



WHAT ARE

HEMOGLOBINOPATHIES?

Hemoglobin is the part of a red blood cell that carries oxygen to all parts of the body. Hemoglobinopathies are a group of disorders that cause changes in the type or amount of the hemoglobin that is produced.

WHAT ARE THE COMMON TYPES OF HEMOGLOBINOPATHY?

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

These diseases are chronic or lifelong. In general, these diseases cause anemia that may require hospitalization or blood transfusions.

HOW ARE SICKLE CELL ANEMIA AND THALASSEMIA INHERITED?

Genes are the units of heredity that determine characteristics such as eye colour or blood type. We have two copies of each gene, one is from our mother and one is from our father. Both parents must be carriers of the gene that causes a particular hemoglobinopathy for there to be a chance of having a child affected with the disease.

WHAT IS A CARRIER?

A carrier is a person who has one working copy of the gene and one gene that does not work normally. Carriers do not show symptoms of the disease because their normal working gene makes enough hemoglobin for them to be healthy. Usually, it is impossible to know if you are a carrier without having a specific blood test. Individuals from certain ethnic origins

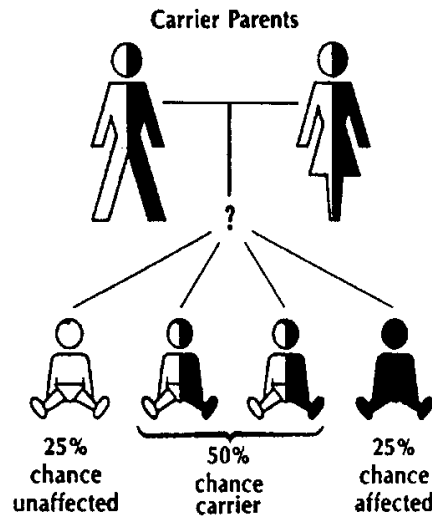
have an increased chance of being a carrier.

WHAT IS THE CHANCE THAT CARRIERS COULD HAVE A CHILD WITH THE DISEASE?

When both parents are carriers, they have an increased chance of having a child with two copies of the non-working gene leading to one of these diseases.

As you can see from the diagram, with each pregnancy the couple faces a 1 in 4 chance of having a child affected. They have a 2 in 4 chance of having a child who is a carrier and a 1 in 4 chance of having an unaffected child who is not a carrier.

WHO MIGHT BE A CARRIER OF A HEMOGLOBINOPATHY?



Although anyone could be a carrier, people whose ethnic (family) background is from certain areas of the world have a greater chance of being a carrier than others do. Sickle cell anemia is more common among people of African background including African-Americans, and Hispanics of Caribbean descent. People with families

from the Mediterranean, middle East, southeast Asia and Africa have an increased chance of being a carrier of thalassemia. If your family comes from any of these regions of the world, you might wish to consider testing for a common hemoglobinopathy.

HOW DO WE TEST FOR SICKLE CELL ANEMIA AND THALASSEMIA?

Screening tests can identify most but not all carriers. Therefore, a negative screen significantly reduces an individual's chance of being a carrier but can not give a 100% guarantee that they are not a carrier.

This screening test, which involves taking a blood sample, can be organized through your health care provider. He/she will order a complete blood count (CBC) including a mean corpuscle volume (MCV) and sickle cell screening. Depending on these results, a blood film exam and hemoglobin electrophoresis may also be necessary, to give more precise information about whether a person is a carrier or not.

It is important to be aware that every pregnant couple in the general population has a 2-3% risk of having a baby with a birth defect. While normal results of prenatal testing are reassuring, they do not guarantee the birth of a healthy baby.

