Hemoglobinopathies

What are hemoglobinopathies?
Hemoglobinopathies are conditions that affect the kind and amount of hemoglobin a person has in their red blood cells. Hemoglobin is the part of the red blood cell that carries oxygen throughout the body and gives the red blood cell its color and shape. Sickle cell anemia is the most common hemoglobinopathy. Sickle cell anemia is an inherited condition that causes misshaped (sickle) red blood cells. When these hard and pointed red blood cells go through small blood vessels, they clog the flow and break apart. This can cause pain, delayed growth, a low blood count (anemia) and other serious health problems.

About one in 350 African Americans are born with a hemoglobinopathy in the United States. Hemoglobinopathies can be found in all ethnic groups but are more common in individuals of African American, Arabian, Greek, Maltese, Italian, Turkish and Indian ancestry.

How does sickle cell anemia affect a child?
The symptoms of this condition vary from person to person and also vary depending on the form of sickle cell anemia. The most common form of this condition is known as sickle cell anemia (SS). Infants and children with sickle cell anemia generally have symptoms that include pain episodes, delayed growth, a low blood count (anemia), jaundice, gallstones and stroke. There can also be damage to most organs including the kidneys, liver and spleen. Damage to the spleen makes individuals with sickle cell anemia easily overwhelmed by bacterial infections.

What causes sickle cell anemia?
Sickle cell anemia is a genetic condition caused by a change in the HBB (hemoglobin beta) gene. A change in the HBB gene results in the production of structurally abnormal hemoglobin, called Hb S. The Hb S causes changes in the shape of the red blood cells to a sickle shape (or crescent moon). These sickle shaped red blood cells are responsible for the symptoms and complication of sickle cell anemia. There are also less common and less severe changes that can produce beta thalassemia hemoglobins (Sβ− or Sβ0), hemoglobin C (Hb C), hemoglobin D (Hb D), hemoglobin E (Hb E) or hemoglobin O (Hb O). These different hemoglobins also affect the shape of red blood cells and lead to different forms of hemoglobinopathies.

Sickle cell anemia is inherited in an autosomal recessive pattern, which means two copies of the HBB gene must be changed (Hb S) for a person to have sickle cell anemia (SS). The parents of a child with an autosomal recessive condition are “carriers” (AS) of one copy of the changed gene (S) and one copy of the normal gene (A). The carriers of sickle cell anemia are referred as having “sickle cell trait”. Individuals that have “sickle cell trait” may experience some of the symptoms associated with sickle cell anemia.

When both parents are carriers (AS), there is a one-in-four (or 25 percent) chance that both parents will pass the changed HBB gene (Hb S) on to a child, causing the child to be born with the sickle cell anemia (SS). There also is a one-in-four (or 25 percent) chance that both parents will pass on the normal HBB gene (Hb A), and the child will be free of the condition (AA). There is a two-in-four (or 50 percent) chance that a child will inherit a changed HBB gene (Hb S) from one parent and a normal HBB gene (Hb A) from the other, making them a carrier (AS) like their parents. These chances are the same in each pregnancy with the same parents.
**Is there a test for hemoglobinopathies?**
Yes. Babies are tested (newborn screening) for hemoglobinopathies before they leave the hospital. The baby’s heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to find out what the type of hemoglobin is in the blood.

**Can sickle cell anemia symptoms be prevented?**
Comprehensive care can reduce some of the complications associated with sickle cell anemia. With newborn screening, we are able to identify babies with sickle cell anemia early and start antibiotic treatment to help prevent most serious infections. It is also very important for infants and children with sickle cell anemia to receive regular childhood vaccinations, yearly influenza (flu) vaccine, yearly hepatitis vaccine and periodic pneumococcal booster. There are also therapeutic drugs to help manage pain episodes, reduce damage to organs and reduce the risk of stroke. Consult with your regional genetic center and/or regional sickle cell center concerning treatment recommendations.

**DISCLAIMER:** The information contained on this page is not intended to replace the advice of a genetic medical professional.

**Resources:**
- The Sickle Cell Information Center  
  PO Box 109  
  Grady Memorial Hospital  
  80 Jessie Hill Jr Drive SE  
  Atlanta, GA 30303  
  Phone: (404) 616-3572  
  Fax: (404) 616-5998  
  Email Address: aplatt@emory.edu  

- Information Center for Sickle Cell and Thalassemic Disorders  
  Kenneth R. Bridges, MD, Founder  
  Phone: (617) 768-8880  
  Email: kbridges@rics.bwh.harvard.edu  
  [http://sickle.bwh.harvard.edu/index.html](http://sickle.bwh.harvard.edu/index.html)

- Sickle Cell Disease Association of America, Inc.  
  200 Corporate Pointe, Suite 495  
  Culver City, California 90230-8727  
  Phone: (310) 216-6363  
  Fax: (310) 215-3722  
  General Public: (800) 421-8453  
  E-Mail: scdaa@sicklecelldisease.org  

- American Sickle Cell Anemia Association  
  10300 Carnegie Avenue  
  Cleveland, Ohio 44106  
  Phone: 216.229.8600  
  Fax: 216.229.4500
E-Mail: irabragg@ascaa.org
http://www.ascaa.org/

References:


- GeneTests (Sickle cell disease) [http://www.geneclinics.org](http://www.geneclinics.org)
