

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 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

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
- Very 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, this copy number is increased to more than 200 repeats, so that the gene is not active and leads to the symptoms seen in Fragile X.

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 with greater than 200 repeats.

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 on the number below 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
- Victorian Fragile X Support Group Tel. 9569 0858

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.
- Genetic counselling is available and recommended.

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

Murdoch Children's Research Institute

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