
INFORMATION FOR PARENTS/GUARDIANS: My Baby Has a Positive Homocystinuria 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 the heel of your baby and is tested for rare, treatable diseases, including homocystinuria.

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

This result does **not** mean that your baby has homocystinuria. It means that more testing is needed because your baby **might** have homocystinuria. Babies with homocystinuria are healthier if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has homocystinuria.

You may feel 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 homocystinuria until follow up testing has been done.

What is homocystinuria?

Homocystinuria is a rare inherited (genetic) disease that does not allow a baby to break down an amino acid (building block of protein) called homocysteine. Homocysteine is made from an amino acid called methionine which is found in most of the foods we eat, including breast milk and infant formula. Homocysteine is important for normal growth and development.

If the body can not break down methionine, harmful substances build up in the body. If untreated, this build up can lead to serious and permanent health problems like poor growth, eye problems, bone problems, life-threatening strokes and mental retardation.

Some people with homocystinuria have a milder form which can be treated with vitamin B6. However, newborn screening can not tell who will have a milder form and who will have more serious problems.

Homocystinuria is very rare. It is thought to affect 1 in every 200 000 – 1 in every 300 000 babies born in Ontario.

What causes homocystinuria?

The most common cause of homocystinuria happens when the enzyme cystathionine beta-synthase (CBS) is either missing or not working properly. The job of this enzyme is to help the body break down methionine.

How do I find out if my baby has homocystinuria?

Blood and urine tests are done to find out if a baby who screened positive actually has homocystinuria.

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 arrange more testing as soon as possible.

Why screen for homocystinuria?

Babies who have homocystinuria look normal when they are born but will develop serious health problems and mental retardation if they are not treated. The goal of newborn screening for homocystinuria is to prevent severe mental retardation, life-threatening strokes and the other serious health problems caused by homocystinuria.



How is homocystinuria treated?

Babies with homocystinuria are treated and monitored by a team of health care specialists including a metabolic doctor and a dietician. The treatment is lifelong and can include large doses of vitamin B6, a special diet that is low in protein and a medical formula. Babies with homocystinuria are checked often for their amino acid levels, development and other health issues associated with homocystinuria.

How does a baby get homocystinuria?

Homocystinuria is an inherited (genetic) disease. A baby with homocystinuria inherited two non-working copies of the cystathionine beta-synthase (CBS) gene, one copy from each parent. People who have one non-working copy of the CBS gene are called "carriers." Carriers of homocystinuria are healthy, do not have, and will never develop, symptoms of homocystinuria.

Where can I get more information?

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

For more information on homocystinuria, please visit the Children Living with Metabolic Diseases website at www.climb.org.uk.

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

