CYSTIC FIBROSIS

CFfacts
cystic fibrosis fact sheet

Carrier Screening

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

Are you thinking about having a family? Carrier screening services are available in Australia to help you be informed 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 on a blood sample or from 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 change. The test can be ordered prior to pregnancy or early in 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 change are healthy and do not have CF nor related symptoms. As CF is an inherited condition, both parents need to be a carrier of the CF gene. If both parents are carriers of a CF gene change, 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.

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. 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 being ordered for you based on the information available about your family history.

Should I be screened?

Making the decision to know your carrier status is a decision only you can make. If you are thinking about being screened then 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 on all babies born in Australia at birth to identify certain diseases including CF. It is commonly known as the 'heel prick' test. The purpose of newborn screening is to identify babies who have CF. A small proportion of carriers of CF are incidentally identified through newborn screening but this is only a very small proportion of all CF carriers. Most babies who are carriers of CF are not identified through newborn screening.

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 to $300 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 access carrier testing themselves.

6 steps to CF carrier screening

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

STEP 2 - **Contact your local genetics service or CF organisation**
Find out the process for CF carrier screening in your state.

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 – depending on what the options are in your State).

Remember, if you have a family history of CF and/or know a relative who is a carrier, to find out the name of the specific gene change and make sure that is written on the test request form.

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's important to find out 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 as per your family history. 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 if or when they decide to plan a family.

Useful Resources

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

Supported by the Rotary Club of Balwyn

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