

## BC Newborn Screening Program

## Information Sheet

### My Baby had a Positive Newborn Screening Result for Cystic Fibrosis *What does this mean?*

#### 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 CF screening tests 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 may have CF or may be healthy "carriers" for CF.

Your baby had a change (mutation) in **both** copies of the gene. This means that your baby is likely to have CF.

#### 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?

Further testing will be done to see for sure if your baby has CF. The main test that is done is called a *sweat test*. Other tests will also be done. It is important that this testing be done quickly as early treatment can greatly improve the health of babies with CF.

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. During your visit, you will meet the CF Newborn Screening Team and your baby will have a sweat test.

### 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.
  - A special machine is used to make a small area on your baby's arm start to sweat. Your baby's skin may feel warm and tingly for 5 minutes while the machine is on. A tight strap is used to keep the machine attached to the baby, which can make the baby a little uncomfortable and he or she may cry a little. Holding your baby or breastfeeding her or him while the sweat is being collected helps comfort your baby.
  - After the machine is removed, a piece of filter paper is put on the baby's arm using tape. The tape is covered with saran wrap and diapers to keep the area warm. The sweat collects on the filter paper for about 30 minutes.
  - When the paper is taken off, part of the arm may be a little red. This is normal and does not hurt.
  - You will be asked to wait 5-10 minutes while the filter paper is weighed to make sure there is enough sample to test.
  - The weighed filter paper is sent to the lab where the amount of sweat and salt is measured.
  - The collection takes about one hour from start to finish. Results of the sweat test are available as soon after the test as possible, often the same day.
- Bring a warm blanket, sweater and hat for your baby. Keeping your baby warm helps your baby to sweat.
  - Do not use lotions or creams (including moisturizing soaps) on your baby's arms and legs on the day of the test.

### Is it ever necessary to repeat a sweat test?

There are times when a baby cannot make enough sweat to complete the test. You will be asked to bring your baby back on another day to try again.

In a small number of cases, the results of sweat tests are "borderline" between maybe having and not having CF. A repeat sweat test, and other tests, will be needed to get more information.

### How do you feel?

Many parents say they feel shocked, scared, and angry when they are told this. These are normal feelings. If you are feeling overwhelmed or have questions, you can talk with your health care provider or call the CF Newborn Screening Team at 604-875-2623. A list of supports is attached. For those that need to travel to Vancouver, travel information is also 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).

### Where is sweat testing done?

Sweat tests are done at BC Children's Hospital in Vancouver. During your visit, you will meet the CF Newborn Screening Team and your baby will have a sweat test.

### What do I need to do to prepare my baby for a sweat test?

- Give your baby plenty of fluids on the day of the test. This makes it easier for your baby to sweat.

Information about CF can be found at:

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

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