatible donor, but is still considered a high-risk procedure.

### ***How Do I Get More Information?***

Talk with your baby's doctor. We recommend that you make an appointment with a pediatric hematologist in the near future. You may also want to have a genetic consultation for you and your family to see how sickle cell disease might affect future children or grandchildren.

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