NEWBORN SCREENING BC

Information sheet

My baby is a Cystic Fibrosis (CF) Carrier

What does this mean for my baby, me, and my family?

Your baby's newborn screening test showed that they are a cystic fibrosis (CF) carrier. 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 does **not** have a condition called CF.

What does it mean to be a CF carrier?

People's bodies are made up of tiny building blocks called cells. Inside the cells are tens of thousands of instructions called genes. A gene is a set of instructions (like a recipe) on how to make a protein. If the gene/recipe has a major change in it then the protein is no longer made or does not work properly. These gene changes (called disease-causing variants) can affect how our body grows, develops, and functions.

We all have two copies of each gene, including the CF gene (CFTR), one copy from each parent

Babies who are CF carriers have one working copy of the CF gene and one non-working copy with a change (disease-causing variant) in it. One working copy of the CF gene is enough to be healthy. In British Columbia, about 1 in 34 babies are CF carriers – that means about 156,000 **healthy** people in BC are CF carriers.

Why is it important to understand that my baby is a CF carrier?

It is important to know so that you can tell your child later in life that they are a CF carrier. Their future partner can then choose to have CF carrier testing. This information will inform them of their chance of having a baby with CF.

Is there any chance my baby has CF even though the screen showed my baby was a CF carrier?

There is a small chance that cystic fibrosis is not picked up by screening. If you have concerns about your baby's health, please discuss these with your family doctor.

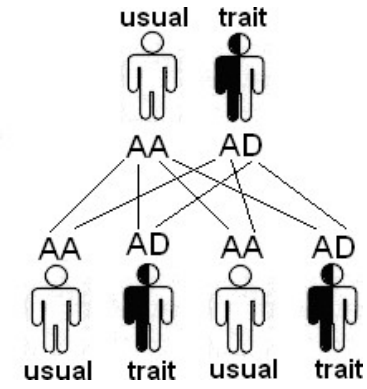
What does having a baby who is a CF carrier mean for me, my partner and for future pregnancies?

Since your baby is a CF carrier, this means that either you or your partner, or both of you are CF carriers. In almost all cases, only one of you will be a CF carrier.

Most people do not know that they are CF carriers. Now that your baby is known to be a CF carrier, both you and your partner have the option of being tested. Testing involves a simple blood test. This can be ordered by your primary care provider.

If only one of you is a CF carrier

There is a 50% chance in every pregnancy to have a baby that is a CF carrier.

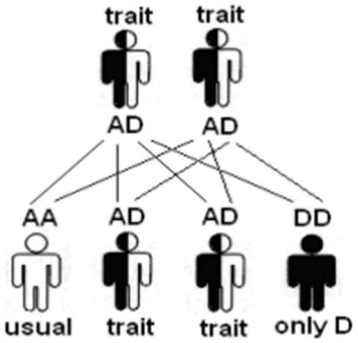


A = working gene
D = non-working gene

If both of you are CF carriers

In the unlikely event that both of you are CF carriers, in every pregnancy there is a:

- 1 in 4 (25%) chance to have a baby that is not a CF carrier and does not have CF
- 1 in 2 chance (50%) to have a baby that is a CF carrier
- 1 in 4 chance (25%) chance to have a baby that has CF



A = working gene
D = non-working gene

If both of you are CF carriers and planning a pregnancy, your doctor can refer you to a genetic counsellor who will review how CF is inherited and all your testing options. These options include testing before, during or after a pregnancy.

Should my other children be tested?

If only one of you is a CF carrier

It is recommended that carrier testing of your other children is delayed until after your child can make their own decision about whether to be tested as adults.

If both of you are CF carriers

In the unlikely scenario that both of you are CF carriers, your other children may be tested to make sure they do not have CF. Your family doctor will help you decide the type of testing that would be best (a DNA test or a sweat test or both) and can arrange the testing.

Should my relatives be tested?

Any blood relative (for example, brother, sister, aunt, uncle, or cousin) of a CF carrier may also be a CF carrier. Your relative may have questions about their chance of having a baby with CF, or may think that one of their children has CF. We suggest they talk to their family doctor who may offer them testing.

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. This is a lifelong condition that requires ongoing medical care. 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.

Where can I get more information?

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

If you and your partner are both carriers then genetic counselling is an option. Ask your family doctor to refer you to the Department of Medical Genetics at BC Children's and Women's Hospital or Vancouver Island Medical Genetics (at Victoria General Hospital) for genetic counselling.

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.