My Baby Has a Positive Newborn Screening Result: Galactosemia

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, including classical galactosemia.

What does it mean if my baby has a positive galactosemia newborn screening result?
This result does not mean that your baby has classical galactosemia. It means that more testing is needed because your baby might have classical galactosemia. Babies with classical 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. Most babies with a positive newborn screen for classical galactosemia prove to either have a mild variant that requires no long term treatment and babies with these variants generally have an entirely normal outcome. Other positive newborn screens may just be “false positives”.

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 classical galactosemia until follow up testing has been done.

What is galactosemia?
Galactosemia is a group of rare inherited (genetic) diseases that does not allow a baby to break down galactose (a form of sugar found in breast milk and many foods) ranging from classical to mild variants. 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 Manitoba.

What causes galactosemia?
The most common cause of galactosemia happens when the enzyme galactose-1-phosphate uridyl transferase (GALT) is either missing [classical galactosemia] or not yet fully active. The GALT enzyme’s job is to break down galactose into other sugars the body can use. If the GALT enzyme cannot do its job, galactose levels become too high and can cause health problems. There are other, rarer 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 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 galactosemia?
Babies who have classical 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 classical galactosemia can include feeding problems, jaundice (yellow colour to the skin and whites of the eyes) and a big liver. When classical 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 classical galactosemia are treated and monitored by a team of health care specialists including a metabolic doctor and a dietician. Babies with classical 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 classical galactosemia are checked often for their galactose levels, development and other health issues associated with galactosemia. Occasionally babies with a mild variant form of galactosemia may require restriction of lactose for the few months of life.

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

NOTE TO PARENTS/GUARDIANS: This information is only applicable if your 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.