Sonic Genetics
Quality is in our DNA

Female 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 men 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 you are 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 you are 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.

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

You have a long FXS gene on one of your X chromosomes and a short (normal) FXS gene on the other. You may be unaffected or have some signs of the condition.

In **every pregnancy** there is a one in four chance (25%) of having a son with the long FXS gene who will develop Fragile X syndrome; his only X chromosome contains the full FXS mutation. There is a one in four chance (25%) of having a daughter who is a genetic carrier of FXS with the full mutation; she may or may not be affected by the condition. There is a two in four chance (50%) that the child (son or daughter) will inherit the normal FXS gene and be unaffected.

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

You have an FXS premutation on one of your X chromosomes and a short (normal) FXS gene on the other. The FXS premutation is not a cause of intellectual disability. Some women will develop premature ovarian failure (before the age of 40). There is also a small chance of developing a tremor later in life. If you pass the premutation to your children, it can expand in length and can become a full mutation.

In **every pregnancy**, there is a one in four chance (25%) of having a son who will inherit either the premutation (no increased risk of intellectual disability) or a full mutation and have Fragile X syndrome. There is a one in four chance (25%) of having a daughter who will inherit either the premutation (no increased risk of intellectual disability) or a full mutation and may be affected by the condition. There is a two in four chance (50%) that the child (son or daughter) will inherit the normal FXS gene and be unaffected.

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

You have an intermediate length FXS gene on one of your X chromosomes and a short (normal) FXS gene on the other. This is not associated with intellectual disability, premature ovarian failure or the development of tremor in later life. If you pass the intermediate length FXS gene to your children, it can expand in length and can become a medium length FXS gene ('premutation').

In **every pregnancy**, there is a one in two chance (50%) of having a child (son or daughter) with an intermediate length FXS gene or a premutation. These children are unaffected. There is a one in two chance (50%) of having a child with the normal FXS gene. These children are also unaffected.

Your test result is important for your relatives

You probably inherited the abnormal FXS gene from one of your parents. That means that your brothers, sisters and 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