Carrier Screening

Genomic Diagnostics Genetic Carrier Screen test is performed to determine whether individuals or couples have a genetic change that may affect their chance of having a child with a genetic condition.

Genomic Diagnostics Genetic Carrier Screen tests for three relatively common genetic conditions in general populations: Cystic Fibrosis (CF), Spinal Muscular Atrophy (SMA) (both result from autosomal recessive inheritance) and Fragile X syndrome (FXS) (resulting from X-linked inheritance). Many children affected by these conditions are born to families with no history of disease.

Importantly, if couples are found to be carriers of these conditions, 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
- Adoption
- The use of a donor sperm or egg that has been screened to ensure that they are not a carrier of the condition.

WHAT TO EXPECT WHEN CONSIDERING CARRIER SCREENING:

- You and your medical practitioner will discuss carrier screening options
- Your doctor can request a Genetic Carrier Screen test on a standard referral form
- Make sure you tell your doctor if you are already pregnant or have a family history of any of the disorders you are being screened for, so that this can be noted on the request form
- **As this test is not Medicare rebated, please contact our Customer Care Team on 1800 822 999 to prepay for your test**
- Go to your nearest Dorevitch Pathology collection centre to have your blood collected
- This test takes approximately 2 weeks to perform and your results will be returned to your medical practitioner.
- **Patients who are identified as CF or SMA carriers by Genomic Diagnostics Genetic Carrier Screen can have their partners tested for the relevant condition free of charge†.**

COST*

Genetic Carrier Screen (GCS)	$345
Cystic Fibrosis (CF) Only	$190
Spinal Muscular Atrophy (SMA) Only	$195

Partner Testing†

Cystic Fibrosis (CF)	Free for partners of patients who are identified as carriers by GCS.*
Spinal Muscular Atrophy (SMA)	

*Prices are correct at October 2018 and are subject to change without notice.
† Free test offer valid at October 2018. This is subject to change without notice.

To talk to our friendly Customer Care team call

1800 822 999

between 9.00 am and 5.00 pm (EST)

or email us at

info@genomicdiagnostics.com.au

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Genomic Diagnostics

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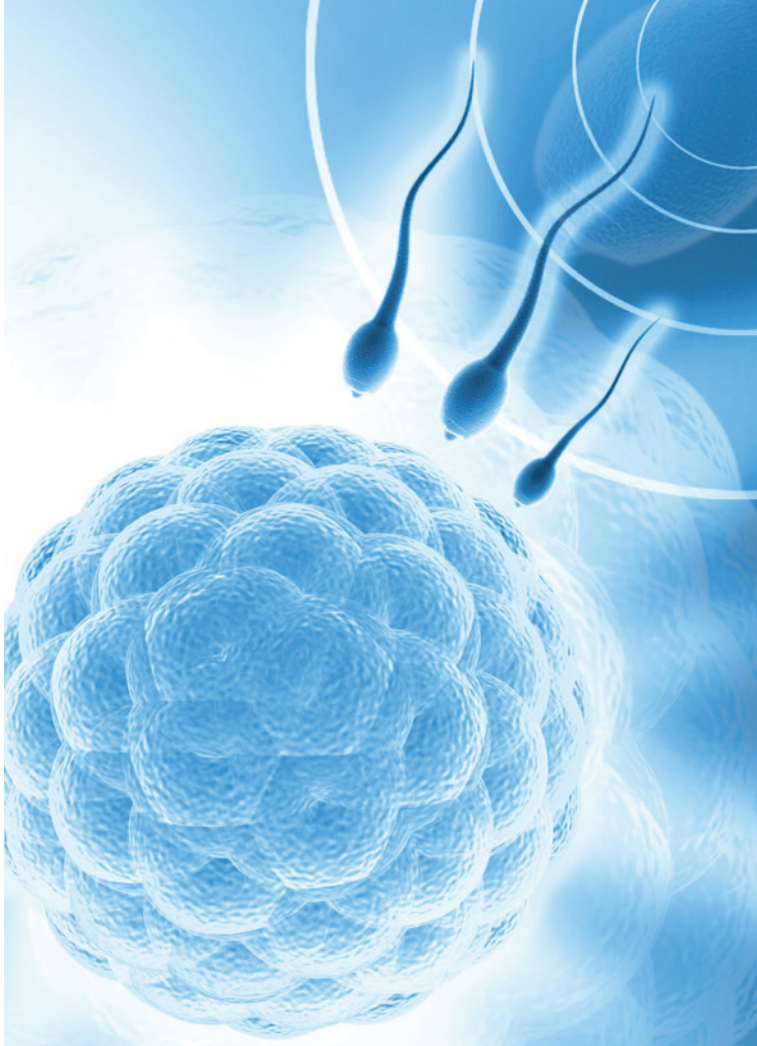

Genetic Carrier Screening

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LEADING THE WAY TO IMPROVE HEALTH

PATIENT INFORMATION

Conditions

WHAT IS CYSTIC FIBROSIS (CF)?

