Information + Taking Control = Best Outcome

Hematology

Fast Facts Information Sheets for Patients

Thalassemia Screening

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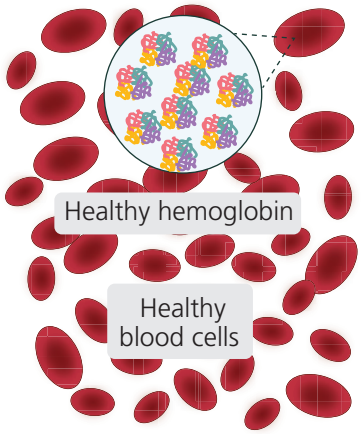
HEALTHCARE

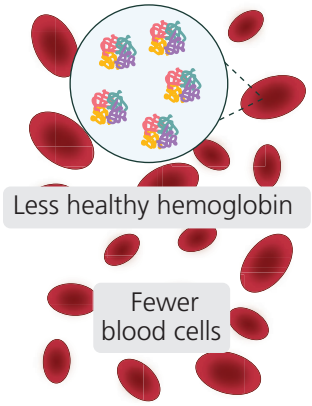
Karger

Thalassemia Screening

This leaflet informs you about thalassemia screening. Screening means doing laboratory tests to look for a condition before it is noticeable or to see if you are at risk of having a child who is affected with a condition.



Healthy blood



Anemic blood

What is thalassemia?

Thalassemia is a blood disorder that causes your body to make less hemoglobin. Hemoglobin is a protein in your red blood cells which transports oxygen from your lungs to the rest of your body. When your body makes less hemoglobin it leads to anemia. The organs in your body do not get enough oxygen and cannot work properly when you have anemia.

Thalassemia is more common in people from, or whose ancestors were from, Southeast Asia, South Asia, the Mediterranean region, the Middle East and Africa.

There are two main types of thalassemia: alpha and beta. There are also different kinds of alpha and beta thalassemia. Some types of thalassemia cause serious health problems.

What is a genetic disease?

Thalassemia is a genetic disease. This means it passes from parents to children through genes. If you have thalassemia, you have inherited a changed (mutated) gene from both of your parents. Healthy carriers often have no symptoms and will never develop the disease.

Why do I need to be tested?

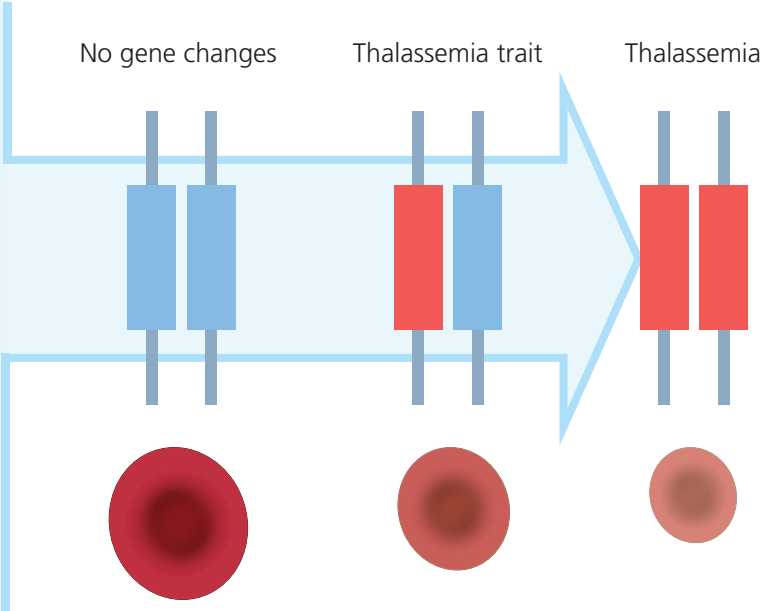
If you are a carrier, you have inherited one or two mutated genes. This is called **thalassemia trait**. You may have no symptoms or just mild anemia with smaller red blood cells.

You will never develop thalassemia.

When you have thalassemia trait, you can pass on the changed gene to your children. And if your partner also has thalassemia trait, your children may be born with thalassemia. This can be a very serious condition.

The only way to know if you are a carrier is to get tested.

Thalassemia trait and disease are not contagious.



How do I get tested?

Trait testing is quick and painless, and can usually be done with just one blood draw which is then sent to a laboratory for analysis. The tests that are done are usually:

- CBC – complete blood count to look at different components of your blood, including the size of red blood cells.
- Iron level (usually done with a test of your ferritin level)
- Hemoglobin analysis by HPLC – high-performance liquid chromatography – or sometimes electrophoresis.
- Genetic testing

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Thalassemia testing: pregnancy and birth

When will I have a thalassemia blood test?

If you are pregnant but have not been tested for thalassemia, you can still be tested. In many countries, thalassemia screening is a routine part of a pregnant woman's prenatal care. The test is usually done before you are 10 weeks pregnant. It is a quick and easy blood test.

What happens after the blood test?

If the first blood test shows that you are a carrier of thalassemia, the baby's father will be invited to have a test.

If the father is also a carrier, or if he is unavailable to take the test, your doctor will offer another kind of test called a **diagnostic test**.

The diagnostic test will tell you if your baby:

- has thalassemia disease
- is a carrier
- is not affected.

What is a diagnostic test?

