CONDITION

Fragile X syndrome

Fragile X syndrome is an inherited condition that affects development, learning, behaviour, and physical features.

WHAT IT IS
Inherited condition affecting development, learning, behaviour, and physical features

BODY SYSTEM
Nervous system and development

MAIN SYMPTOMS
Developmental delay, learning disability, anxiety, attention difficulties

WHAT CAUSES IT
Inherited change in the FMR1 gene

HOW IT IS DIAGNOSED
FMR1 DNA testing, usually from blood

MAIN TREATMENT
Developmental, educational, behavioural, and symptom-based support

CAN IT BE CURED?
No - the genetic change remains lifelong

IS IT AN EMERGENCY?
No, except prolonged seizures or immediate safety risks

LONG-TERM OUTLOOK
Lifelong differences; many people continue building skills

Overview

Fragile X syndrome is an inherited condition that affects brain development. It can influence learning, speech, attention, emotions, behaviour, and social communication. Some people also have recognisable physical features or medical concerns.

The condition is caused by a change in the FMR1 gene on the X chromosome. This change reduces or stops production of a protein needed for healthy brain development.

Effects vary widely. Some people have mild learning or emotional difficulties. Others have an intellectual disability and need substantial help with communication, education, daily activities, or safety.

Symptoms are often greater in boys and men because they usually have one X chromosome.

Girls and women usually have two, so the unaffected copy may partly compensate. However, some girls and women have significant support needs.

Fragile X is associated with autism, but they are not the same condition. A person may have fragile X syndrome without autism or meet the criteria for both.

Symptoms

Signs may not be obvious at birth. Concerns often emerge during infancy or early childhood as speech, movement, behaviour, and social skills develop.

Developmental and learning features may include:

Behavioural and emotional features can include a short attention span, impulsivity, anxiety, sensory sensitivity, and distress when routines change. Repetitive movements, such as hand flapping or hand biting, may occur. Some people avoid eye contact, particularly when anxious or overstimulated.

Autism-related differences may include difficulty with social communication, intense interests, repetitive behaviour, and a strong preference for predictability. A separate assessment is needed if an autism diagnosis is being considered.

Physical features are often subtle in young children and may become more noticeable with age. They can include a long, narrow face, prominent ears, flexible finger joints, flat feet, and a high-arched roof of the mouth. Boys may develop enlarged testicles after puberty. Appearance alone cannot confirm the condition.

Associated concerns include recurrent ear infections, hearing problems, crossed eyes, seizures, sleep difficulties, constipation, feeding problems, and loose joints. Not everyone develops them.

- Delayed speech, language, sitting, walking, or toilet training
- Intellectual disability or specific learning difficulties
- Difficulty planning, solving problems, or changing activities
- Stronger practical or visual skills than language-based skills

Causes and risk factors

Fragile X syndrome usually results from an expanded section of DNA in the FMR1 gene containing a repeated pattern called a CGG repeat .

In a full mutation, the pattern is usually repeated more than 200 times. This commonly switches off the gene through methylation, leaving the body with little or none of the FMRP protein that supports connections between brain cells.

The altered gene can pass through families. A smaller expansion, called an FMR1 premutation , does not usually cause fragile X syndrome. However, it can expand into a full mutation when passed from a mother to a child.

Some premutation carriers develop related conditions. These include fragile X-associated primary ovarian insufficiency, which can affect menstrual periods and fertility, and fragile X-associated tremor/ataxia syndrome, which affects movement and balance, particularly in older men.

A family may have no previous diagnosis because a premutation can pass silently through generations before expanding.

Diagnosis

Fragile X syndrome is confirmed with an FMR1 DNA test , usually using blood. It measures the number of CGG repeats and may assess whether methylation has switched off the gene.

Testing may be suggested for unexplained developmental delay, intellectual disability, autism-related features, or a family history linked to fragile X. It may also be considered when a relative has an FMR1 premutation or full mutation.

Standard chromosome tests, chromosome microarray testing, and many forms of exome sequencing do not reliably detect the usual FMR1 expansion. A specific fragile X test may therefore be needed even when other genetic tests were unremarkable.

Genetic counselling can explain results described as a typical repeat size, intermediate result, premutation, or full mutation, including possible implications for biological relatives.

Carrier testing, prenatal testing, and testing embryos created through in vitro fertilisation are available in some settings. These are personal choices, and genetic counselling can provide balanced information.

Treatment and management

There is no treatment that removes the FMR1 change or cures fragile X syndrome. Care supports development, reduces distress, treats associated concerns, and helps the person participate in family, school, work, and community life.

Early support may include speech and language therapy, occupational therapy, physiotherapy, and help with play or social communication. Services should reflect the person's abilities and priorities.

At school, an individual learning plan may provide structured teaching, visual instructions, movement breaks, a quieter setting, or extra time. Predictable routines and preparation for changes can reduce anxiety.

