

# Fragile X

Fragile X is recognised as the most common form of inherited learning disability, caused by a gene defect in the X chromosome. The disorder may be passed from one generation to the next. It can occur in both boys and girls, but is twice as common in boys, at an incidence of approximately 1 in 4,000. Learning disabilities vary considerably from mild to severe.

## Diagnosis

Many cases of Fragile X go undetected: people may be unaware of its presence in their families and themselves. This is because some people who carry the damaged gene might not be affected by it at all. They can however, pass it on to their children. DNA testing can detect Fragile X in children and adults. In the unborn child, diagnosis can be by amniocentesis or by CVS (chorionic villus sampling) as early as 10 weeks. This testing may not always distinguish affected from unaffected carrier females.

## The Genetics

The inheritance pattern in Fragile X is very complex, and it may be present for many generations before it causes any problems. Once a diagnosis has been made, parents should see a genetic counsellor to discuss the implications for their family. Very briefly, the FMR1 gene that causes Fragile X is on the X chromosome. A woman has two X chromosomes (XX); a man has one X and one Y chromosome (XY). It can be passed on by a woman or a man who has an affected X chromosome, to a child of either sex.

## The Characteristics

Boys are more severely affected than girls. This is possibly because girls have two X chromosomes, and the unaffected one may partly be able to compensate for the other.

### 1. Cognitive

The range of abilities is wide. A majority of boys with Fragile X will have some form of learning disability ranging from moderate to severe. Girls are usually less affected.

### 2. Physical

The physical features of Fragile X may be very subtle, and are seldom so marked as to give an unusual appearance. Some people with Fragile X may have heads that are larger than average, long faces, large jaws, protruding ears and high palates and dental overcrowding.

### 3. Behavioural

Behaviours can vary considerably. People with Fragile X can exhibit many autistic-like features, such as high anxiety in a crowded or noisy place - hand flapping, spinning - and a dislike of direct eye contact. They are comfortable in routines they know well, and will find changes stressful. They like the company of others and do not avoid social contact, although their autistic-like reactions to some situations may give rise to misunderstanding. The main problems appear to be impulsivity, inattentiveness and, especially in boys, hyperactivity. They may have motor coordination problems. Speech and language difficulties are common: speech may be repetitive, with strong use of learned stereotypic phrases. They may have trouble staying on topic, will throw in inappropriate comments, impulsive replies, and will talk at varying speed and volume.

## The Treatment

There is no cure for Fragile X at this present time. Treatment should be multi-disciplinary with input from the family's medical team, the child's school, speech therapists and occupational therapists. Medication may be suggested to improve concentration, and may be required if epilepsy is also present. Early intervention is highly recommended.

## Hints

Many children with Fragile X will be seen initially as relatively able, as they have high verbal abilities and will be alert to what is happening around them. They also have a good sense of humour and are good imitators. Their visual skills are good and they will learn better if visual clues and aids are used: their auditory skills are generally weaker. Their reading and spelling abilities may be higher than their comprehension skills. Mathematical skills, however, are usually poor.

Parents and teachers should be aware of the 'overload' effects of too much sound, movement, touch and even smell and taste. The resulting anxieties may produce non-stop chatter, total withdrawal, or autistic traits such as hand flapping or hand biting. Their anxieties and coordination problems may limit their use of playthings such as swings, bicycles, etc.

## In general

- use the child's interests, ability to mimic and his sense of humour as much as possible
- use lots of visual clues to augment auditory instruction
- break tasks down into small units
- prepare the child well for any changes to established routines
- keep all distractions to a minimum
- do not talk excessively; allow time for instructions to be assimilated, or answers given
- do not give too many instructions at once
- reinforce and encourage all attempts at speech
- bring the level of your speech to that of the child. Allow the child to work in small groups and, because of their ability to mimic, preferably with higher-functioning children
- maintain close home / school contact so that goals can be synchronised.

## Associated problems

See Behavioural paragraph above. Some people with Fragile X may also develop epilepsy, and there is a tendency to short or long sightedness, squints, and - in children - recurrent ear infections (glue ear).

## Other useful contacts

### Fragile X Society

Rood End House, 6 Stortford Road, Great Dunmow, Essex CM6 1DA.  
Provide support, information and friendship to families with a child with Fragile X and support to professionals.

**Web:** [www.fragilex.org.uk](http://www.fragilex.org.uk)

**Tel:** 01371 875100

**E-mail:** [info@fragilex.org.uk](mailto:info@fragilex.org.uk)

### Reading

See also OAASIS information sheet '**Books – where to find them**'

Available from [www.amazon.co.uk](http://www.amazon.co.uk) + p&p:

**Children with Fragile X Syndrome:** A Parent's Guide. Author J Dixon Weber. 2000, £6.95.

**Educating Children with Fragile X:** A Multi-Professional View  
Editor D Dew-Hughes. 2003, £28.99

**Fragile X Syndrome - A Guide for Teachers;** Author Suzanne Saunders. 1999, £20.99.

**Routledge Education** (member of the Taylor Francis Group)

**Web:** [www.routledgeeducation.com](http://www.routledgeeducation.com)

**Tel:** 020 7017 6000

**Fax:** 020 7017 6699

### Internet sites:

**The Fragile X Society** at [www.fragilex.org.uk](http://www.fragilex.org.uk)

**Contact a Family** at [www.cafamily.org.uk/Direct/f33.html](http://www.cafamily.org.uk/Direct/f33.html) for an information sheet about Fragile X and more information from the USA at <http://genetics.faseb.org/genetics/acmg/pol-16.htm>.

References: Use was made of Fragile X Society literature, Contact a Family information sheet and Internet papers attributed to The Children's Hospital, Denver, Colorado, USA.



[www.cambianeducation.com](http://www.cambianeducation.com)

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### OAASIS publications

- SEN Information Sheets
- First Guide to ...
- Cards explaining 9 learning disabilities
- Legal advice Information Sheets
- SEN 'How to' guides

All OAASIS publications can be downloaded from [www.oaasis.co.uk](http://www.oaasis.co.uk)