Does Medicare cover screening?

Genetic Carrier Screening for CF, SMA and FXS will be Medicare rebateable from November 1st, 2023, and is available for:

- females planning a pregnancy
- females who are pregnant
- male reproductive partners of female carriers of cystic fibrosis or spinal muscular atrophy

The test is available free of charge once per lifetime.

Why choose carrier screening with Genomic Diagnostics?

- **Simple and safe:** a one-tube blood test
- **Convenient:** available at a collection centre near you at anytime
- **Fast results:** within 10 working days of collection
- **Accurate:** looks for the most common gene changes for CF, SMA and FXS in the Australian population
- **Supportive:** dedicated customer care team to answer all your queries

Please refer to genomicdiagnostics.com.au for more information

How do I get the Genetic Carrier Screen?

Step 1: See your doctor to get a request

After discussing **Genetic Carrier** screening with you, your doctor will complete a request form.

Step 2: Have your sample collected

Attend your most convenient Healius Pathology collection centre to have your blood sample taken.

Step 3: See your doctor to get your results

Genetic Carrier Screen results will be delivered to your doctor within 10 working days of your sample collection.

If you and your partner are identified as a carrier couple you will be offered the option to talk with a genetic counsellor free of charge.

Genomic Diagnostics

For more information please contact us at info@genomicdiagnostics.com.au

1800 822 999

genomicdiagnostics.com.au

PO Box 250, Heidelberg West, VIC 3081

Healius Pathology Pty Ltd (ABN 84 007 190 043) t/a Genomic Diagnostics_139_GD_v4_Aug23

Genomic Diagnostics

Genetic Carrier Screening

Informed Reproductive Decision Making

Healius companies

abbott pathology | dorevitch pathology | laverty pathology | qml pathology | tml pathology | western diagnostic pathology

Genetic Carrier Screening

Genetic carrier screening (GCS) is testing to identify couples at increased risk of having children with three of the most common, severe inherited genetic disorders in the Australian population:

- Cystic fibrosis (CF)
- Spinal muscular atrophy (SMA)
- Fragile X syndrome (FXS)

One in twenty Australians will be a carrier for at least one of these conditions. Most will not have a family history of the disease.

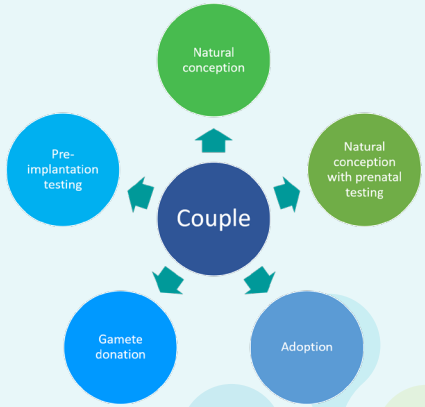
Carriers are usually unaffected by the condition themselves, but are at increased chance of having an affected child.

Why and when should I be tested?

Knowing your carrier status can be extremely useful when making reproductive choices, and the only way to know your carrier status is through genetic testing. If couples are found to be carriers, they can consider several reproductive options.

The best time to have carrier testing is before pregnancy. This offers you the widest range of reproductive choices and allows more time to make important decisions. However, testing can still be performed during pregnancy, ideally before 12 weeks.

You will only need to be tested once in your lifetime, as your carrier status does not change.

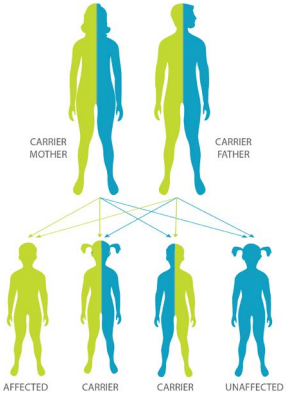


Conditions screened by GCS

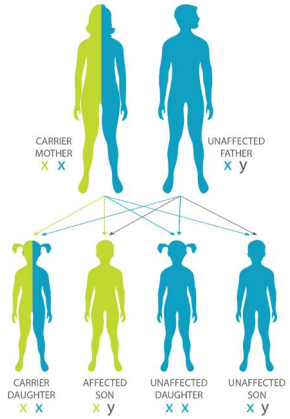
CF, SMA and FXS can have devastating effects on life-expectancy and quality of life.

- CF is the most common inherited disorder in Caucasians
- SMA is the most common genetic cause of mortality in children under two
- FXS is the most common form of inherited intellectual disability

CF and SMA are autosomal recessive conditions, which means that both parents must be carriers to have an affected child. FXS is an X-linked condition, so the mother must be a carrier for children to be affected.



Autosomal recessive inheritance



X-linked inheritance

How does genetic carrier screening work?

GCS tests for the most common genetic changes that cause CF, SMA and FXS using DNA from a sample of your blood.

It is recommended that the female partner is tested first, followed by testing of the male partner for CF or SMA if required.

When both partners are identified as carriers for CF or SMA, or the female partner is identified as a carrier for FXS, they are considered to be a carrier couple.

When the female partner is not identified as a carrier through GCS, the couple is considered to be at greatly reduced chance of having an affected child..

Genetic Counselling

If you and your reproductive partner are shown to be a carrier couple for any of these conditions, genetic counselling is recommended to provide more information about the condition and enable detailed discussions regarding your reproductive options.

This support is provided free of charge through Genomic Diagnostics for carrier couples where at least one partner is tested through the Healius Pathology Network.