

Carrier Screening

Cystic fibrosis (CF) is an inherited condition that mainly affects the lungs and digestive system. An estimated one in 25 Australians carry the gene mutation that can cause CF.

Carrier screening is the only way to find out if you are a carrier of the CF gene mutation. In Australia, carrier screening services are available for people aged over 18 years.

What is CF Carrier Screening?

CF carrier screening is a genetic test to identify if people have changes to the gene that causes CF. This test can be done using a blood sample, cheek swab or saliva sample. The test can be ordered any time prior to pregnancy or in the early stages of pregnancy.

What Does it Mean to Be a CF Carrier?

Carriers of a single CF gene are healthy and do not have CF or CF symptoms. To be diagnosed with CF you need to receive a CF gene mutation from each parent.

If both parents are carriers of a CF gene mutation, the chance of having a child with CF is one in four (or 25%), for each pregnancy.

What if There is No Family History of CF?

Almost all children born with CF are born to parents who have no family history, so, having no family history does not mean you are not a carrier. Most carriers do not know that they are carriers.



What if I Have a Family History of CF?

If you do have a family history of CF and you would like to know your carrier status, speak to your doctor. Give them as much information as possible e.g. the name and DOB of the family member who has CF or is a carrier should be entered on the request form, as well as the specific gene mutation and the year of diagnosis/screening if known. This will ensure you are tested for that specific gene mutation as well as other common CF gene changes.

Should I Be Screened?

Carrier screening is a very personal decision. If you are considering pregnancy and decide to be screened, it is preferable to have it done before you and your partner conceive so you have time to consider your reproductive options.

Should your results indicate that you are a carrier of the CF gene, talk to your doctor or referring health professional for more information.

Is Carrier Screening the Same as Newborn Screening?

No. Newborn screening is conducted soon after birth, on all babies born in Australia, to identify certain diseases, including CF.

Screening Options

Carrier screening is available throughout Australia. There are several different screening options which may be available to you including:

- A single panel screen for CF or expanded panel for several of the most common mutations or full sequencing of the CF gene.

The following options include screening for CF and other conditions and may be accessed on discussion with an appropriate health professional:

- A 3-panel screen for CF, Spinal Muscular Atrophy and Fragile X syndrome.
- An expanded carrier screening – ability to screen for over 1,000 autosomal recessive and X-linked conditions.

How Much Will It Cost?

You should expect to pay between \$150-380 for carrier screening, which is not covered by Medicare. For individuals with a family history or clinical suspicion of CF, the fee may be reduced or waived but a PathWest laboratory will need to be used. A blood test is preferred, and the CF carrier screening sample will need to be sent to the PathWest Dept of Diagnostic Genomics, QEII.

Specifically, testing will be covered for individuals with a relationship closer than first cousins once removed and where there is clinical suspicion of CF.

6 Steps to CF Carrier Screening

STEP 1 - Get Informed- discuss with your family, partner and doctor.

STEP 2 - Find out the Process- ask your GP or genetic services for referral and testing procedures.

STEP 3 - Request the Test- and provide your sample.

STEP 4 - Get Your Results- you may be contacted directly to discuss your results, or you may be offered an appointment with a genetic counsellor.

STEP 5 - Understand Your Results- your results will identify if you are a CF carrier for the common gene mutations as well any specific gene changes that you have requested testing for. A negative result usually means there is a low risk that you are a carrier. There remains a very small risk that you could be a carrier for a very rare CF gene mutation.

STEP 6 - Inform Others- if your result shows you are a CF carrier, it is important that you let others in your family know as they may also be a CF carrier and may like to have this information.

Genetic Services WA (GSWA)

GSWA provide free genetic counselling for couples if both partners are carriers of a CF gene mutation. However, if you are already pregnant and have a family history of CF you and your partner will be seen as a matter of importance to expedite testing. If you have any questions about whether you qualify for genetic testing please call [GSWA](#) during business hours and ask to speak with a genetic counsellor.

Useful Resources

- [CF Carrier Screening Program \(CFCC\)](#)

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