



Carrier Screening:

How to be tested

1 in 20 people carry a gene change for cystic fibrosis, fragile X syndrome, or spinal muscular atrophy, but most people are unaware that they are carriers.

Genetic carrier screening services are available in Australia. This screening helps people find out whether they have a chance of having a child with a genetic condition.

Genetic carrier screening is becoming increasingly available in Australia

Several different screening options are available:

- A 3-panel screen, which screens for carriers of three of the most common inherited conditions:
 - ▲ cystic fibrosis (CF)
 - ▲ spinal muscular atrophy (SMA)
 - ▲ fragile X syndrome (FXS).
- A single panel screen, which screens for carriers of one of these conditions.
- A larger panel, which screens for carriers of a wider range of common and rare inherited conditions.

People with a family history of a particular genetic condition may wish to request testing for that specific condition only, or they may wish to request a test that also includes other commonly inherited conditions.



If you decide to have carrier screening, we recommend you follow the steps below:

1. Visit your healthcare provider

Genetic carrier screening is usually requested by general practitioners, obstetricians, fertility specialists, midwives, genetic counsellors or medical geneticists.

Let your healthcare provider know that you wish to have genetic carrier screening.

Your healthcare provider will need to complete a test request form that you can take to a local pathology collection centre.

Alternatively, if you do not live near a pathology collection centre you can order a saliva sample kit that can be mailed to you.

If you are aware of a family history of an inherited condition, knowing the specific gene change in your family and communicating this to the healthcare provider arranging your testing, will ensure the appropriate test is ordered for you.

It is recommended that your testing be performed by a specialist genetic testing laboratory.





2. Have the test

It is recommended that the female partner is tested first, as testing may include conditions carried on the X chromosome.

Take your test request form to an affiliated pathology collection centre and have your blood sample taken.

If you have a family history of some conditions, such as FXS or CF, Medicare and Private health insurance may subsidise the cost of carrier screening.

3. Visit your healthcare provider to get your results


Typically your results will be communicated to your healthcare provider within 2-4 weeks. They will inform you if you carry a gene change for any of these conditions.

4. If the test shows you are a carrier

It is recommended that you discuss your result with a genetic counsellor.

Some carrier screening services provide genetic counselling as part of their service.

Depending on your result, your healthcare provider or a genetic counsellor will discuss carrier testing for your partner.



Carrier screening is recommended either before pregnancy or early in pregnancy (during the first 12 weeks)

Screening is relevant to everyone - most people who are carriers do not have a known family history of the condition. Making a decision about screening is a choice only you can make.



Recommended carrier screening provider

Victorian Clinical Genetics Services (VCGS)

VCGS provides carrier screening services including genetic counselling throughout Australia and New Zealand.

w. vcgs.org.au/tests/prepair

e. vcgs@vcgs.org.au

p. 1300 118 247

For more information

Cystic Fibrosis Community Care (CFCC)

w. cfcc.org.au

p. (03) 9686 1811



Fragile X Association of Australia (FXAA)

w. fragilex.org.au

p. 1300 394 636



Spinal Muscular Atrophy Australia Inc. (SMA Australia)

w. smaaustralia.org.au

p. (03) 9796 5744



This brochure was produced by the Carrier Screening Community Education Campaign Reference Group which includes representatives from CFCC, FXAA, Genetic Support Network of Victoria, Royal Children's Hospital Melbourne, SMA Australia, and VCGS. VCGS has no financial affiliation with CFCC, FXAA, or SMA Australia.

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