

NEWBORN SCREENING BC

Information sheet

My baby had a positive Newborn Screening Result for Cystic Fibrosis

What does this mean?

Your baby had a positive newborn screening result for cystic fibrosis (CF). A positive screen does **not** mean that your baby has CF. A positive screen tells us that there is a small chance that your baby has CF and more testing is needed.

What is newborn screening?

Your baby had a newborn screen. This test was done on a blood sample collected by heel prick shortly after birth. This screen tests for a number of rare conditions like Cystic Fibrosis (CF). Knowing if your child has one of these conditions early in life is important because early diagnosis allows for early treatment and better health outcomes.

What screening tests for cystic fibrosis have been done?

A protein was measured in your baby's blood. It was found to be increased. This protein is called immunoreactive trypsinogen (IRT). Babies with CF have higher levels of this protein but it can also be increased in babies for other reasons. Because your baby's IRT level was increased, the lab did another test on the blood sample which is called a DNA test.

The DNA test looks for common changes in the CF gene seen in people with the condition. A gene is a set of instructions (like a recipe) on how to make a protein. The CF gene makes a chloride channel protein called CFTR. When the instructions are changed the protein does not work properly or is not made. The DNA test looks for common changes (called disease-causing variants) in the CF gene.

We all have two copies of the CF gene, one copy from each parent. Babies with a change (disease-causing variant) in both copies of the gene are likely to have CF.

Your baby's testing showed a change (disease-causing variant) in **one** copy of the gene. This means that your baby is most likely a healthy "carrier" for CF, however more testing is needed to rule out CF. Babies who are CF carriers are no more

likely to get sick than any other baby. They do not need any special medical treatment.

What is cystic fibrosis?

Cystic fibrosis (CF) is an inherited disorder that causes thick mucus to build up in the lungs, digestive system (and pancreas) and other organs. Most people with CF get frequent chest infections. They may also have problems digesting their food and, as a result, may not gain weight as well as they should. Newborn screening means that babies with CF can receive early treatment with medicines and physical therapy that help them digest food and keep their lungs clear of mucus. This treatment will help prevent serious illness and allow the child to live a healthier life. About one baby in 4,300 in BC is born with CF.

What happens next?

Another blood sample needs to be taken from your baby's heel when your baby is 3 weeks (21 days) old. This sample is tested to see if your baby's IRT level is still increased. This second IRT level is a better indicator of CF than the one done at 24-48 hours of age. You may wish to hold your baby or breastfeed them while the blood sample is being taken to provide comfort.

If your baby's IRT level on the second blood sample taken at 3 weeks of age is normal, it is **very unlikely** that your baby has CF but instead is a healthy "CF carrier."

If your baby's IRT level is still increased, this may mean that your baby has CF or it may mean that your baby is a healthy "CF carrier." Further testing will be done to learn more.

After the second IRT results are ready, your primary care provider will discuss next steps.

How do you feel?

For most parents, this is an unexpected result. You may feel scared and upset which are normal feelings. Remember there is only a small chance that your baby has CF. If you are feeling

overwhelmed or have questions, you can talk with your healthcare provider to discuss supports available or call the CF Newborn Screening Team at 604-875-2623. A list of supports is attached.

Where can I get more information?

Talk to your family doctor. You may also call the CF Newborn Screening Team at 604-875-2623.

Information about the BC Newborn Screening Program can be found at

www.newbornscreeningbc.ca

Information about CF can be found at:

- Canadian Cystic Fibrosis Foundation
www.cysticfibrosis.ca/
- Canadian Cystic Fibrosis Foundation (CCFF), Vancouver & Lower Mainland Chapter
www.cfvancouver.ca/
- For a listing of contacts for other CCFF chapters in BC:
www.cfvancouver.ca/
- GeneTests (hosted by the National Center for Biotechnology Information (NCBI))
www.genetests.org

This fact sheet provides basic information only. It does not take the place of medical advice, diagnosis or treatment. Always talk to your health care provider about specific health concerns.

