

## Carrier Screening

CF is an inherited condition that mainly affects the lungs and digestive system. An estimated one in 25 Australians, over 1 million people in Australia, 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 generally healthy and do not have CF. 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 they have together.

### What if there is no family history of CF?

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



### What if there is 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- the name and date of birth of the family member who has CF or is a carrier should be provided and if known, add the specific gene mutation and the year of diagnosis or screening.

This will ensure you are tested for that specific gene mutations 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 mutation, talk to your doctor or referring health professional for more information.

## Screening options

The test, which should be ordered by your GP, obstetrician or gynaecologist, involves taking a blood sample, a swab from inside the mouth or a saliva sample. There are several different screening options which may be available to you including:

- A single panel screen for CF, an expanded panel for several of the most common CF mutations, or a complete 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.

## Where to go for the screening test?

Carrier screening is available throughout Australia. In WA, your GP will possibly refer you to PathWest laboratory or to a private laboratory for your CF carrier screening.

## How much will it cost?

Genetic testing for CF is now bulk billed for Medicare eligible patients. If you are not eligible for Medicare it will cost ~\$389. For individuals with a family history or clinical suspicion of CF, the fee may be reduced or waived under certain circumstances.

## What if the results show you are a CF carrier?

Please ask your GP for a referral to see a genetic counsellor who will elaborate further on the findings and discuss your options and the potential impacts with you and your family. Other family members may choose to also access carrier screening or genetic counselling following a positive result.

## Genetic Services WA

GSWA provides free genetic counselling for couples if both partners are carriers of a CF gene mutation. 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

- [Reproductive carrier testing for CF](#) (Dept. Health and Aged Care)
- [Genetic carrier screening medicare](#) (Australian Clinical Labs)