CYSTIC FIBROSIS

CFfacts
cystic fibrosis fact sheet

Carrier Screening

Cystic fibrosis (CF) is an inherited condition that mainly effects the lungs and digestive system. An estimated 1 in 25 Australians carry the gene mutation that can cause CF.

If you are thinking about having a family, carrier screening services are available in Australia to help you get information and understand your options.

Knowing my options – 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 (the preferred method in WA) or using cells taken from inside the mouth with a cheek swab or saliva sample.

Carrier screening is only available to those 18 years and over and is the only way to find out if you are a carrier of the CF gene mutation. The test can be ordered any time prior to pregnancy or in the early stages of pregnancy. If you are interested in CF carrier screening, speak to your doctor or a genetic counsellor.

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 a person needs to have 2 copies of the CF gene, one from each parent. If both parents are carriers of a CF gene, 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 or know that a relative is a carrier of CF and you would like to know your carrier status then it is important to find out the specific gene change in your family. This will ensure that you are being screened for that specific gene change as well as the other common gene changes. For people with a family history of CF, it is best to speak to a genetic counsellor prior to testing to ensure that the correct test is ordered based on your family history.

:

Should I be screened?

Only you can decide whether or not to find out your carrier status. If you are thinking about being screened, speak with your doctor or a genetic counsellor. Your GP, obstetrician or genetic counsellor can request a carrier screen for you.

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 newborn screening the same as carrier 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. It is commonly known as the 'heel prick' or Guthrie test. A small proportion of CF carriers are incidentally identified through newborn screening. Most babies who are carriers of CF are not identified through newborn screening.

If a baby is found to be a carrier of the CF gene, further investigations will be offered to his/her parents to identify whether they are affected or only a carrier.

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 please tell your GP as you may be entitled to a rebate.

How do I tell others if I am a CF carrier?

If you are a carrier of CF it can be useful to tell your relatives so that they can make the decision to access carrier testing themselves. It is also important to tell your partner particularly if you are considering having children.

:

CYSTIC FIBROSIS

CFfacts
cystic fibrosis fact sheet

6 steps to CF carrier screening

STEP 1 - **Get informed**
Discuss CF carrier screening with your family, partner and doctor and decide if it is important for you to know your CF carrier status

STEP 2 - **Find out the process**
Check with your GP or the genetic service provider in your state for referral criteria and testing procedures.

STEP 3 - **Request the test**
Ask your doctor or genetic counsellor to request CF carrier screening. They will complete a test request form and then you will need to provide your sample (either blood sample or cheek brush/saliva sample). Remember, if you have a family history of CF or a relative who is a carrier, the name and DOB of the family member should be entered on the request form and the year of diagnosis/screening if known. It is also helpful to find out the specific gene and make sure it is written on the test request form.

NB: In Western Australia, 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.

STEP 4 - **Get your results**
Notification of results will vary depending on who ordered your test, some health professionals may contact you directly to discuss your results, others will arrange for you to discuss your results with a genetic counsellor. It is important to find out when and how you will receive your results prior to being screened.

STEP 5 - **Understand your results**
Genetic counsellors are available to discuss the outcome of 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 gene change causing CF.

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

- Cystic Fibrosis Community Care Carrier Screening Program www.cfscreening.com.au

Special thanks to Cystic Fibrosis Community Care for their input into this factsheet.

Last revised November 2018

: