
Fragile X syndrome

Summary

- Fragile X syndrome is the most common inherited cause of intellectual disability.
 - Fragile X syndrome is the most common known single-gene cause of autism.
 - About 1 in 3,600 boys and 1 in 4,000–6,000 girls have Fragile X syndrome.
 - Every week in Australia, a child is born with Fragile X syndrome.
 - The effects of Fragile X syndrome vary widely but most people experience lifelong difficulties.
 - Both men and women can be carriers of the Fragile X gene and be at risk of having one or more children with Fragile X syndrome.
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Fragile X syndrome is a genetic disorder caused by a change to one of the genes on the X chromosome. It is the most common inherited cause of intellectual disability and the most common single gene cause of autism. Fragile X syndrome affects around 1 in 3,600 boys and between 1 in 4,000 – 6,000 girls. Fragile X syndrome causes a wide range of physical, developmental, behavioural and emotional difficulties although the level of severity between people with the condition varies enormously.

Effects of Fragile X syndrome

Fragile X syndrome can cause a range of physical, developmental, behavioural and emotional difficulties in people. Although there is no typical presentation, some boys with Fragile X syndrome have a long narrow face, prominent forehead and large testicles.

The most significant effects of Fragile X syndrome are:

- global developmental delay, including speech
- language and communication difficulties
- intellectual disability and learning problems
- anxiety
- autistic-like behaviours such as hand flapping, repeating words and sentences, and difficulty with social interactions
- ADHD
- poor eye contact
- difficulty processing sensory information.

The level of severity of these effects varies considerably and not every person with Fragile X syndrome will have every one of these characteristics.

Cause of Fragile X syndrome

Fragile X syndrome is caused by a change to a gene on the X-chromosome called the FMR1 gene. This gene produces a protein that helps the brain to function normally, but if this gene is changed or altered in any way, it cannot produce its normal protein, which can result in Fragile X syndrome.

Fragile X syndrome is inherited in a way that is known as 'X-linked', as the changed gene is on the X chromosome. This means that men with Fragile X syndrome are often more severely affected than women. This is because men only have one X chromosome, whereas women have two X chromosomes, only one of which is changed.

Pre-mutation carriers of the Fragile X gene

Although Fragile X syndrome is not that common, affecting around 1 in 3,600 boys and between 1 in 4,000 – 6,000

girls, the number of men and women who are carriers of the Fragile X gene is significantly higher. It is estimated that 1 in 150 women and 1 in 800 men are carriers of the gene. These people are known as 'Fragile X pre-mutation carriers'. Pre-mutation carriers may not have any symptoms of Fragile X syndrome but they are at risk of passing on the changed FMR1 gene and having a child with Fragile X syndrome.

People who are Fragile X pre-mutation carriers are also at risk of developing health problems later in life. Around 20 per cent of female Fragile X pre-mutation carriers experience reduced fertility or early menopause. This is called Fragile X-associated Primary Ovarian Insufficiency, or FXPOI. Both male and female Fragile X pre-mutation carriers are also at risk of developing a neurological condition called FXTAS, or Fragile X-associated Tremor Ataxia syndrome. Problems with balance and gait, tremors and gradual intellectual decline are common in FXTAS. These conditions are called Fragile X-associated Disorders.

Testing and diagnosis of Fragile X syndrome

Fragile X syndrome (and Fragile X-associated Disorders) can only be diagnosed by DNA testing – usually by a blood test but sometimes via cheek swab or mouthwash. If possible, testing should be done at a recognised genetics service [<http://www.genetics.edu.au/Genetics-Services/genetic-testing-services>]. If there is a diagnosis of Fragile X syndrome, it is important that immediate and extended family members are also tested to identify others who may have the changed FMR1 gene. This is called cascade testing.

DNA testing is recommended for:

- people with a family history of Fragile X syndrome or intellectual disability
- people with intellectual disability, developmental delay or learning disability together with features of Fragile X syndrome such as anxiety, ADHD or autistic behaviour
- men or women over 50 with balance or gait problems, tremor or dementia
- any woman with problems with fertility or early menopause (under 40)
- women with family history of primary ovarian insufficiency (loss of function of the ovaries before age 40) for testing before or during pregnancy.

Unfortunately diagnosis of Fragile X syndrome is often delayed and it is not uncommon for families to have had a second child born with Fragile X syndrome before the first child has been diagnosed. A prompt diagnosis of Fragile X syndrome is important so that families can get access to appropriate early intervention services to help their child reach their full potential.

Genetic counselling services and Fragile X syndrome

The facts about Fragile X syndrome are complicated and the ramifications for families can be serious. It is recommended that parents and family members ask their doctor to refer them to a genetics service, both for testing and follow-up counselling. The genetics counsellor can also provide information about the range of reproductive options that are now available.

Clinics in Victoria are located in metropolitan Melbourne and major regional centres. Contact **Victorian Clinical Genetics Services** for more information about Fragile X syndrome or to organise an appointment.

Where to get help

- Your doctor
- Paediatrician
- **Fragile X Association of Australia** Tel: 1300 394 636 (freecall) or E: support@fragilex.org.au
- **Victorian Clinical Genetics Services** Tel: (03) 8341 6201
- **Fragile X Alliance Clinic** Tel: (03) 9528 1910
- **Genetic Support Network Victoria** Tel: (03) 8341 6315 or E: info@gsnv.org.au
- **Carers Victoria** Tel: 1800 242 636 (freecall) or E: reception@carersvictoria.org.au

Things to remember

betterhealth.vic.gov.au

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- Fragile X syndrome is the most common known single-gene cause of autism.
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This page has been produced in consultation with and approved by:

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