

Fragile X syndrome

Fragile X syndrome is a genetic disorder caused by an alteration in the X chromosome (that is, a change in the DNA structure). It results in a wide range of developmental, physical and behavioural problems, and is the most common known cause of inherited intellectual disability. Prevalence estimates for Fragile X syndrome vary – the best estimate is that about one in 4,000 males has this syndrome.

Fragile X syndrome varies in its presentation

The most significant effects of Fragile X syndrome are:

- Developmental delay
- Intellectual disability
- Behavioural problems
- Mimicking.

The degree of intellectual disability can vary from mild learning difficulties through to severe intellectual impairment. Behavioural problems are present in some Fragile X individuals. These effects tend to be more severe in males than in females.

Characteristics of Fragile X syndrome

Some children with Fragile X syndrome will display behaviours similar to those of children with autism, including:

- Hand flapping
- Repeating of words and sentences.

Fragile X males may have certain physical features, including:

- Large, prominent ears
- Enlarged testes
- Flexible joints.

Not all of these characteristics are seen in every person with Fragile X syndrome.

Chromosomes explained

Chromosomes are located in the cells of the human body and contain all our genetic information. In every cell, there are 23 pairs of chromosomes, one pair of which determines the sex of a person. A male has an X and a Y, a female has two X chromosomes. A child will receive one of their sex chromosomes from the father's sperm (X or Y) and one from the mother's egg (X).

Fragile X syndrome is inherited

The Fragile X syndrome is inherited in a way known as 'X-linked'. The altered gene is on the X chromosome.

On the X chromosome, there is a gene called FMR1, which produces a protein that helps the brain to function normally. If this gene is altered, it cannot produce its normal protein. Our genes often contain regions that are repeated. In the 'normal' form of the gene, there will be between five and 55 repeated copies of a small section of the gene. In a person with Fragile X syndrome, this copy number is increased to more than 200 repeats (called a 'full mutation'). When there are more than 200 repeats, the gene is switched off, leading to the symptoms seen in Fragile X syndrome.

A repeat number of between 55 and 200 is classed as a 'pre-mutation'. The number of repeats may increase when the mother passes on her X chromosome to her child. Therefore, an apparently unaffected woman – with fewer than 200 repeats – may have an affected son or daughter with greater than 200 repeats.

Other effects of Fragile X premutations

In the past it was thought that the only significance of a person having a Fragile X premutation was that women with permutations were at risk of having a child with Fragile X syndrome. Recently it has been discovered that Fragile X permutations can have additional health effects. Approximately 20 per cent of females with a Fragile X premutation experience premature menopause (menopause prior to age 40 years), and males (and some females) with permutations are at risk of a late onset neurological disorder called Fragile X Tremor Ataxia syndrome (FXTAS).

Counselling is available

The facts about Fragile X are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic. The genetics clinic can provide diagnostic and counselling services, including information about reproductive options. Clinics are held in Victoria in metropolitan Melbourne, Geelong, Ballarat, Bendigo, Albury/Wodonga, Mildura, Shepparton, Warragul, Warrnambool, Frankston and Sale/Traralgon. Contact Genetic Health Services Victoria for more information about Fragile X syndrome or to organise an appointment.

Where to get help

- Your doctor or paediatrician
- Genetic Health Services Victoria Tel. (03) 8341 6201
- Fragile X Alliance Clinic Tel. (03) 9528 1910

Things to remember

- Fragile X is the most common cause of inherited intellectual disability.
- Fragile X affects males more than females.
- The children of unaffected carriers may be affected by Fragile X syndrome.
- Genetic counselling is available and recommended.

This page has been produced in consultation with, and approved by:

Genetic Health Services Victoria

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