My Baby Has a Positive Newborn Screening Result: Cystic Fibrosis

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 cystic fibrosis.

What does it mean if my baby has a positive newborn screening test for cystic fibrosis?
A positive screen for cystic fibrosis means that a molecule called immunoreactive trypsinogen (IRT) is elevated in the baby’s blood. This result does not mean that your baby has cystic fibrosis. It means that more testing is needed because your baby might have cystic fibrosis. It is important to diagnosis cystic fibrosis early so that if the baby needs specific treatments they can be implemented as soon as possible.

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 cystic fibrosis until follow up testing has been done.

What is IRT?
Manitoba currently uses a two-tier screening program for cystic fibrosis in which IRT is measured in the filter paper blood sample collected from the baby at 1-2 days of life. If the initial IRT is elevated, a second newborn screen card will be requested at 21 days of age and IRT will be measured again. If the second IRT is not elevated, no additional testing will be required. If the second IRT level is also elevated, the baby will be referred by the baby’s health care provider to Winnipeg Children’s Hospital Section of Pediatric Respirology for further investigations.

IRT is a molecule that is part of a certain pancreatic enzyme. Studies have shown that IRT levels are high in babies with cystic fibrosis. However, IRT levels can be increased for reasons other than cystic fibrosis so follow-up testing is essential to determine if the baby has cystic fibrosis. Only a small percentage of babies with two elevated IRT levels will have cystic fibrosis.

What is cystic fibrosis?
Cystic fibrosis is an inherited (genetic) disease that causes thick mucus to build up in the lungs, digestive system and other organs. This leads to problems like frequent lung infections and problems gaining weight and growing.

Cystic fibrosis affects about 1 in every 3,600 babies born in Manitoba. Cystic fibrosis health problems can range from mild to severe.
How do I find out if my baby actually has cystic fibrosis?
If your baby’s second newborn screen IRT level is still elevated, a test, called a sweat test, is the best way of checking for cystic fibrosis. People with cystic fibrosis have a large amount of salt in their sweat, and measuring the amount of salt in the sweat can help determine whether or not your baby has cystic fibrosis. The sweat test causes no discomfort. Some babies will also need other tests including additional blood tests.

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 cystic fibrosis?
Screening means that babies with cystic fibrosis can be treated early with diet, medications and chest physiotherapy. The goal is to help people with cystic fibrosis live longer, healthier lives.

How does a baby get cystic fibrosis?
Cystic fibrosis is an inherited (genetic) disease. A baby with cystic fibrosis inherited two non-working copies of the cystic fibrosis transmembrane conductance regulator (CFTR) gene, one copy from each parent. People who have one non-working copy of the CFTR gene are called “carriers.” Carriers of cystic fibrosis are healthy, do not have, and will never develop symptoms of cystic fibrosis.

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 CF, please visit the Canadian Cystic Fibrosis Foundation website at www.cysticfibrosis.ca or call 1-800-378-CCFF(2233).

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