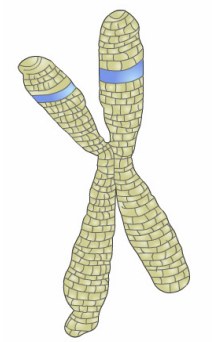
Fragile X Syndrome

Introduction

Fragile X syndrome is the most common form of inherited intellectual disability. A problem with a specific gene causes the disease. People with only a small change in the gene might not show any signs of fragile X. People with bigger changes can have severe symptoms. Getting treatment early for fragile X can help.



FMR1 Gene

This reference summary explains what fragile X syndrome is and what causes it. It also covers how fragile X is diagnosed and the available treatment options.

What is Fragile X Syndrome?

Fragile X syndrome is the most common form of inherited intellectual and developmental disability. Fragile X happens when there is a change, or mutation, in a single gene called the FMR1 gene.

The FMR1 gene normally makes a protein the body needs for the brain to develop. But when there is a change in this gene, the body makes only a little bit or none of the protein. This can cause the symptoms of fragile X.

Fragile X is inherited, which means it is passed down from parents to children. Parents can have children with fragile X even if the parents do not have fragile X themselves. The changes in the gene can become more serious when passed from parent to child.

Some people may only have a small change in their FMR1 gene, called a permutation. These people may not show any signs of fragile X. Other people may have bigger changes in the gene, called a full mutation, that cause the symptoms of fragile X syndrome.

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Fragile X syndrome can affect both males and females. However, females often have milder symptoms than males. This is because females have 2 X chromosomes while boys have 1 X and 1 Y chromosome. A single fragile X is likely to affect them more severely.

Symptoms

Not everyone with fragile X has the same signs and symptoms, but they do have some things in common. This section explains some common signs of fragile X.

Many people with fragile X have intellectual disabilities. These problems can range from mild learning disabilities to more severe intellectual and developmental disabilities.

Most children with fragile X have some behavioral challenges. They may be afraid or anxious in new situations. Many children, especially boys, have trouble paying attention or may be aggressive. Girls may be shy around new people.

Most boys with fragile X have some problems with speech and language. They may have trouble speaking clearly. They may stutter or leave out parts of their words.

Boys may also have problems understanding "clues" when talking to other people, such as understanding the speaker's tone of voice or body language. Girls usually do not have severe problems with speech or language.

Many children with fragile X are bothered by certain sensations, such as bright light, loud noises or the way something feels. Some do not like to be touched. They may have trouble making eye contact with other people.

Teens and adults with fragile X may have long ears, faces and jaws. Many people with fragile X may also have loose, flexible joints. They may have flat feet and be able to extend joints like the thumb, knee and elbow further than normal.

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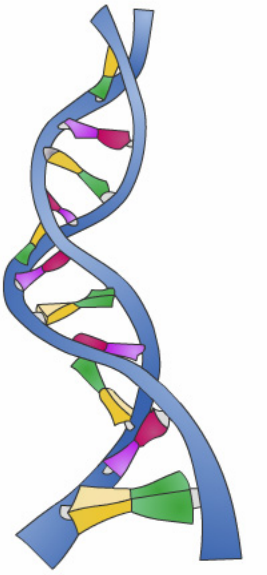
Causes

Each cell in the human body contains thousands of genes. Genes tell the body to make certain substances. Every person has thousands of genes. Our genes make us look the way we do. They also have a lot to do with our health.

Fragile X syndrome is caused by a change in the Fragile X Mental Retardation 1, or FMR1, gene. The FMR1 gene makes a protein called Fragile X Mental Retardation protein, or FMRP, that is needed for normal brain development.

Genes are found on chromosomes. Every non-reproductive human cell contains 23 pairs of chromosomes. People get their chromosomes from their parents. People get 1 of each pair of chromosomes from their mother and 1 of each pair from their father.



The chromosomes that form the 23rd pair are called the sex chromosomes. They decide if a person is male or female. Females have 2 X chromosomes, or XX. Males have 1 X and 1 Y chromosome, or XY. The FMR1 gene is on the X chromosome.

