



## Hemoglobin Disorders (Hemoglobinopathies) Information for Parents

### ➤ Overview

Hemoglobin is a protein in the blood that carries oxygen from the lungs to all the tissues in the body. Most hemoglobin is made up of 2 parts, alpha globin and beta globin. The instructions for making the alpha globin and beta globin are contained in our DNA (which makes up our genes). There are many changes (or mutations) that can take place in these instructions. Often, these changes can result in a *hemoglobin variant*, which has no effect on the individual's health. Sometimes, however, the hemoglobin is changed enough that the health of the individual is affected.

### ➤ What is a hemoglobinopathy?

Hemoglobinopathy is a term describing a number of inherited disorders involving differences in the structure or amount of hemoglobin in the blood. If there is not enough hemoglobin in the body, or the hemoglobin is not formed correctly, oxygen cannot be carried through the body as efficiently. Abnormally low levels of healthy hemoglobin or red blood cells is called anemia. Depending on how much abnormal hemoglobin is being produced, symptoms can range from mild to life-threatening. Sickle cell anemia and thalassemia are examples of hemoglobinopathies.

### ➤ How are hemoglobinopathies inherited?

Hemoglobinopathies are inherited as autosomal recessive conditions. Most people have two complete sets of genes, one inherited from each of our parents. Genes are the instructions for growth and development of the body. In the case of recessive inheritance, two genes with mutations need to be inherited to have the condition. People with only one copy of a changed gene, also called a mutation, do not have the condition, but can pass the mutation on to their offspring. Individuals with one changed gene copy for a hemoglobin disorder are called *carriers*, or are said to have the *trait*. Hemoglobinopathies are inherited when both parents are carriers. With each pregnancy between two carriers, there is a 25% chance that the child will have a hemoglobin disorder, a 50% chance that the child will be an unaffected carrier, and a 25% chance that the child will not be a carrier nor have the disorder.

### ➤ Types of hemoglobinopathies

- **Sickle Cell Disease (Hemoglobin SS, SB<sup>0</sup>, SB<sup>+</sup>, or SC):** a serious condition in which most of the red blood cells are sickled (crescent-shaped), instead of their normal round shape. This sickle shape is the result of a mutation in the beta globin genes. The abnormal shape is less efficient at carrying oxygen. In addition, sickled cells have a tendency to get stuck in smaller blood vessels, causing blockages. People with sickle cell disease tend to get tired easily, have episodes of pain, called "sickle crises", and are prone to fevers and infections. Sickle cell anemia is more common in people of African descent.
- **Beta Thalassemia (Thalassemia Major/Intermedia/Minor):** a form of anemia in which the amount of beta hemoglobin is decreased. Depending on how serious the anemia is (Thalassemia Major), regular blood transfusions may be needed. It is more common in people with southern European (especially Greek and Italian), Middle Eastern, African and southeast Asian ancestry.
- **Alpha Thalassemia (Hemoglobin Barts):** occurs when one or more of the four genes responsible for alpha globin production is lost, resulting in a decrease in the amount of alpha globin. Depending on how many genes are missing, the individual can range from having no symptoms to severe, life-threatening anemia. Alpha thalassemia is most common among people of Asian descent.

➤ **Why is newborn screening done for hemoglobinopathies?**

Newborn screening is done so that babies with a hemoglobin disorder can be diagnosed quickly, and treatment, if needed, can be started.

➤ **Does a positive result from the Kansas Newborn Screening Lab mean that my baby has a hemoglobinopathy?**

No, not necessarily. Newborn screening tests the baby's level of different types of hemoglobin. The amount of these different types of hemoglobin in relation to each other may indicate that your child either is affected with a hemoglobin disorder or has the trait. Consultation with a specialist with experience in hemoglobin disorders can best determine if your baby needs treatment.

➤ **How common are hemoglobinopathies?**

Hemoglobinopathies are more common in certain ethnic groups, in particular, people of African, Asian, Arabic or Mediterranean descent. In the U.S., sickle cell anemia affects approximately 1 in every 250-600 African Americans. Beta thalassemia and alpha thalassemia are rare in the U.S., affecting less than 1 in 200,000 people. However, it is possible for two carriers of different hemoglobinopathies to have children with both traits; that is, two hemoglobin variants and no normal hemoglobin. This is called *compound heterozygosity*. The effect on the health of compound heterozygotes is variable, but can be significant.

➤ **What are the signs and symptoms of a severe hemoglobinopathy, such as sickle cell disease?**

Some of the more common signs and symptoms of a severe hemoglobinopathy include:

- fatigue, shortness of breath
- jaundice (yellow tint to skin and whites of eyes)
- slow growth, late puberty
- joint, bone and chest pain
- enlarged spleen and liver

➤ **How are hemoglobinopathies diagnosed?**

Hemoglobinopathies are diagnosed by a CBC (complete blood count) and gel electrophoresis testing to confirm the amounts of different kinds of hemoglobin. Follow up genetic testing on family members may also be performed.

➤ **Is there a cure for hemoglobin disorders?**

No, there is no cure, but new treatments and therapies have improved the quality of life for many patients with severe anemia, allowing them to live longer, more healthy lives.

➤ **How are hemoglobinopathies treated?**

- Many minor forms of thalassemia require no treatment.
- For more severely affected individuals, treatment may involve antibiotics to prevent infections, blood transfusions to increase the amount of normal hemoglobin in the body, and healthy nutrition to provide the best growth and development in the child.

**Where can I get additional information?**

1. American Sickle Cell Anemia Association (ASCAA)  
<http://www.ascaa.org/>
2. Thalassemia Patients and Friends: <http://www.thalassemiapatientandsfriends.com/>