Sonic Genetics
Quality is in our DNA

Male genetic carriers of Fragile X syndrome

Information for patients

The healthcare professional responsible for your care has given you this leaflet because you have been identified by Preconception Carrier Screening as a carrier of a change in the gene that causes Fragile X syndrome. This fact sheet contains more information about the particular genetic disorder mentioned in your report. We recommend that you also discuss your result with an experienced doctor or genetic counsellor. There is a separate leaflet for women who are shown to be carriers of this condition.

Each year, about one in 4,000 Australian boys and one in 8,000 Australian girls are born with Fragile X syndrome (FXS). This inherited genetic condition is the most common cause of intellectual disability and can range from learning difficulties through to severe developmental delay. People with FXS can also have anxiety, autism, epilepsy and a range of emotional and behavioural problems.

Genetic conditions are caused by inheriting faulty genes. Our genes are made of DNA and tell the body how to function, grow and develop. The genes are packaged into chromosomes; men have an X and a Y chromosome, while women have two copies of the X chromosome. The FXS gene (its scientific name is FMR1) contains information that is important for brain development. As the FXS gene is located on the X chromosome, women will have two copies of the FXS gene and men will only have one copy.

How to interpret a carrier result for Fragile X syndrome

Hearing about the carrier result will usually be unexpected and may lead to a range of physical and emotional reactions. In the midst of this emotional confusion, there may be some decisions that need to be made. You need as much information and support as possible when making these decisions. Genetic counsellors are experienced in providing support and information that will help you and your partner make an informed decision. The right decision is the one that is best for you and your family.

If your partner is not pregnant, you have time to think about the carrier test result when discussing your family planning options. Among these options is testing the baby in pregnancy for FXS (prenatal testing). Another is using IVF and then testing the embryo for FXS before implantation. However, having no testing for FXS at all is also an option.

If your partner is pregnant, your options include prenatal testing or no testing for FXS. Again, seeking support and information when making these decisions is important.

How does a person inherit Fragile X syndrome?

Variations in the length of part of the FXS gene can alter the information it provides to the brain cells. As the gene is passed down from parents to children over the generations, it can expand from the usual short length, through to being intermediate, medium and long. The length of the FXS gene, i.e. the type of FXS mutation, inherited will have different implications for you and your children. Your doctor will advise you which type of FXS mutation has been identified by Preconception Carrier Screening. It is important to discuss the risks summarised overleaf with your doctor or genetic counsellor.

Male genetic carriers of Fragile X syndrome

Information for patients

If you have a long FXS gene (a 'full mutation')

You have a long FXS gene on your X chromosome. You have no working copy of the FXS gene. You have some signs of the condition.

In **every pregnancy**, each of your daughters will inherit the full mutation and be genetic carriers of the full FXS mutation. They will have a normal short copy of the FXS gene on their other X chromosome. They may or may not be affected by FXS. Your sons will not inherit the full mutation from you, as they would inherit their X chromosome from their mother.

If you have a medium length FXS gene (a 'premutation')

You have an FXS premutation on your X chromosome. This is not associated with intellectual disability. Some men with a premutation develop a tremor and unsteady gait (ataxia) later in life. It is important to discuss this risk with your doctor or genetic counsellor.

In **every pregnancy**, each of your daughters will inherit the FXS premutation gene and be genetic carriers of the FXS premutation. They will have a normal short copy of the FXS gene on their other X chromosome. They will not have the condition but can pass the premutation onto their children. Each of your sons will not inherit the premutation from you, as they would inherit their X chromosome from their mother.

If you have an intermediate length FXS gene ('grey zone')

You have an intermediate length FXS gene on your X chromosome. This is not associated with intellectual disability or tremor/ataxia in later life.

In **every pregnancy**, each of your daughters will inherit the intermediate gene and will be a carrier of the FXS intermediate length gene. This gene works normally, and they will also have a normal short copy of the FXS gene on their other X chromosome. Your daughters will not have FXS. Their children may inherit an intermediate gene or it may expand to a premutation; they will not have FXS. Your sons will not inherit the intermediate length FXS gene, as they would inherit their X chromosome from their mother.

It is important to discuss this result with your doctor or genetic counsellor.

Your test result is important for your relatives

You probably inherited the abnormal FXS gene from your mother. That means that your brothers, sisters and maternal cousins, if you have any, also have a chance of being an FXS carrier. Telling them about the availability of the carrier test may help them make informed family planning decisions as well.

Sonic Genetics offers free genetic counselling for couples where the female partner is identified as a carrier for Fragile X syndrome. If you are eligible for this service, your doctor may refer you for genetic counselling. Conditions apply and can be discussed with your doctor.

Further information and contacts

The Centre for Genetics Education www.genetics.edu.au