

### My baby had a positive Newborn Screening Result for Cystic Fibrosis

#### What happens next?

Your baby had a positive newborn screening result for cystic fibrosis (CF). This result makes it more likely that your baby has CF but it does not confirm a diagnosis. More testing is needed to find out for sure.

#### 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 **both** copies of the gene. This means that your baby may have CF. More testing is needed to find out for sure.

#### 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?

The CF Newborn Screening Team will contact you to discuss your options, answer questions and organize a visit for your family at BC Children's Hospital in Vancouver.

The CF Newborn Screening Team will arrange further testing for your baby to find out if your baby has CF. The main test that is done is called a sweat test. Other tests may include bloodwork and testing of urine and stool samples. The CF doctor will also examine your baby.

It is important that this testing be done quickly, as early treatment can greatly improve the health of babies with CF.

#### What is a sweat test?

- A sweat test measures how much salt is in your baby's sweat. A large amount of salt in sweat indicates CF.
- To find out more information about a sweat test, please refer to the following [information sheet](#)

#### How do you feel?

For most parents, this is an unexpected result. You may feel scared and upset which are normal feelings. 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](http://www.newbornscreeningbc.ca)

Information about CF can be found at:

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