The chromosomes and genes have a special code called DNA. DNA has 4 chemical letters, called "bases": A, C, T and G. The order of the letters determines the information carried in each gene, like the way that a specific pattern of letters makes up the words in a sentence.

There is a place in the FMR1 gene where the DNA pattern CGG is repeated over and over again. In most people, the number of repeats is small, which is normal. If the number of repeats is too large, the gene turns off. This is called a "trinucleotide repeat disorder." People inherit the disorder from their parents.

When the gene is turned off, no protein is made. Without the protein, the person develops fragile X syndrome.

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Diagnosis

Fragile X syndrome is not usually diagnosed until a child is school age. The first clue in children often is a delay in reaching one or more developmental milestones.

Developmental milestones are things most children can do by a certain age. For example, most children can walk without help by 15 months of age.

Fragile X syndrome can be diagnosed with a blood test. Testing can show changes in the FMR1 gene.

A diagnosis of fragile X syndrome can be helpful to the family because it can provide a reason for a child's intellectual disabilities and behavior problems. This allows the family and other caregivers to learn more about the disorder and manage care so that the child can reach his or her full potential.

Anyone who is thinking about fragile X syndrome testing should consider having genetic counseling prior to getting tested. The genetic counselor determines whether a condition in the family may be genetic and estimates the chances that another relative may be affected.

Genetic counselors offer and interpret genetic tests that may help to estimate risk of disease. The genetic counselor also provides psychological counseling to help families adapt to their condition or risk.

Treatment

There is no cure for fragile X syndrome. However, there are a variety of ways to help minimize the symptoms of the condition.

The symptoms of fragile X syndrome are most often treated using a combination of the following:

- Special education.
- Behavioral and physical therapy.
- Medication.

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Children with fragile X who receive appropriate education, therapy and medication have the best chance of using their individual capabilities and skills. Even those with significant intellectual disability can learn to master many self-help skills.

The sooner a child begins to get help, the more opportunity there is for learning. Because a young child's brain is still forming, early intervention gives children the best start possible and the best chance of developing their full potential.

Speech-language therapists can help fragile X patients that have speech problems. The therapists work on improving pronunciation of words and sentences, slowing down speech and using language more effectively. They may set up social or problem-solving situations to help a child practice using language in meaningful ways.

Rarely, some children with fragile X syndrome may not be able to speak well enough to communicate with others. For these children, nonverbal ways of communication may be taught.

Occupational therapists help find ways to adjust tasks to match a person's needs and abilities. For example, this type of therapist might help a teenager with fragile X identify a job, career or skill that matches his or her interests and individual capabilities.

Physical therapists design activities and exercises to build motor control and to improve posture and balance. They can teach parents ways to exercise their baby's muscles.
At school, they may help a child who is easily over-stimulated or who avoids body contact participate in sports and games with other children.

Behavioral therapists try to identify situations in which a child acts in a negative way and then seeks ways to prevent or cope with these stressful occasions.
These therapists also work with parents and teachers to find useful responses to desirable and undesirable behavior.

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Currently, there is no medication that can cure fragile X. No drugs have been approved specifically for the treatment of fragile X or its causes. But in many cases, medications have been used to treat the symptoms of fragile X.

Medications may be used to treat:

- Aggression.
- Attention problems.
- Mood changes.
- Seizures.
- Sensory overstimulation.
- Sleep disturbances.
- Other symptoms.

Even if treatment for fragile X doesn't begin until late childhood or adulthood, it is never too late to benefit from treatment.

Summary

Fragile X syndrome is the most common form of inherited intellectual disability. A problem with the FMR1 gene causes the disease. When there is a change in this gene, the body makes only a little bit or none of a certain protein, which can cause the symptoms of fragile X.

Fragile X syndrome affects both males and females. However, females often have milder symptoms than males. Many people with fragile X have intellectual disabilities. These problems can range from mild learning disabilities to more severe intellectual and developmental disabilities. Most children with fragile X have some behavioral challenges. They may be afraid or anxious in new situations.

Fragile X syndrome is not usually diagnosed until a child is school age. The first clue in children often is a delay in reaching one or more developmental milestones. Fragile X syndrome can be diagnosed with a blood test. There is no cure for fragile X syndrome. However, there are a variety of ways to help minimize the symptoms of the condition.

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