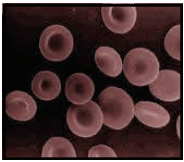


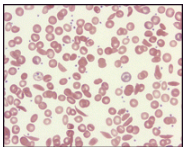
Parents' Guide to Sickle Cell Hemoglobin C Disease (SC Disease)

You have just learned that your infant has sickle cell hemoglobin C disease (hemoglobin SC). Naturally you are concerned and have many questions. This information sheet 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).



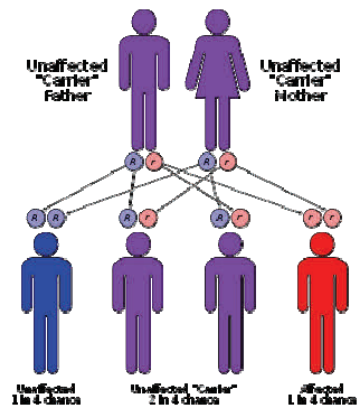
What is Hemoglobin?

Hemoglobin is a protein in the red blood cells. It carries 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 is inherited through genes, one from each parent. Most people have hemoglobin A (also called A Adult or normal hemoglobin) and a small amount of both hemoglobin A₂ and F (F Fetal), called minor hemoglobins. The presence of hemoglobin A 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). 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 Hemoglobin C Disease (Hemoglobin SC)?

Sickle cell hemoglobin C disease, also called SC disease (hemoglobin SC) occurs when a person has both hemoglobin S and hemoglobin C and no normal or adult hemoglobin. Children *inherit* this disease from both parents as a recessive genetic disorder. This means a sickle cell (hemoglobin S) gene is passed down from *one* parent and a hemoglobin C gene from the *other* parent causing SC disease. There is a 1 in 4 or 25% chance with each pregnancy that an infant will inherit two abnormal hemoglobin genes (S and C). Sickle cell hemoglobin C disease is found in males and females equally and occurs in all races. The disease is most common among in people of African, Caribbean and South American ancestry. It is less common in Mediterranean and Middle Eastern people. All newborn babies in Utah are tested for SC disease, regardless of their race or ethnic background. Sickle cell hemoglobin C disease (SC disease) is not contagious.



Persons with SC disease will always have it lifelong. Hemoglobin C interacts with hemoglobin S. This leads to a mixture of hemoglobins with the potential to sickle in certain circumstances, similar to the situation when an S hemoglobin interacts with another S, D, or O hemoglobins. All produce a mixture which has potential to sickle. As a result SC disease causes problems like sickle cell disease (SS disease) does. Those with SC disease may have periods of feeling well and times of sudden intense pain that can be anywhere in the body. This happens when sickled red blood cells (SC) become hard and sticky and clog up small blood vessels. Some hemoglobin SC red blood cells become rod-shaped with straight edges, because six-sided crystals form inside in the red blood cell (called hemoglobin C crystals). Damaged red blood cells cannot deliver oxygen to the body and over time may damage the body's tissues and organs.

What Problems can Sickle Cell C Disease Cause?

Hemoglobin SC red blood cells are not very flexible in moving through the blood vessels. The sickle C (SC) red blood cells sickle and 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 cell to hold onto oxygen. Bilirubin is produced from the remains of the ruptured cells which, in turn, can cause jaundice. Children with SC disease sometimes may have serious health problems that, if not treated, can lead to death. These problems are: *infection of the blood* (septicemia) and *sudden enlargement of the*

spleen with a drop in the red blood cell count due to trapping of a large amount of blood in the spleen (acute splenic sequestration), a condition that occurs more often in SC disease than in SS disease, due to the usually larger spleen in SC disease. 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, yellow skin (jaundice), organ damage, kidney failure, painful erections, and problems during pregnancy.

What can be done to Treat Sickle Cell C Disease?

If your child with sickle cell hemoglobin C disease (SC) is having any of the symptoms listed above, parents should first consult the 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 hemoglobin SC cell disease. Penicillin prescribed by your child's doctor taken two times each day can help decrease these infections in your child. Keeping your child's immunizations up-to-date will significantly decrease the chance 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 with them and call your child's doctor with questions.

■ Your child should avoid extreme hot and cold temperatures and exhaustion, get plenty of rest, and drink lots of liquids to reduce his or her 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. Those with severe SC disease may be treated by a bone marrow transplant from a compatible donor, but it 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 hemoglobin SC disease might affect future children or grandchildren.

Page 2 of 2

Utah Department of Health

44 N Mario Capecchi Drive
PO Box 144710
Salt Lake City UT 84114-4710

Phone: 801.584.8256
Fax: 801.536.0966
www.health.utah.gov/newbornscreening



Newborn
Screening Program