

Carrier Screening

Cystic fibrosis (CF) is an inherited condition that mainly affects the lungs and digestive system. An estimated 1 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 by using a saliva sample. The test can be ordered any time prior to pregnancy or in the early stages of pregnancy. If you are interested in carrier screening, speak to your doctor.

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 1 in 4 (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, it is important you tell your doctor. Please give your doctor as much information as possible, for example, 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 and the year of diagnosis/screening if known. This will ensure you are tested for that specific gene as well as other common CF gene changes.

NB: In WA, if you have a family history of CF, your carrier screening test fee may be 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.

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 you can ask for a referral to a genetic counsellor for more information.

Is Carrier Screening the Same as Newborn Screening?

No, carrier screening is not the same as newborn screening. Newborn screening is conducted soon after birth, on all babies born in Australia, to identify certain diseases, including CF.

Where Can I Get Screened?

Carrier screening is available throughout Australia. Discuss CF carrier screening with your doctor or to find out more about CF screening in your state contact your nearest genetic service or your local CF organisation.

How Much Will It Cost?

You should expect to pay between \$200 and \$350 for CF carrier screening, which is not covered by Medicare. However, if you have a relative or family member with CF you may be entitled to a rebate.

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 CF carrier for the common gene changes 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 carrier 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.

Useful Resources

- [CF Carrier Screening Program \(CF Community Care\)](#)
- [Genetic Carrier Screening \(CFWA\)](#)
- [Genetic Services WA](#)

Special thanks to Rebecca Freedman, Genetic Services WA for input into this factsheet.

Cystic Fibrosis WA
The Niche
11 Aberdare Road
Nedlands WA 6009

T: +61 8 6457 7333
F: +61 6457 7344
E: admin@cfwa.org.au

Disclaimer: This publication is for general education and information purposes. Contact a qualified healthcare professional for any medical advice needed.
© Cystic Fibrosis Western Australia