



INFORMATION FOR PARENTS/GUARDIANS: My Baby Has a Positive Newborn Screening Result for a Hemoglobin Disease

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 newborn screening result?

This result does **not** mean your baby has a hemoglobin disease. It means that more testing is needed because your baby **might** have a hemoglobin disease. Babies with hemoglobin diseases 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 a hemoglobin disease.

You may feel concerned or worried about your baby's screen positive result. Many parents in this situation feel this way. Remember, we do not know for sure that your baby has a hemoglobin disease until follow up testing is done.

What is hemoglobin?

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.

What are hemoglobin diseases?

Hemoglobin diseases are a group of inherited diseases that affect the hemoglobin in red blood cells and cause anemia (decreased amounts of red blood cells). The goal of newborn screening is to detect hemoglobins that cause disease.

Sometimes, variant (slightly different) hemoglobins are found. Most times, these variant hemoglobins do not cause disease and do not need treatment. However, some babies with variant hemoglobins have serious disease and do need treatment.

Variant hemoglobins are common. Anyone can have a variant hemoglobin or a hemoglobin disease.

How do I find out if my baby has a hemoglobin disease?

Blood tests are done to find out if a baby actually has a hemoglobin disease.

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 hemoglobin diseases?

Screening means that babies with a hemoglobin disease can be treated early by specialists called hematologists (doctors who specialize in blood diseases). The goal is to help people with hemoglobin diseases live healthier lives.





How are variant hemoglobins inherited?

Hemoglobin variants are inherited (genetic). A baby with a hemoglobin variant inherited a variant copy of the β -globin (or α -globin) gene from either their father or their mother. Sometimes, a baby inherits a variant hemoglobin from both parents. The parents are often not aware that they carry variant hemoglobins because variant hemoglobins often do not cause disease.

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.

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

