Genomic Diagnostics

Genetic Carrier Screening

A guide for parents-to-be

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Genetic carrier screening

Genetic carrier screening is used to identify individuals and couples at increased chance of having children with a genetic condition by testing for genetic variants (changes) that can cause these conditions.

Most carriers of genetic conditions do not have any associated health concerns, and are often unaware that they carry a genetic variant. However, if one or both partners of a reproductive couple are carriers for a genetic condition, they are at increased chance of having affected children.

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 including:

- Natural pregnancy, with consideration of prenatal diagnosis (CVS or Amniocentesis)
- Pre-implantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test and then transfer embryos that are free of the condition
- The use of sperm or an egg from a donor who has been screened to ensure that they are not a carrier of the condition
- Adoption

Core Genetic Carrier screening with Genomic Diagnostics

Genomic Diagnostics offers the Core Genetic Carrier Screen, which looks for genetic variants in genes that can cause some of the most common genetic disorders in the Australian population: cystic fibrosis (CF), spinal muscular atrophy (SMA) and fragile X syndrome (FXS). Many children affected by these conditions are born to families with no history of these disorders.

How common are these conditions?

CF, SMA and FXS are amongst the most common inherited disorders in the Australian population:

Carrier	Frequency of carrier	Number of births per year
Cystic Fibrosis (CF)	1 in 25	120
Fragile X syndrome (FXS)	1 in 150	75 males 38 females
Spinal Muscular Atrophy (SMA)	1 in 40	35-50

What is Cystic Fibrosis (CF)?

Cystic fibrosis mainly causes breathing and digestion problems. People with CF have chronic and recurrent infections that cause damage to the lungs and gut. They may require frequent medical treatment and severely affected individuals have a reduced life expectancy.

What is Fragile X syndrome (FXS)?

Fragile X syndrome (FXS) is the most common inherited cause of intellectual disability. People with FXS can have intellectual disability, developmental delay, anxiety disorders, autism, ADHD, behavioural and learning challenges and various physical characteristics.

What is Spinal Muscular Atrophy (SMA)?

Spinal muscular atrophy (SMA) is a degenerative neurological disease that results in progressive muscle weakness. Over time, these weaknesses increase and can become life-threatening. It is the most common genetic cause of death in children under 2 years of age.

When should I be tested?

The ideal setting for carrier screening is during family planning, to have the most time to deal with all possible testing outcomes. However, testing can also be done during early pregnancy (ideally before 12 weeks). It is generally recommended the female partner is tested first should time permit as there may be no need for the male partner to get tested. You will only need to be tested once in your lifetime, as your carrier status does not change.

Should my partner be tested?

If an individual is identified as a carrier, their reproductive partner may need to have carrier testing. If partner testing is required, it should be organised by their doctor without delay.

What does it mean if I am identified as a carrier of a condition?

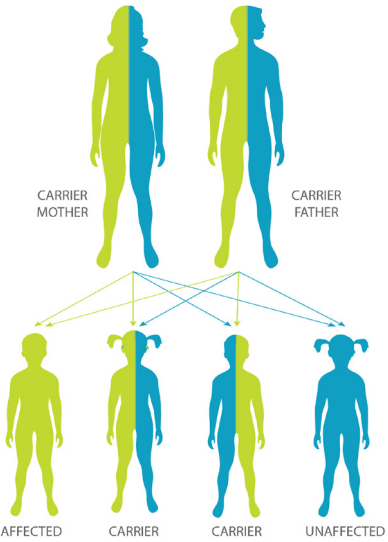
Every person has 22 sets of chromosomes plus 2 sex chromosomes (called X and Y), on which their genes are found. One chromosome from each set, and either X or Y, is inherited from each parent.

If one of your chromosomes has a gene change that causes CF, SMA or FXS you are a carrier of this condition.

- Cystic Fibrosis (CF) and Spinal Muscular Atrophy (SMA)

  A couple can only have a child with CF or SMA if both parents are carriers of the gene change for that condition and they each pass the gene change on. This means that if you are a carrier, your partner should also be offered testing. Two people who are carriers of the same condition have a 1 in 4 (25%) chance of having a child with the condition in each pregnancy they have together.

  CF and SMA carriers do not develop symptoms.

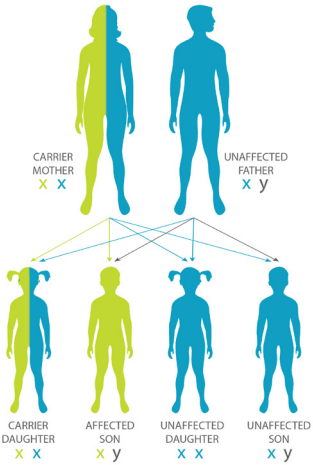


- Fragile X syndrome (FXS)

  FXS is carried on the X chromosome. Only women who are carriers for FXS will have a greater chance of having a child with the condition. This means that the male partner will not need to be tested for fragile X syndrome.

  Female carriers of FXS have a 1 in 2 (50%) chance of passing that gene onto each child they have. This is because females have two X chromosomes. Male children that inherit the gene change will have FXS. This is because males have an X and a Y chromosome. Female children can also be affected but usually have milder features.

  Some female carriers of FXS may develop fertility problems and go through menopause early (before 40 yrs). Some male and a small number of female carriers of FXS may develop a late-onset neurological condition which causes tremors and balance problems which worsen with age.



Counselling

If you or your partner are shown to be carriers for any of these conditions, genetic counselling is recommended to provide more information and enable detailed discussions regarding your reproductive options. This support is provided free of charge through Genomic Diagnostics.

Please refer to genomicdiagnostics.com.au for more information

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:** from 8 – 10 days following receipt of sample at the laboratory*
- **Accurate:** looks for the most common gene changes that cause CF, SMA and FXS in the Australian population

How do I get the Core Genetic Carrier Screen?

Step 1: See your doctor to get a request

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

Step 2: Prepare for your collection

Medicare and private health insurance do NOT cover the cost of testing. Prepay online via genomicdiagnostics.com.au. If you are unable to pay online contact our Customer Care Team on 1800 822 999.

Step 3: Have your sample collected

Write the receipt number on the request form and take it with you to your closest Healius Pathology collection centre to have your sample taken.

Step 4: See your doctor to get your results

Core Genetic Carrier Screen results will be delivered to your doctor 9-12 days from your sample arriving at the laboratory*

* Samples may take several days to reach the laboratory if coming from regional Australia, or over the weekend or a public holiday

Genomic Diagnostics

Healius Pathology Pty Ltd (ABN 84 007 190 043) t/a Genomic Diagnostics_139_GD_v2 (Sep21)