Carrier Screening:

How to be tested

Reproductive genetic carrier screening services are available in Australia. This screening could help you find out if you have an increased chance of having a child with a serious genetic condition.

Most people who are carriers of a genetic condition do not have a known family history of the condition. Making a decision about screening is a choice only you can make.

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

1 in 20 people carry a gene change for cystic fibrosis (CF), fragile X syndrome (FXS), or spinal muscular atrophy (SMA), and most are unaware that they are carriers. Being a carrier may not impact your own health, but there is a chance that your future children may have the condition. There is currently no cure for these conditions.

What is Spinal Muscular Atrophy?

SMA is an autosomal recessive genetic condition which weakens the muscles over time, making it hard to move limbs, walk, swallow, and breathe. There are different types of SMA, varying in severity and age of onset. Some new treatments can prevent this muscle weakening, and a promising new gene therapy may help those with SMA live a longer, easier life.

What is Cystic Fibrosis?

CF is an autosomal recessive genetic condition that causes too much mucus to form on and around the major organs. Different types of CF can cause different problems, including lung problems (infections, reduced function, or failure), and reduced nutrient absorption (making it harder to grow and gain weight). There are many treatments for CF including physiotherapy to clear the lungs and airways, medications, and other clinical interventions.

What is Fragile X Syndrome?

FXS is caused by a lengthened fragile X (FX) gene on the X chromosome. Females with a medium (premutation) or long (full mutation) FX gene have an increased chance of having a child with FXS, whereas males with a medium or long-length FX gene generally do not. People with the long-length FX gene can have a range of intellectual and behavioural symptoms including developmental delay, intellectual disability, autism, anxiety, ADHD, and sensory sensitivity. FXS is not life-limiting and a range of treatments, interventions and management strategies can benefit people with FXS.

People with a medium-length FX gene (carriers) are at risk of developing fragile X-associated tremor ataxia syndrome or fragile X-associated primary ovarian insufficiency, but may have no symptoms associated with fragile X at all.

Genetic carrier screening is becoming increasingly available in Australia

Several different screening options are available:

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

This screening is relevant to anyone planning a family or in early pregnancy. Most carriers have no family history of the condition, however, if you do have a family history of a genetic condition you should discuss this with your healthcare provider. You may wish to discuss testing for that specific condition only, or you and your healthcare provider may discuss having a test that also includes other commonly inherited conditions.

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

1. Visit your healthcare provider

It is recommended that carrier screening is organised with the guidance and support of your healthcare provider. Carrier screening is usually requested by general practitioners, obstetricians, fertility specialists, midwives, genetic counsellors, or medical geneticists.

Let your healthcare provider know that you are interested in carrier screening and would like to learn more.

If you decide to have screening your healthcare provider will need to complete a test request form. This form can be taken to a local pathology collection centre, or used to order a saliva sample kit from a genetic testing laboratory which can be mailed to you.

If you have a family history of a genetic condition, tell your healthcare provider so they can ensure they arrange the most appropriate test for you.

2. Have the test

It is recommended that the person seeking to become pregnant is tested first, as testing may include conditions carried on the X chromosome.

Take your test request form given to you by your healthcare provider to an approved pathology collection centre and have your blood sample taken. Alternatively, complete and return the at-home saliva sample kit.

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 the conditions that you were screened for.

4. If the test shows you are a carrier

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

Some carrier screening services provide genetic counselling as part of their service. If this is not included, accessing a genetic counsellor can be organised through your healthcare provider.

Depending on your results, your healthcare provider or a genetic counsellor may also discuss carrier testing for your partner.

Where can I go for more information?

carrierscreening.org.au

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