This test is done by either:

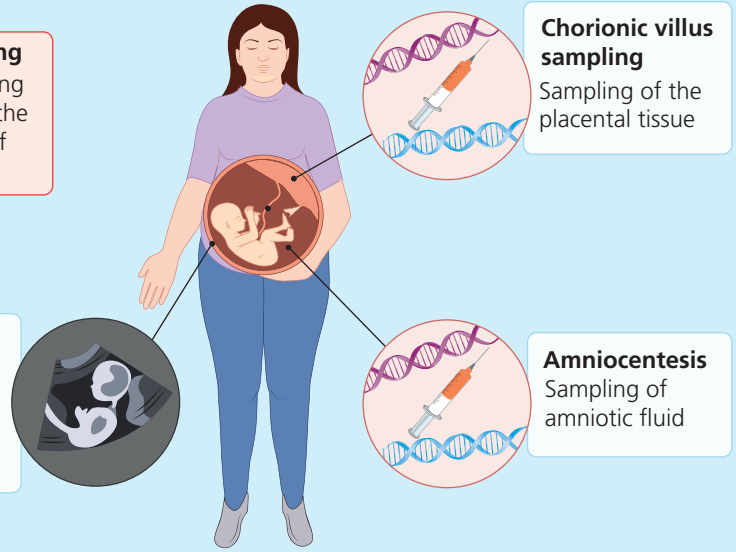
- taking a small sample of the placenta (chorionic villus sampling)

OR

- testing the fluid surrounding the baby (amniocentesis).

The choice of test depends on how far through the pregnancy you are.

Both tests carry a small risk of miscarriage, so your doctor will only suggest testing if it is absolutely necessary.



For more information about thalassemia

- ukts.org
- thalassemiapatientsandfriends.com/
- thalassemia.org
- thalassaemia.org.cy

The results of the test

If tests suggest that your baby may be born with severe thalassemia, a counselor may guide you through the difficult decision of whether to continue with the pregnancy. There is no single answer that suits every couple. The decision depends on many factors, including cultural, social, spiritual and religious beliefs. The aim of counseling is to help you and your partner to make a fully informed and independent decision.

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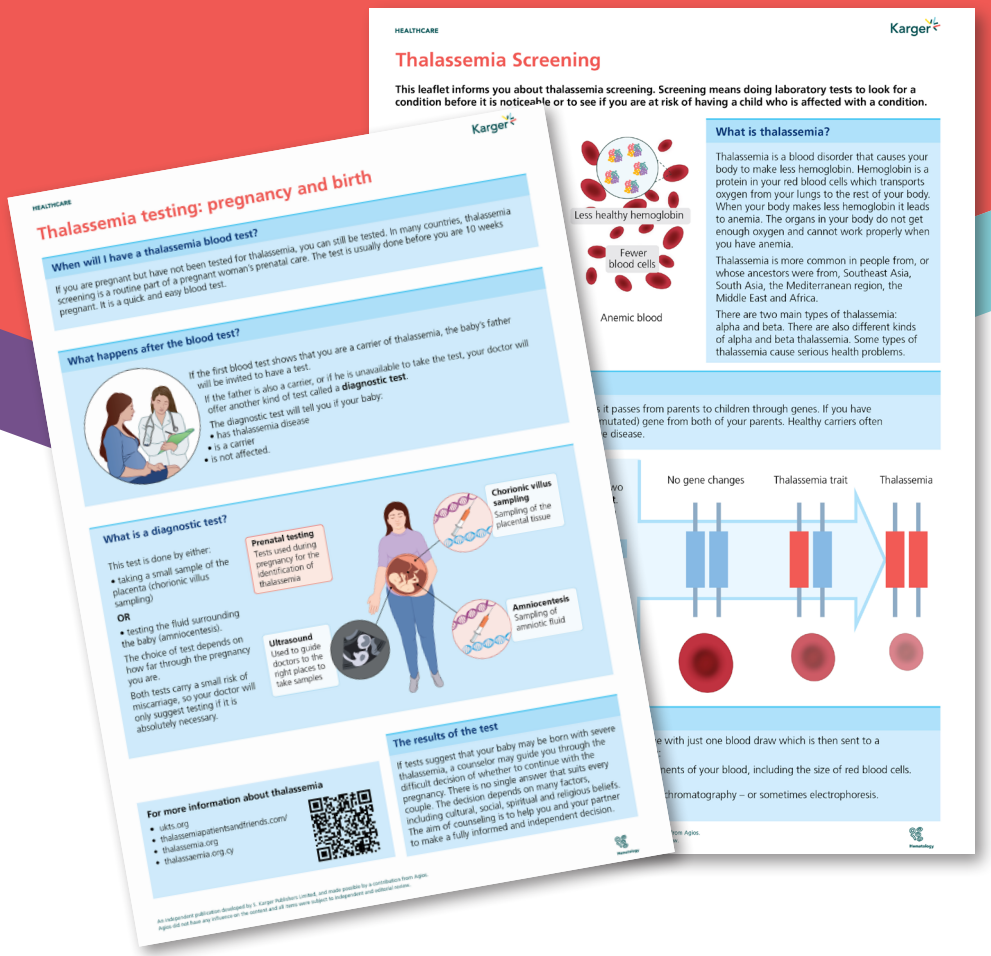

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