Cystic Fibrosis is a genetic condition that mainly causes breathing and digestion problems. People who have Cystic Fibrosis carry variant changes (mutations) in both copies of their CFTR genes. This gene makes an important protein that transports salt in and out of our cells. People with Cystic Fibrosis 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 a genetic condition that is the most common inherited cause of intellectual disability. People with Fragile X syndrome can have intellectual disability, developmental delay, anxiety disorders, autism, ADHD, behavioural and learning challenges and various physical characteristics. Features of Fragile X syndrome can vary from mild to severe, and whilst Fragile X syndrome occurs in both sexes, males are often more severely affected than females. There is no cure for Fragile X syndrome, however some behavioural, educational and medical interventions can improve outcomes.

WHAT IS SPINAL MUSCULAR ATROPHY (SMA)?

Spinal Muscular Atrophy (SMA) is a genetically inherited degenerative neurological disease that results in progressive muscle weakness. SMA affects both males and females. Over time, these weaknesses increase and can become life-threatening. Treatment options and support services are available to assist those living with SMA. There is no cure for SMA.

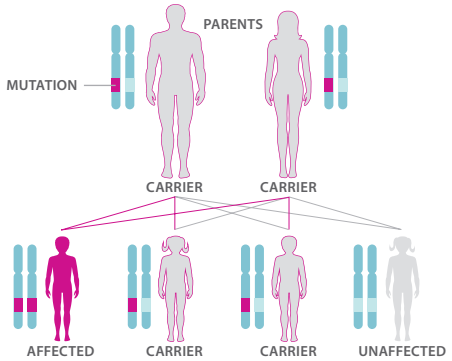
HOW COMMON ARE THESE CONDITIONS?

These three conditions combined are amongst the most commonly carried mutations in European populations:

Carrier	Frequency	Number of live births (per year)
Cystic Fibrosis (CF)	1 in 25	1 in 2,500
Fragile X syndrome	1 in 250	1 in 4,000 males (1 in 8,000 females)
Spinal Muscular Atrophy	1 in 49	1 in 6,000 – 1 in 10,000

WHAT IS AUTOSOMAL RECESSIVE INHERITANCE?

Genes come in pairs, one is inherited from each parent. Autosomal recessive is a mode of inheritance where an individual must have a genetic change in both copies of the specific disease gene pair, for them to have the genetic condition. Individuals who are carriers of the condition are healthy. They carry one changed copy of the specific disease gene pair and one normal copy of the gene.

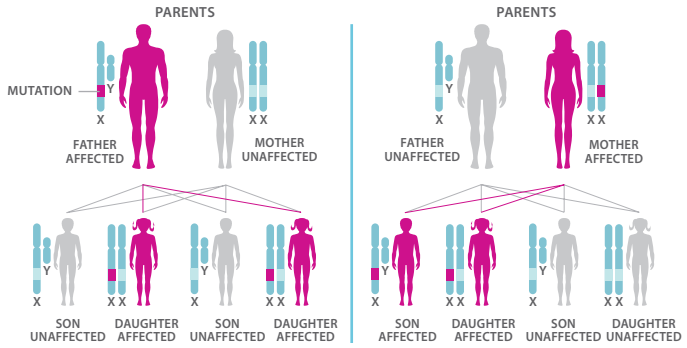


WHAT IS X-LINKED INHERITANCE?

X-linked is a mode of inheritance in which a genetic change on the X chromosome causes the expression of the genetic condition.

Males with an X-linked condition (i.e. Fragile X syndrome) are often more severely affected than females because males have only one X chromosome, it's pair is the Y chromosome; whereas females have two X chromosomes, only one of which carries the changed copy of the gene.

Because females have two X chromosomes, female carriers of X-linked conditions have a 1 in 2 (50%) chance of passing on their X chromosome with the gene change onto each child they have, and a 1 in 2 (50%) chance of passing on their X chromosome without the gene change.



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 pregnancy, ideally during the first three months of pregnancy.

WHAT DOES IT MEAN IF I AM IDENTIFIED AS A CARRIER OF A CONDITION?

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

If your test shows that you have one copy of the gene change for Cystic Fibrosis (CF) or Spinal Muscular Atrophy (SMA), you are a carrier of this condition. Being a carrier means you have a change in one of your two genes associated with CF or SMA. CF and SMA carriers do not develop symptoms.

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. If you are a carrier, your partner will 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.

Fragile X syndrome (FXS)

For Fragile X syndrome, only women who carry an expansion of in the FMR 1 gene have a greater chance of having a child with the condition. This means that your 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.

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.

Should my partner be tested?

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