My Baby Has a Positive Newborn Screening Result: Homocystinuria

What is newborn screening?
These are routine tests done shortly after birth on every baby born in Manitoba. A small sample of blood is taken from the heel of your baby and is tested for rare, treatable diseases, like homocystinuria.

What does it mean if my baby has a positive newborn screening test for homocystinuria?
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 will not know for sure if 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 homocysteine, one of the 20 building blocks of protein called amino acids. Homocysteine is important for normal growth and development. However in homocystinuria abnormally high levels of homocystine and other harmful substances build up in the body. If untreated, this build up can lead to serious and permanent health problems like poor weight gain, eye problems, bone problems, learning difficulties and strokes.

Some people with homocystinuria have a milder form which can be treated with vitamin B6. However, newborn screening cannot 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 Manitoba.

What causes homocystinuria?
The most common cause of homocystinuria happens when the enzyme cystathionine betasynthase (CBS) is either missing or not working properly. The job of this enzyme is to help the body break down homocysteine.

How do I find out if my baby actually has homocystinuria?
Blood and urine tests are done to find out if a baby who screened positive actually has homocystinuria.

This parent information brochure was developed using materials from and with the assistance of the Ontario Newborn Screening Program, located at the Children’s Hospital of Eastern Ontario.
When can my baby have these tests?
Your baby’s doctor or health care provider and a member of the metabolic team at the Winnipeg Children’s Hospital 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 like all other healthy babies when they are born but they will develop serious health problems and learning problems if they are not treated. The goal of newborn screening for homocystinuria is, by early detection of an affected baby and early treatment before symptoms develop, to prevent severe learning problems, potentially life-threatening strokes and the other serious health problems such as osteoporosis.

How is homocystinuria treated?
Babies with homocystinuria are treated and monitored by a team of health care specialists including a metabolic doctor, metabolic genetic counselor and a dietician. The treatment is lifelong and can include large doses of vitamin B6 and other vitamins, a medical formula designed to treat homocystinuria and when on solids a diet that is low in protein. Babies with homocystinuria are checked often for their amino acid levels their growth and development and other health issues associated are regularly monitored.

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 Winnipeg Metabolic Program website www.wrha.mb.ca/prog/genetics or the newborn screening section of the Cadham Provincial Laboratory website www.gov.mb.ca/health/publichealth/cpl/baby.html

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.