INFORMATION FOR PARENTS/GUARDIANS:
My Baby Has a Positive Galactosemia 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 galactosemia.

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

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

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 galactosemia until follow up testing has been done.

What is galactosemia?

Galactosemia is a rare inherited (genetic) disease that does not allow a baby to break down galactose (a form of sugar found in breast milk and many foods). Levels of galactose and other harmful substances build up in babies with galactosemia. If untreated, this build up can lead to serious and permanent health problems like poor growth, liver damage, kidney problems, cataracts, life-threatening infections and mental retardation.

Galactosemia affects about 1 in every 60 000 babies born in Ontario.

What causes galactosemia?

The most common cause of galactosemia happens when the enzyme galactose-1-phosphate uridyl transferase (GALT) is either missing or not working properly. This enzyme's job is to break down galactose into other sugars the body can use. If the GALT enzyme can not do its job, galactose levels become too high and cause health problems.

There are other, more rare forms of galactosemia that may not be picked up by newborn screening.

How do I find out if my baby has galactosemia?

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

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 galactosemia?
Babies who have galactosemia look normal when they are born but will develop serious health problems, mental retardation and sometimes die if they are not treated. Early signs of galactosemia can include feeding problems, jaundice (yellow colour to the skin and whites of the eyes) and a big liver. When galactosemia is diagnosed and treated early, severe mental retardation and many of the serious health problems can be prevented.

How is galactosemia treated?
Babies with galactosemia are treated and monitored by a team of health care specialists including a metabolic doctor and a dietician. Babies with galactosemia are given a special diet that is low in galactose as well as extra vitamins and minerals that are important for normal growth. Babies with galactosemia are checked often for their galactose levels, development and other health issues associated with galactosemia.

How does a baby get galactosemia?
Galactosemia is an inherited (genetic) disease. A baby with galactosemia inherited two non-working copies of the GALT (the enzyme that breaks down galactose) gene, one copy from each parent. People who have one non-working copy of the GALT gene are called "carriers." Carriers of galactosemia are healthy, do not have, and will never develop, symptoms of galactosemia.

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 galactosemia, please visit the Parents of Galactosemic Children, Inc. website at www.galactosemia.org

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

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