

BC Newborn Screening Program

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 tells us that there is a small chance that your baby has CF. **A positive screen does not mean that your baby has CF.**

What is newborn screening?

A small spot of your baby's blood was used to get important information about your baby's health. In the first 24-48 hours of life, your baby's heel was pricked and a few drops of blood were put onto a special card. That card was sent to the newborn screening lab at BC Children's Hospital. It was checked for 19 rare disorders, including cystic fibrosis (CF). If found and treated early, the effects of these disorders can be greatly reduced and sometimes prevented.

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.

A DNA test looks for common changes that happen with CF. People's bodies are made up of tiny building blocks called cells. Inside the cells are tens of thousands of instructions called genes. Genes tell the body how to grow and develop. Two copies of each gene are present, one copy from the mother and

one from the father. The DNA test looks for changes (called *mutations*) that happen in one of the baby's genes (CF gene).

Babies with a change (mutation) in both copies of the gene are likely to have CF. Babies with a change (mutation) in **one** copy of the gene are most likely healthy "carriers" for 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.

Your baby had a change (mutation) in **one** copy of the gene. This means that your baby is most likely a healthy "carrier" for CF but there is a small chance that your baby has CF. More testing is needed to find out for sure.

What is cystic fibrosis?

Cystic fibrosis is an inherited life-limiting disorder. It causes thick mucus to build up in the lungs, digestive system (and pancreas) and other organs. Most people with CF get chest infections. They also have problems digesting their food and, as a result, they may not gain weight as well as they should. CF affects about 1 in every 3,600 babies in BC.

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 her or him 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 you wish, you may have your baby tested (*sweat test*) to be sure.

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 IRT results are ready, the CF Newborn Screening Team will contact you to discuss next steps.

How do you feel?

Many parents say they feel shocked, scared, and angry when they are told this. These 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 health care provider or call the CF Newborn Screening Team at 604-875-2000, loc 7005. 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:

- GeneTests (hosted by the National Center for Biotechnology Information (NCBI)) www.genetests.org
- 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/

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.

Revised November 2009