VCGS
Leaders in Genetic Health

Victorian Clinical Genetics Services
Murdoch Childrens Research Institute
The Royal Children's Hospital
Flemington Road, Parkville VIC 3052
P (03) 8341 6201 W vcgs.org.au

prepair™ fact sheet

Fragile X syndrome

What does this result mean?

The results of your test show that you are a carrier of a fragile X premutation or full mutation. This means that there is a chance that you could have a child with fragile X syndrome (FXS). The chance you will pass this gene change to a child is 1 in 2. The chance your child will be affected by FXS will vary depending on the exact size of your FXS gene.

What happens next?

Only female carriers of FXS are at increased chance of having a child with FXS. Therefore, your partner does not need to be tested.

- Your sample will be tested further to determine the exact size of your FXS gene. FXS is a complex genetic disorder and genetic counselling is recommended.

If you are not pregnant there are a number of family planning options available, which a genetic counsellor will discuss with you. This includes pre-implantation genetic diagnosis using in vitro fertilisation.

If you are pregnant a genetic counsellor will discuss prenatal testing for FXS. It is your choice whether or not to have further testing. However, prenatal testing can confirm if the pregnancy is affected.

What does prenatal testing involve? A prenatal diagnostic test will tell you if your baby has inherited the FXS gene change. Diagnostic testing involves passing a fine needle into the developing placenta or the amniotic fluid and drawing a small amount of sample into a syringe, under ultrasound guidance. The sample is sent to the laboratory for testing. The FXS testing results (often including full chromosome analysis) are ready in approximately 2-3 weeks. Diagnostic tests carry a small risk of miscarriage.

If your baby has inherited the FXS gene change: Your baby may either be a carrier of FXS (premutation) or may have FXS (full mutation). Your genetic counsellor will discuss this result with you. Depending on the result, you may need to face the difficult decision of whether to continue your pregnancy.

With prepair™, genetic counselling is available to you through the Victorian Clinical Genetics Services (VCGS). A genetic counsellor can talk with you about what your result means for you and your family. You can contact a genetic counsellor directly on 03 9936 6402. It is also advisable to discuss your results with your doctor.

What does my carrier result mean for my relatives?

FXS is an inherited condition which means it is likely some of your relatives may also be affected by FXS. FXS testing is available to your relatives and testing can be accessed through Victorian Clinical Genetics Services or through their local clinical genetics service.

Details of genetics services in Australia and New Zealand are available at: www.genetics.edu.au/Genetics-Services/genetic-counselling-services

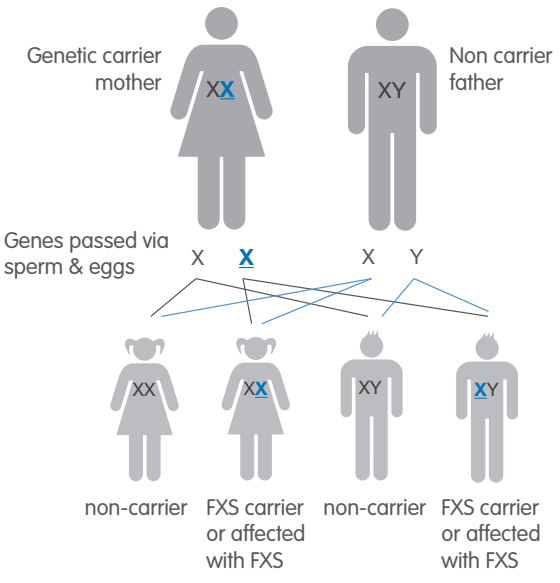
What is fragile X syndrome?

FXS is the leading cause of inherited intellectual disability. People with FXS can have developmental delay, learning difficulties, anxiety, autism and epilepsy. FXS is caused by a change in the length of the FXS gene (*FMR1*), located on the X-chromosome.

There are three classes of gene length: short (normal), medium (premutation) and long (full mutation). Females at risk of having a child with FXS carry either the premutation or full mutation.

Features of FXS vary from mild to severe with males more likely to be severely affected than females. Females with a premutation do not have FXS, but may experience early menopause (before age 40). Females with a full mutation may not show obvious features of FXS.

There is no cure for FXS, although there are some educational, behavioural and medical interventions that can improve outcomes for people with FXS.



Inheritance of fragile X syndrome.
X = normal size FXS gene
X = pre or full mutation FXS gene

Victorian Clinical Genetics Services
P +61 3 8341 6201
W vcgs.org.au

prepair™ genetic carrier screening

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