SHOULD I UNDERGO CARRIER SCREENING TEST FOR HEMOGLOBINOPATHIES?

In all ethnic groups, there are specific hereditary conditions that are known to be more frequent. This information sheet will give you information about conditions seen more frequently in people of Mediterranean, Middle Eastern, Asian, Indian, and African ancestry.

What are hemoglobinopathies?

Hemoglobinopathies are a group of inherited diseases that affect the production of hemoglobin. Hemoglobin is the protein in red blood cells that carries oxygen to all parts of the body. The production of abnormal or deficient hemoglobin results in mild to severe anemia.

The two main hemoglobinopathies are *Thalassemia* and *Sickle cell disease*.

**Thalassemia** is due to reduced hemoglobin production and causes anemia due to fewer red blood cells than normal. There are two main types of thalassemia: *beta-thalassemia* (also called Mediterranean or Cooley’s anemia) and *alpha-thalassemia*. **Beta-thalassemia** can be very severe and manifest in the first two years of life. People with a severe form are often treated by monthly blood transfusions. The most severe form of **alpha-thalassemia**, which affects mainly individuals of Southeast Asian, Chinese and Filipino ancestry, results in fetal and newborn death. However, most individual with alpha-thalassemia have milder forms of the disease.

**Sickle cell disease** is due to the production of a variant hemoglobin that leads to hard, sticky, and crescent-shaped red blood cells. When these red blood cells go through the small blood vessels, they tend to get trapped, block the blood flow, and get destroyed. This can cause severe pain, damage to vital organs, risk of severe infections, and anemia.

How are hemoglobinopathies transmitted?

A severe hemoglobinopathy can be transmitted to a child when both parents are carriers of the condition. We all have two copies for each of our genes, one copy we received from our father and one from our mother. A person who is a carrier for a hemoglobinopathy possesses one defective copy of the gene responsible for the condition and one working copy. One working copy of the gene is enough to be healthy or have only mild symptoms of the disease. However, when two hemoglobinopathy carriers have children together, they have a ¼ (25%) risk to each transmit the defective copy of the gene and have a child with a severe hemoglobinopathy. Individuals who are carriers for thalassemia or sickle cell disease are often said to have the trait.
What is the chance that I am a carrier?

Your chance of being a carrier for one of the hemoglobinopathies depends on your family history and also on your ancestry. If you do not have a family history of thalassemia or sickle cell disease, then your risk of being a carrier is the same as the general population.

The following table gives you the approximate risk of being a carrier for thalassemia and sickle cell disease according to your ancestry. These numbers, taken from March of Dimes Genetic Screening pocket facts, may not be exact.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>beta-thalassemia trait</th>
<th>alpha-thalassemia trait</th>
<th>Sickle-cell trait</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mediterranean</td>
<td>1/20-1/30</td>
<td>1/30-1/50</td>
<td>1/30-1/50</td>
</tr>
<tr>
<td>Middle Eastern</td>
<td>1/50</td>
<td>variable</td>
<td>1/50-1/100</td>
</tr>
<tr>
<td>Southeast Asian</td>
<td>1/30</td>
<td>more than 1/20</td>
<td>rare</td>
</tr>
<tr>
<td>Asian</td>
<td>1/50</td>
<td>1/20</td>
<td>rare</td>
</tr>
<tr>
<td>Indian, Pakistani</td>
<td>1/30-1/50</td>
<td>variable</td>
<td>1/50-1/100</td>
</tr>
<tr>
<td>African American</td>
<td>1/75</td>
<td>1/30</td>
<td>1/12</td>
</tr>
<tr>
<td>Non-Hispanic Caribbean/West Indian</td>
<td>1/50-1/75</td>
<td>1/30</td>
<td>1/12</td>
</tr>
<tr>
<td>West African</td>
<td>1/50</td>
<td>1/30</td>
<td>1/6</td>
</tr>
<tr>
<td>Hispanic Caribbean</td>
<td>1/75</td>
<td>variable</td>
<td>1/30</td>
</tr>
<tr>
<td>Hispanic Mexican/Central American</td>
<td>1/30-1/50</td>
<td>variable</td>
<td>1/30-1/200</td>
</tr>
<tr>
<td>Caucasians/other populations</td>
<td>rare</td>
<td>rare</td>
<td>rare</td>
</tr>
</tbody>
</table>

Your risk of having a child with a hemoglobinopathy would be your risk to be a carrier times the risk of your spouse to be a carrier times ¼, which is the risk to have an affected child when both parents are carriers.

Example: 1/25 X 1/30. X 1/4 = 1/3000

How can I find out if I am a carrier?

Through a blood draw for CBC and hemoglobin electrophoresis, your doctor and us can interpret if you are a carrier of a hemoglobinopathy trait.

Why would I want to find out if I am a carrier?

The advantage of undergoing carrier screening is to find out if you are at increased risk for having a child with a form of hemoglobinopathy. Furthermore, if both you and your spouse are found to be carriers, you may have the option of undergoing prenatal diagnosis to find out the status of the baby. There are couples for whom it is important to know this information because they want to have time to prepare to the eventuality of having a child with a hemoglobinopathy and others who would consider an abortion for severe forms. What you would do with this information is very personal. There are also couples who do not want to know if they are carriers.
When should I undergo hemoglobinopathies carrier screening?

It is best to do the test before you are pregnant because it gives you more options if you and your spouse are found to be carriers. If you are already pregnant, the test should be done as soon as possible.

What should I do if I want to undergo hemoglobinopathies carrier screening?

First, we recommend that you check with your referring physician whether this test has been done already or not. Some physicians will include this test with the routine blood work that you do in the first trimester of the pregnancy. It may also have been at a previous pregnancy. This test needs to be done only one time.

If this test has not been done, you can arrange for a blood draw through your physician’s office. If the service is not available at his/her office, we can arrange this test for you.

What if I am found to be a carrier?

If you are found to be a carrier, your spouse should be tested as soon as possible. Remember that the risk is high to have an affected child only if both you and your spouse are carriers. We are available to meet with you again in a genetic counseling session to review the implications of your results and your options.

If you are interested in more information about thalassemia and sickle cell disease, you can also visit the following Websites:

- www.thalassemia.com
- www.cooleyasanemia.org
- www.thalassaemia.org.cy
- www.sicklecellinfo.net
- www.ascaa.org
- www.sicklecelldisease.org