



Thalassemia & Pregnancy

What is thalassemia?

Thalassemia is the name of a group of inherited blood disorders. There are two main types of thalassemia: alpha and beta, in reference to the alpha and beta proteins that form hemoglobin in the blood. Hemoglobin is the oxygen-carrying component of red blood cells, so if the body doesn't produce enough of either of these two proteins, the result is anemia that begins in early childhood and lasts throughout life.

What are the effects of thalassemia?

Thalassemia ranges widely in severity. Babies born with thalassemia may have mild to severe anemia, may develop jaundice, organ damage or even die.

How is thalassemia transmitted?

Thalassemia is an inherited disease, i.e. it is passed on by parents who have the thalassemia gene.

Because the gene is recessive, both parents must each pass on the thalassemia gene in order for the baby to have the full disease.

If the baby only inherits one gene, s/he will become a carrier but not express the full disease. Sometimes this carrier state is referred to as "thalassemia trait". Most carriers lead normal, healthy lives. They may not even realize that they carry this gene.

When both parents are carriers:

- 1 in 4 chance that their child will inherit 2 thalassemia genes and have severe outcomes of the disease = **Thalassemia major**
- 2 in 4 chance that the child will inherit the thalassemia trait, i.e. become a carrier = **Thalassemia minor**
- 1 in 4 chance that the child will inherit 2 **normal** genes

How is thalassemia diagnosed?

When you do your routine blood test at the beginning of your pregnancy, one result we review is the Mean Corpuscular Volume, or MCV. The MCV reading determines the size of your red blood cells. For adults, if the MCV reading is less than 75 you may be a trait carrier. If your MCV reading indicates that you may have the thalassemia trait, additional blood tests can be performed to make sure.

Genetic counseling can also aid in identifying if you should have this blood test. Because thalassemia occurs most commonly among Mediterranean, Middle-Eastern, Asian, and African people, if you and your baby's father are from any of these groups, you may want to consider genetic counseling and/or further testing.

During pregnancy, chorionic villus sampling (CVS) or amniocentesis can detect or rule out thalassemia in the fetus. Early diagnosis is important so that treatment can prevent complications.

What is the treatment for thalassemia?

The use of frequent blood transfusions and antibiotics has greatly improved the outlook for children born with thalassemia. Still, there is a chance that these babies will be born with major organ damage and may not live a full life span.