

Frequently Asked Questions

- 1. Do I need to be tested every time I have a baby?**
NO. If you are not a carrier, you remain at very low risk of having a child with CF. If you are a carrier and you have a new partner, your new partner will need to be tested.
- 2. If no gene change is found, could I still be a carrier?**
YES. The current carrier test detects about 80% of carriers, and cannot detect every gene change that causes CF. If no gene change is found, you are not a carrier of the most common gene changes, but there is still a small risk that you are a carrier.
- 3. If I am a carrier, can I develop CF?**
NO. If you are a carrier, you have a gene change in one of your two CF genes. Relatives of carriers are more likely to also be carriers and may wish to have testing.
- 4. What if I have a relative who is a carrier or has CF?**
YOUR CHANCE of being a carrier of CF is greater than most people and you and your partner should consider testing. If you or your partner have a family history of CF contact Genetic Health Services Victoria to discuss testing. There is no charge for testing where an individual residing in Victoria has an immediate blood relative with CF.
- 5. If I have no family history of CF can I have a child with CF?**
YES. Most families where a child is born with CF have no family history of this condition.
- 6. How do I arrange to have a test?**
Your doctor needs to sign your request form.
This is sent with your cheek brush in the reply paid envelope.
- 7. How long until I get a result?**
It takes 5-7 working days from when your sample arrives for the laboratory to process and send the result to your doctor.
- 8. How do I get my result?**
Results are mailed to your referring doctor.

Contact Details

Further information regarding the CF Screening Program may be found at:
<http://www.cfscreening.com.au>

or

Genetic Health Services Victoria
10th Floor
Royal Children's Hospital
Flemington Road
Parkville VIC 3052
Ph: (03) 8341 6201
<http://www.genetichealthvic.net.au>



For Information on Cystic Fibrosis
Contact:
Cystic Fibrosis Victoria Inc.
80 Dodds Street
Southbank Victoria 3006
Ph: (03) 9686 1811
FreeCall: 1800 633 685
<http://www.cfv.org.au>



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carrier testing for
Cystic Fibrosis

www.cfscreening.com.au



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What is Cystic Fibrosis?

Cystic fibrosis (CF) is an inherited disease that affects breathing and digestion in infants, children and young adults. It causes thick mucus which traps bacteria, resulting in recurrent infections that damage the lungs. Thick mucus in the gut also makes digestion of food difficult.

Infants and children with CF require daily chest physiotherapy to clear mucus from their lungs, frequent courses of antibiotics and must take medication to assist digestion.

Until recently, many children with CF died in early childhood, but now many live to be 30, 40 or more. There is no cure for CF but better treatments are constantly under research and development.

Is our baby at risk?

CF is most common in individuals of European descent but can occur in other ethnic groups. Most couples who have a child with CF have NO family history of this condition.

Babies inherit one copy of each gene from each parent. Genes can have changes, known as mutations, which affect their function. A baby will have CF only if they inherit a gene change for cystic fibrosis from **both** parents. This is only possible if **both** parents are carriers of a gene change for CF.

About 1 million Australians 'carry' a gene change for cystic fibrosis as well as a healthy copy. **Carriers are completely healthy.**

If you are...	Then your risk of being a carrier is:
Caucasian	1 in 25 (4%)
Asian	1 in 90 (1.1%)

(For individuals with no family history of CF)

How can we find out our risk of having a child with CF?

A couple is only at risk of having a child with CF if BOTH the father and mother are carriers. We cannot test every gene change for CF, but we can test for the 12 most common gene changes that result in severe CF and identify about 80% of carriers amongst those of European origin. Testing both you and your partner will give you as a couple the most accurate risk assessment of having a child with CF.

The carrier test involves firmly wiping your inner cheek with a soft swab that is similar to a large cotton bud - a totally painless procedure. In the laboratory, DNA from the cheek cells on the swab will be tested for the most common CF gene changes.

When should we have carrier testing?

It is up to you whether you choose to be tested. To ensure there is time to make decisions it is best to test either before pregnancy, or early in pregnancy.

We recommend testing before 14 weeks of pregnancy. Then you have the maximum time to decide what to do if you are among the small number of couples who are both carriers.

How do we get tested?

Speak to your Doctor or Midwife. As yet there is no Medicare rebate for this test and there will be a charge.

What do the results mean?

There are two possible results from the test; carrier or non-carrier.

Carrier

If your test shows that you have one copy of the gene change, you are a carrier. You will be offered genetic counselling. Your partner's result will determine your risk as a couple of having a child with CF.

Non-Carrier

This means that you do not have one of the common gene changes and so your risk of being a carrier is greatly reduced. However this test can not absolutely rule out the possibility that you may have a gene change that is not tested for.

Couple Results	Risk of having a child with CF
Not tested	1/2,500
Both non-carriers	Less than 1/60,000
1 non-carrier, 1 not tested	Less than 1/12,000
1 carrier, 1 non-carrier	Less than 1/500
1 carrier, 1 not tested	Approximately 1/100
Both carriers	1/4

What if we are both carriers?

Two people who are carriers have a 1 in 4 (25%) chance of having a child with CF in each pregnancy. If you are both carriers you will be provided with genetic counselling. Following counselling you may choose to have tests to identify whether your baby has CF. If the tests diagnose CF you have a choice about whether to continue or terminate the pregnancy. If you are identified as being carriers prior to a pregnancy, then you also have the option of preimplantation genetic diagnosis using in vitro fertilisation.