Behavioural and psychological support can address anxiety, attention difficulties, sleep, aggression, or self-injury. Assessment should consider communication difficulties, pain, sensory overload, and environmental demands rather than treating behaviour as deliberate misconduct.

Medicines do not treat fragile X itself but may be considered for attention-deficit/hyperactivity disorder, severe anxiety, mood symptoms, sleep problems, or seizures. Benefits and side effects require individual assessment.

Regular care may include checks of hearing, vision, growth, dental health, joints, sleep, and emotional well-being. Seizures, persistent digestive problems, and heart findings are treated according to the specific problem.

Complications

Fragile X can affect education, communication, relationships, employment, and independent living. Support needs vary widely.

Anxiety and sensory overload may make busy places, transitions, medical appointments, or unexpected changes difficult. Communication barriers can make pain or illness harder to recognise, and a behaviour change may be the first sign of a medical problem.

Seizures occur in some people and often respond to standard treatment. Sleep disruption, hearing loss from repeated ear problems, and vision difficulties can interfere with learning if unrecognised.

Loose connective tissue can contribute to flexible joints, flat feet, or, less commonly, heart valve changes. These may require only observation unless they cause symptoms.

Families may face pressures involving caregiving, education, finances, or planning for adulthood. Support for parents, siblings, and other caregivers can be an important part of care.

Prevention

Fragile X syndrome cannot be prevented through lifestyle changes because it is inherited.

If an FMR1 change is known in the family, genetic counselling can explain the chance of passing it on. Reproductive options may include pregnancy without genetic testing, prenatal testing, in vitro fertilisation with embryo testing, donor eggs or sperm, and adoption. Availability and personal views differ.

Testing relatives can identify premutation carriers who may want information about reproductive choices or related adult health concerns. Predictive or carrier testing is a personal decision, particularly for children without symptoms.

Living with it and what to expect

Fragile X syndrome is lifelong, but it does not usually cause steady loss of previously learned skills. Children and adults can continue developing communication, self-care, social, and practical abilities, although progress may be uneven.

Support needs change with age. Childhood may focus on communication, education, sensory needs, and behaviour. Adolescence can bring anxiety, social pressures, and challenges related to puberty. Adult planning may cover work, housing, transport, relationships, money, and support with decisions.

Some adults live with limited assistance, work in paid or supported employment, and participate in community life. Others need daily supervision or lifelong help with personal care and safety. Childhood test results cannot precisely predict adult independence.

Strengths may include visual memory, humour, empathy, practical interests, and strong attachment to familiar people. Plans that build on these strengths can be more effective than focusing only on difficulties.

Do not automatically attribute major changes to fragile X. New loss of skills, reduced mobility, pain, severe sleep disruption, or marked behaviour change needs assessment for another medical or emotional cause.

When to get medical help

Arrange routine medical follow-up for concerns about development, speech, learning, attention, anxiety, hearing, vision, sleep, feeding, or bowel habits. Assessment is also appropriate if fragile X or an FMR1 premutation has been found in a biological relative.

Seek prompt medical advice for a first suspected seizure, new loss of established skills,

persistent pain, a major unexplained behaviour change, or medication side effects. These concerns require assessment but are not always emergencies.

Call emergency services if:

Follow any existing seizure or crisis plan.

- A seizure lasts five minutes or longer
- Seizures repeat without recovery between them
- Breathing is difficult or the person does not recover as expected
- There is an immediate risk of serious self-harm or harm to others

Common questions

Is fragile X syndrome the same as autism?

No. Fragile X is caused by an FMR1 gene change. Autism is diagnosed from patterns of social communication and behaviour. Some people with fragile X are also autistic, while others are not.

Can someone have mild fragile X syndrome?

Yes. Effects can be mild, particularly in some girls and women. A mildly affected person may have learning difficulties, anxiety, attention problems, or social challenges without a major intellectual disability.

Does fragile X get worse with age?

Fragile X is not usually progressive and does not typically cause steady loss of brain function. Skills and support needs can change, but new neurological decline should be assessed rather than assumed to be part of the syndrome.

Can fragile X syndrome be missed on genetic testing?

Yes, if the correct test was not performed. Chromosome microarray and many sequencing tests do not reliably detect the usual repeat expansion. Confirmation requires a test designed to analyse the FMR1 gene.

Can a person with fragile X have children?

Some people with fragile X can have biological children. Fertility, ability to consent, support needs, and the chance of passing on an FMR1 change require individual discussion. Genetic and reproductive counselling can explain the options.

What is the difference between fragile X syndrome and a premutation?

Fragile X syndrome usually results from a full mutation that switches off the FMR1 gene. A premutation is a smaller expansion that usually does not cause the syndrome, but it can be inherited, may expand in a future generation, and can cause other health conditions in some carriers.

Sources

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