Fragile X syndrome

Fragile X syndrome is a genetic condition involving changes in a gene on the X chromosome. It is the most common form of inherited intellectual disability in boys.

Causes

Fragile X syndrome is caused by a change in a gene called *FMR1*. A small part of the gene code is repeated several times in one area of the X chromosome. The more repeats (also called gene expansion), the more likely the condition will occur.

The *FMR1* gene makes a protein needed for your brain to function properly. A defect in the gene makes your body produce too little of the protein, or none at all.

Boys and girls can both be affected, but because boys have only one X chromosome, a single fragile X expansion is likely to affect them more severely. You can have fragile X syndrome even if your parents do not have it.

A family history of fragile X syndrome, developmental problems, or intellectual disability may not be present.

Symptoms

Behavior problems associated with fragile X syndrome include:

- Autism spectrum disorder
- Delay in crawling, walking, or twisting
- Hand flapping or hand biting
- Hyperactive or impulsive behavior
- Intellectual disability
- Speech and language delay
- Tendency to avoid eye contact

Physical signs may include:

- Flat feet
- Flexible joints and low muscle tone
- Large body size

- Large forehead or ears with a prominent jaw
- Long face
- Soft skin

Some of these problems are present at birth, while others may not develop until after puberty.

Family members who have fewer repeats in the *FMR1* gene may not have intellectual disability. Women may have premature menopause or difficulty becoming pregnant. Both men and women may have problems with tremors and poor coordination.

Exams and Tests

There are very few outward signs of fragile X syndrome in babies. Some things that your health care provider may look for include:

- Large head circumference in babies
- Intellectual disability
- Large testicles after the start of puberty
- Subtle differences in face features

In females, excess shyness may be the only sign of the disorder.

Genetic testing can diagnose this disease.

Treatment

There is no specific treatment for fragile X syndrome. Instead, training and education have been developed to help affected children function at the highest possible level. Clinical trials are ongoing (www.clinicaltrials.gov [https://www.clinicaltrials.gov]) and looking at several possible medicines for treating fragile X syndrome.

Support Groups

More information and support for people with Fragile X condition and their families can be found at:

- National Fragile X Foundation -- fragilex.org [https://fragilex.org/]

Outlook (Prognosis)

How well the person does depends on the amount of intellectual disability.

Possible Complications

Complications vary, depending on the type and severity of symptoms. They may include:

- Recurrent ear infections in children
- Seizure disorder

Fragile X syndrome can be a cause of autism or related disorders, although not all children with fragile X syndrome have these conditions.

Prevention

Genetic counseling may be helpful if you have a family history of this syndrome. All pregnant women and those planning a pregnancy should be offered genetic screening for these conditions.

Alternative Names

Martin-Bell syndrome; Marker X syndrome

References

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