Does Medicare cover the cost?

Yes, genetic carrier screening for cystic fibrosis, spinal muscular atrophy and fragile X syndrome is bulk-billed. Medicare eligibility criteria apply, please speak to your doctor for more information.

Why choose us for screening?

- ✓ **Simple and safe:** A one-tube blood test
- ✓ **Convenient:** Available at a collection centre near you
- ✓ **Fast results:** Within 10 working days of collection
- ✓ **Accurate:** Identifies the most common gene changes for these three conditions
- ✓ **Supportive:** Dedicated customer care team to answer your questions and provide support

dorevitch.com.au

Steps to get a genetic carrier screen

1 Step 1: See your doctor to get a request

Your doctor will discuss genetic carrier screening with you and complete a request form.

2 Step 2: Have a blood test

Visit your closest Dorevitch Pathology collection centre to have your blood sample taken.

3 Step 3: See your doctor to get your results

Your genetic carrier screen results will be delivered to your doctor within 10 working days.

If you and your partner are identified as a carrier couple, we will offer free genetic counselling to support you with your next steps.

Genetic Carrier Screening helps you make informed choices for you and your family.

Learn more:

Healius companies

dorevitch pathology | Genomic Diagnostics

Healius Pathology Pty Ltd (ABN 84 007 190 043) t/a Dorevitch Pathology_Pub/Mr/1746_V2_Oct23

Genetic Carrier Screening

Helping you to make informed choices for you and your family

dorevitch pathology | Genomic Diagnostics

What is genetic carrier screening?

Genetic carrier screening is testing to identify an increased risk of having children with three of the most common inherited genetic disorders:

- **Cystic fibrosis**
- **Spinal muscular atrophy**
- **Fragile X syndrome**

One in 20 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 have a higher risk of having an affected child.

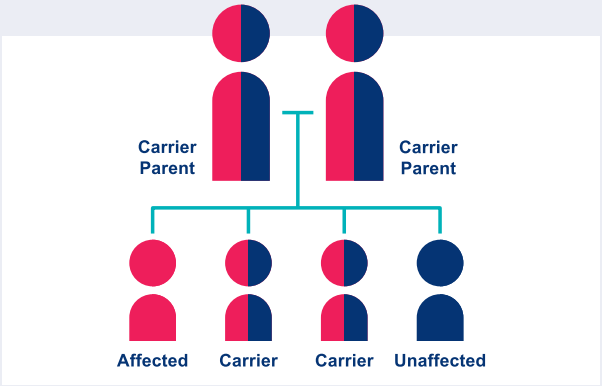
Why should I consider getting tested?

Genetic testing is the only way to find out whether or not you are a carrier for one of these conditions.

Knowing your carrier status can help you make informed choices. For many people, genetic carrier screening provides peace of mind. For those with a higher risk of having an affected child, it provides the opportunity to consider a range of options.

When should I get tested?

The best time to have carrier testing is before pregnancy, if possible. This gives 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.



Autosomal recessive inheritance

Genetic conditions screened

Cystic fibrosis, spinal muscular atrophy and fragile X syndrome can have devastating effects on life-expectancy and quality of life.

- Cystic fibrosis is the most common inherited disorder in Caucasians.
- Spinal muscular atrophy is the most common genetic cause of mortality in children under two.
- Fragile X syndrome is the most common form of inherited intellectual disability.

Cystic fibrosis and spinal muscular atrophy are autosomal recessive conditions, which means both parents must be carriers to have an affected child. Fragile X syndrome is an X-linked condition, so only the mother must be a carrier to have an affected child.

How do I get tested?

Genetic carrier screening involves getting a simple blood test at your local Dorevitch Pathology collection centre.

Available at more than 500 Dorevitch Pathology collection centres

We recommend that the female partner get tested first, followed by the male partner if required for cystic fibrosis or spinal muscular atrophy.

How does the screening work?

Our expert Genomic Diagnostics team will use the DNA from your blood sample to test for the most common genetic changes that cause cystic fibrosis, spinal muscular atrophy and fragile X syndrome.

If both partners are identified as carriers for cystic fibrosis or spinal muscular atrophy, or the female partner is identified as a carrier for fragile X syndrome, they are considered to be a carrier couple.

When the female partner is not identified as a carrier, the couple is considered to have a much lower risk of having an affected child.

Genetic counselling

If you and your reproductive partner are found to be a carrier couple for any of these conditions, genetic counselling is available. This will provide more information about the condition and enable in-depth discussions about your options, whether you are already pregnant or planning to have a child.

We provide genetic counselling free of charge for carrier couples when at least one partner has their test at one of our Dorevitch Pathology or Healius Pathology network collection centres.