



INFORMATION FOR PARENTS/GUARDIANS: My Baby Has a Positive Beta Thalassemia Newborn Screening Result

What is Newborn Screening?

These are routine tests done shortly after birth on every baby born in Ontario. A small sample of blood is taken from your baby and is tested for rare treatable diseases.

What does it mean if my baby has a positive β thalassemia newborn screening result?

This result does not mean your baby has β thalassemia but it does mean that your baby very likely has β thalassemia. More testing is needed to confirm the diagnosis. Babies with β thalassemia can be healthier if treatment begins early, so it is important to have follow-up testing done to find out if your baby truly has β thalassemia.

You may feel concerned or worried about your baby's screen positive result. Many parents in this situation feel this way.

What is β thalassemia?

β thalassemia is an inherited (genetic) disease that affects the amount of hemoglobin in the red blood cells. Hemoglobin is the protein in the red blood cell that carries oxygen to every part of the body. Hemoglobin is made up of different building blocks called the beta (β)-globin chains and the alpha (α)-globin chains. Babies with β thalassemia do not have enough of the beta building block to make normal amounts of hemoglobin.

If untreated, babies with β thalassemia develop serious anemia and may die in childhood.

Anyone can have β thalassemia; however, β thalassemia is more common in Mediterranean, Middle Eastern and African communities.

How do I find out if my baby has β thalassemia?

Blood tests are done to find out if a baby actually has β thalassemia.

When can my baby have these tests?

Your baby's doctor or a health care professional at a Newborn Screening Treatment Centre will call you to talk about the results of your baby's positive newborn screen and make arrangements for the follow up testing to happen as soon as possible.

Why screen for β thalassemia?

Screening means that babies with β thalassemia can be treated early with blood transfusions to prevent serious anemia. The goal of screening is to help people with β thalassemia live healthier lives.

How does a baby get β thalassemia?

β thalassemia is an inherited (genetic) disease that causes lower than normal levels of hemoglobin. Hemoglobin is made up of 4 building blocks, two alpha(α)-globin chains and two beta(β)-globin chains.





A baby with β thalassemia inherited a non-working copy of the β -globin gene from each parent (each parent is a “carrier” of β thalassemia). The parents may not be aware that they are carriers of β thalassemia because β thalassemia carriers do not have, and **will not develop**, β thalassemia.

Where can I get more information?

For more information on newborn screening, please visit the Parents section of our website at www.newbornscreening.on.ca or talk to your local health care provider.

For more information on β thalassemia, please visit the Thalassemia foundation of Canada website at www.thalassemia.ca or call 416-242-THAL (8425).

NOTE TO PARENTS/GUARDIANS: This information is only for parents whose baby has had a positive newborn screening result for β thalassemia. Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

