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.

**Effects of Fragile X syndrome**

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 people with Fragile X. 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, and 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).

**Cause of Fragile X syndrome**

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.

**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, and major regional centres. Contact Victorian Clinical Genetics Services (VCGS) for more information about Fragile X syndrome or to organise an appointment.

**Where to get help**

- Your doctor
- Paediatrician
- The Fragile X Association of Australia Helpline Tel. 1300 394 636
- Victorian Clinical Genetics Services Tel. (03) 8341 6201
- Fragile X Alliance Clinic Tel. (03) 9528 1910
- Better Start for Children with a Disability Tel. 1800 242 636

**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:**

Victorian Clinical Genetics Services (VCGS)

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