

**CALIFORNIA DEPARTMENT OF EDUCATION
DIAGNOSTIC CENTER, SOUTHERN CALIFORNIA**

FRAGILE X SYNDROME

Fragile X syndrome is a genetic disorder that affects mental development, physical appearance, and emotions. It is the most common hereditary form of intellectual disability. It gets its name from an abnormal appearance of the X chromosome during special chromosome culture. It is usually caused by an abnormality in a single gene, referred to as the FMR1 gene. Fragile X syndrome is seen most often in boys.

MENTAL DEVELOPMENT

Most children with Fragile X syndrome have a mild to moderate intellectual disability (mild to moderate mental retardation). A small number will have learning disabilities without an overall reduced mental ability. In addition, the tested IQ profile may change over time. A substantial percentage of boys will have a drop in their IQ scores during the prepubertal and early puberty years, and boys initially identified as having mild intellectual disability when they are young, may end up with moderate intellectual disability. This appears to be due to a slowing of development starting around age 10, not a loss of skills.

Children with Fragile X tend to have a particular intellectual profile, regardless of their intellectual level. Most boys have delayed language development, and continue to be better at processing visual information than verbal information, especially when the visual information is meaningful, rather than abstract. There are also common weaknesses in understanding and using numbers. Arithmetic skills frequently lag well behind reading and spelling when children are acquiring basic academic skills. Weaknesses show up in standardized testing in quantitative reasoning tasks, and in developmental testing in poor development of conservation of number. Standardized psychological testing has also shown a relative strength in simultaneous processing of information compared to a relative weakness in sequential processing.

PHYSICAL APPEARANCE

There is a characteristic physical appearance associated with adults with Fragile X, and minor physical signs may be present during childhood. However, many children with Fragile X are difficult to distinguish from other children by their appearance. The most common physical signs before puberty are low muscle tone, hyperextensible joints, or large ears. The long face and jaw that are described in textbooks generally develop after puberty. This is also true of the large testicular size.

BEHAVIOR AND EMOTIONS

Children with Fragile X may suffer from both unusual behavior and poorly regulated emotions. Unusual behavior is often the earliest sign of the disorder. Many children have difficulty with social interaction, with a tendency to be shy and avoid eye contact. Unusual movement patterns such as hand flapping may also occur, especially in younger children. There may also be a pattern of sensory disturbance, such as a

fascination with spinning objects. These symptoms overlap with those of Autistic Spectrum Disorders, and as many as one third of children with Fragile X syndrome will also be diagnosed with Autistic Disorder or Pervasive Developmental Disorder, Not Otherwise Specified (PDD-NOS).

Anxiety is also a common problem in children and adults, and will aggravate all of the social, emotional and behavior symptoms of Fragile X.

GENETICS

Fragile X is an X linked chromosome disorder, like hemophilia and muscular dystrophy. This means that it is carried by females who generally do not show symptoms and then expressed in half of their sons.

Fragile X syndrome is caused by a mutation of a single gene on the X chromosome. This gene is called FMR1 and makes a protein called FMRP. The abnormal gene state may exist as a premutation, or a full mutation. The premutation is generally asymptomatic.

The severity of symptoms is related to two factors, the size of the mutation and whether or not the mutation is methylated. Individuals interested in more detailed information on the mutation and genetic research can find updated information at the websites for the National Fragile X Foundation or FRAXA.

TREATMENT

At this time (2009), there is no specific treatment for Fragile X syndrome, although research is active and medication trials are either pending or in progress. Medications are used to treat specific symptoms, such as hyperactivity, inattention, aggressive behavior, sleep disturbances, and unstable mood. The medications used to treat these symptoms are the same as those used to treat children with ADHD, Autism, and mental health problems. Children with Fragile X may also be at risk for seizures, and may need to be on seizure medication.

Since recommendations for the medical treatment of Fragile X may change, it is best to check with your physician. Up to date information on treatment, research and clinical trials can be found at the following websites:

FRAXA Research Foundation - <http://www.fraxa.org/>
National Fragile X Foundation - <http://www.fragilex.org/>

MORE INFORMATION

The National Institute of Child Health and Human Development has published a helpful pamphlet *Families and Fragile X*. It contains information that can be helpful to anyone interacting with a child with Fragile X syndrome. It is available online at: <http://www.nichd.nih.gov/publications/pubs/fragileX/index.cfm>.

EDUCATION

A child's educational program should always be based on his individual tested strengths and weaknesses. Some common educational supports are as follows:

- Children with Fragile X will need curricular accommodations and/or modifications for either intellectual deficiency or learning disability.
- Functional application of academic skills should be part of the curriculum.
- Most will benefit from visual supports.
- Many will need supports for a specific weakness in math skills.
- Accommodations will be needed for language processing problems.
- Social skills will need to be supported. Social skills training is frequently needed.
- The classroom setting should have high structure and predictable routines.
- Physical accommodations may be necessary for low muscle tone and poor coordination.
- Accommodations and interventions may be necessary for sensory issues.