



Cystic Fibrosis Canada

Testing for Cystic Fibrosis

Who should be tested?

Carrier testing for cystic fibrosis (CF) is recommended for anyone with a family history of cystic fibrosis, for anyone whose partner has a family history of cystic fibrosis, and for anyone with a medical condition which might possibly be connected to cystic fibrosis, such as male infertility.ⁱ

In general, carrier testing is performed for the purpose of family planning. Since carriers are unaffected by the disease, the fact that they are carriers is relevant only if they — or their grown children — wish to have children. A genetic counsellor can help you decide whether or not to be tested.



“With cystic fibrosis carrier testing and the knowledge of whether or not they are a carrier, our children are now able to plan their families knowing if their partner and children need to be tested to see if they carry a CF gene. Carriers can also be prone to CFTR-Related Metabolic Syndrome (CRMS), where they may have some of the complications of cystic fibrosis. With the information of your carrier status, appropriate medical treatment can be received.”

Marc and Anne-Marie Beausoleil, Tilbury, Ontario, parents of four children (one with CF, two carriers, one not a carrier)

How reliable is the test?

Since the discovery of the gene responsible for cystic fibrosis in 1989 and the development of new technologies, it has become possible, in most families, to detect the mutations in the gene through laboratory tests, using blood samples or cheek swabs. Samples are sent to specialized molecular diagnostic laboratories for analysis.

“In Canada, laboratory tests detect mutations that cover for approximately 98% of the Canadian CF population.”

Routine genetic tests cannot yet detect all cystic fibrosis carriers. Currently, over 1,900 mutations in the gene responsible

for cystic fibrosis have been identified, and most medical diagnostic laboratories screen for approximately 40 of the most common mutations. In Canada, laboratory tests detect mutations that cover for approximately 98% of the Canadian CF population.ⁱⁱ

If medically indicated, a complete mutation sequencing can be arranged to provide a more detailed analysis.

ⁱⁱ CF Registry 2012 Annual Report

What does a positive test result mean?



A positive test result means that you are a cystic fibrosis carrier. In other words, you do not have cystic fibrosis, but you could have a child with cystic fibrosis, if and only if, your partner is also a CF carrier or has cystic fibrosis.

If your test result is positive and you are thinking of having a child, your partner should also be tested. If your partner's test result is negative, there is still a possibility of having a child with cystic fibrosis, because there is a small chance that your partner carries a mutation which was not detected through testing. There is a greater chance that the child will be a CF carrier.

What does a negative test result mean?

A negative test result means that the likelihood of being a cystic fibrosis carrier is very low. However, because not all the mutations in the gene have been identified and extremely rare mutations are not included in the test, there is still a small possibility that you may be a carrier. The following table summarizes the risks of having a child with cystic fibrosis, prior to and following carrier testing.

Carrier status of parents ⁱⁱⁱ	Risk of having a child with cystic fibrosis
Both partners unknown	1 in 2,500
One partner tested negative; other partner unknown	1 in 24,000
One partner tested positive; other partner unknown	1 in 100
Both partners tested negative	1 in 230,000
One partner tested positive; other partner tested negative	1 in 960
Both partners tested positive	1 in 4

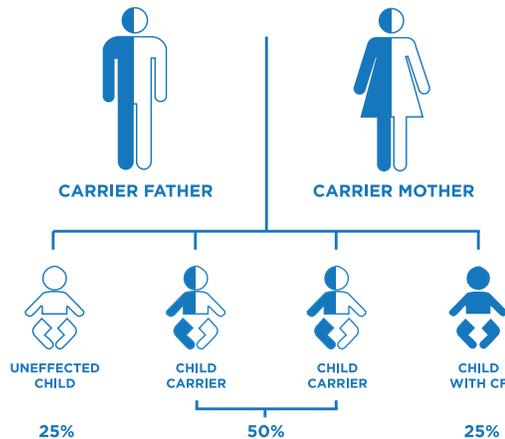
ⁱⁱⁱ Based on Caucasian status.

What if both you and your partner test positive?

Cystic fibrosis occurs when a child inherits two defective copies of the gene responsible for cystic fibrosis, one from each parent.

The possible outcomes of each pregnancy are:

- ◆ A 25 percent chance of a child who does not have cystic fibrosis, and is not a cystic fibrosis carrier
- ◆ A 50 percent chance of a child who does not have cystic fibrosis, but does carry one defective copy of the gene, and is therefore a cystic fibrosis carrier
- ◆ A 25 percent chance of a child who has two defective copies of the gene, and therefore has cystic fibrosis



What are your options?

If both you and your partner have tested positive as carriers, you may wish to consider genetic counselling prior to conceiving a child.

A genetic counsellor will explain in greater detail the implications of your test results, and offer other family planning options, including adoption, pre-implantation genetic diagnosis, pregnancy with prenatal diagnosis, or pregnancy without prenatal diagnosis.



One family planning option is pre-implantation genetic diagnosis. Through in vitro fertilization, eggs are fertilized to produce embryos that are then tested for genetic defects. Only those free of the genetic defects are implanted in the womb.

There are also prenatal tests available which can detect the presence of cystic fibrosis in the fetus. These tests are:

- ◆ Chorionic villus sampling (CVS): At 8 to 12 weeks of pregnancy, a small number of cells are removed from the membranes surrounding the fetus for laboratory analysis.
- ◆ Amniocentesis: At 16 to 18 weeks of pregnancy, a sample of the fluid surrounding the fetus is removed for laboratory analysis.

Both of these tests involve a small risk of miscarriage.

If a prenatal test indicates that the fetus does have cystic fibrosis, you and your partner will have to decide on a course of action most appropriate to your beliefs and circumstances.

Testing your baby for cystic fibrosis after birth is another option for couples who do not wish to undergo pre-implantation diagnosis or prenatal testing. Many Canadian provinces screen all newborns for cystic fibrosis.

All of these procedures can be discussed with a genetic counsellor who will explain the process and associated risks, and can help you consider your options. A genetic counsellor will not make a decision for you — only you and your partner can make a decision.

For more information about CF carrier tests

For information on the availability of cystic fibrosis carrier testing, contact your family physician or CF Clinic, and ask to be referred to the nearest Genetics Centre.

How can you connect with other Canadians affected by cystic fibrosis?

Join the My Cystic Fibrosis (CF) Canada Network, Canada's first-ever social network designed by CF patients specifically for CF patients to connect and share important information about living with this severe and potentially fatal genetic disease. Canadians with cystic fibrosis can discuss the treatments, programs and services available in their province/region, via video chat and instant message, share stories and personal issues, and receive support for advocacy and fundraising initiatives from a much broader community.

Cystic Fibrosis Canada acknowledges Novartis Pharmaceuticals Canada Inc. for the collaboration that has made the My CF Canada Network possible.

Sign-up today at www.mycfnetwork.com

For more information about cystic fibrosis, visit cysticfibrosis.ca

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