If a person or their family is from a country in the *Mediterranean, Middle East, Asia or Africa,* they have the highest chance of being a carrier of *thalassemia.* If a person is from a country in the *Caribbean, and South and Central America*, then the chance of being a carrier is also higher than average.

If your family is from these regions of the world, you might wish to consider carrier testing for a hemoglobinopathy. This testing can be done through your doctor's office, or it can be organized through your regional Genetics centre. The testing should include both a complete blood count (CBC) **and** a hemoglobin electrophoresis. Both of these are simple blood tests.

If you have more questions, please contact the Genetics Clinic at The Credit Valley Hospital by calling:
905-813-4104
or
1-877-292-4284 (toll free)
and ask the switchboard to transfer you to Genetics.

Your genetic counsellor's name is

_____

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Information about Hemoglobinopathy Screening

at

The Credit Valley Hospital

Genetics Clinic

What are hemoglobinopathies?

Hemoglobin is part of a red blood cell. Hemoglobin carries oxygen around the body. The word “hemoglobinopathy” means a problem with the hemoglobin. Hemoglobinopathies can affect the number of red blood cells or how well the hemoglobin carries oxygen around the body. Hemoglobinopathies are the most common genetic conditions in the world.

How do hemoglobinopathies affect people?

There are different types of hemoglobinopathies. Each one has different effects. If the red blood cells do not work properly, then there might be medical problems. Effective treatments are available for most of these problems. For example, a person with a hemoglobinopathy might need blood transfusions on a regular basis. Others must take medicine daily. When treatment is available, many people with a hemoglobinopathy have active and productive lives. For some less common types, effective treatment might not be available.

What are the common types of hemoglobinopathies?

There are two common types of hemoglobinopathies, called **sickle cell anemia** and **thalassemia**. Thalassemia is further divided into *two* types, *alpha* thalassemia and *beta* thalassemia.

How do hemoglobinopathies happen?

A person is born with a hemoglobinopathy. Hemoglobinopathies happen because of differences in a person’s genetic information. Each piece of genetic information is called a gene. Genes provide the instructions for a person’s growth and development. There are many different genes. Specific genes are responsible for making hemoglobin.

Genes come in pairs. We get one copy of each gene from each of our parents. Usually, both copies of a gene are working. If a person has one copy of a gene that works normally and one copy of a gene that does not work, they are called a *carrier.* A carrier of a hemoglobinopathy usually is healthy and does not have the disease.

When both parents are carriers and have a child together, each can pass on either the working or non-working copy of a hemoglobin gene. There is a 1 out of 4 (25%) chance a child will get two non-working copies of a hemoglobin gene. This child will have a hemoglobinopathy. There is a 2 out of 4 (50%) chance a child will be a carrier, like their parents. There is a 1 out of 4 (25%) chance a child will get two working copies of the gene. These chances are the same for each child when both parents are carriers. Boys and girls have the same chance of being affected by a hemoglobinopathy.

If only one parent is a carrier, they will not have a child with a hemoglobinopathy. In this case, each of their children will have a 1 out of 2 (50%) chance of being a carrier, like one of the parents.

Is there testing for hemoglobinopathies?

Yes. A blood test can be done to determine if a person is a carrier of a hemoglobinopathy. If both parents are carriers of a hemoglobinopathy, testing can be done in a pregnancy or after birth to see if a child has a hemoglobinopathy.

Can hemoglobinopathies be prevented or cured?

Hemoglobinopathies cannot be prevented but treatments are available for many of them.

Who could be a carrier of a hemoglobinopathy?

Anyone could be a carrier. However, in certain areas of the world, hemoglobinopathies are more common.

If a person or their family is from a country in *Africa, the Middle East*, or the *Caribbean,* they have the highest chance of being a carrier of *sickle cell anemia.* If a person is from a country in the *Mediterranean, South East Asia (including India, Pakistan, Bangladesh, Indonesia, Thailand and Myanmar) and South and Central America,* then the chance of being a carrier is also higher than average.

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