

## Fragile X syndrome

---

### What is Fragile X syndrome?

Fragile X is a genetic condition. The gene that causes Fragile X is found at the tip of the X chromosome and shows as a fragile site - hence the name. Both men and women can be carriers of Fragile X and the gene may pass through several generations of carriers before a child affected by the syndrome is seen. Diagnosis is established by a DNA or Chromosome test but a Fragile X test must be specified.

### Learning difficulties

Fragile X is the most common inherited cause of learning disability. It affects boys and girls and it is found in all populations and ethnic groups. Learning disabilities vary from subtle educational delays to severe mental impairment. Boys who are affected almost always have some learning difficulty which can range from moderate to severe. Up to half of the girls have learning problems which are occasionally severe.

### Behavioural features

The behavioural features include inattentiveness, distractibility and poor impulse control often associated with hyperactivity. Shyness and social withdrawal are striking features in girls with Fragile X syndrome who may experience difficulty in making friendships and feel alienated from peers.

Poor eye contact, difficulty in relating to other people, anxiety in social situations often leading to tantrums, insistence on familiar routines and hand flapping or hand biting may also occur.

### Physical features

Physical features may include a largish head and prominent ears but these may not be obvious in young children. Twenty percent of people with Fragile X have epilepsy.

### Speech and language delay

Speech and language delay is almost always present but its severity varies considerably. There may be an entire absence of speech through to milder and more

subtle communication difficulties. The characteristic speech pattern is fast and fluctuating. Generally the speech pattern has a jocular, running-on narrative style with frequent repetitions and swings of pitch described as "litany-like". There is a tendency to stick to one theme in conversation (topic perseveration) and children may repeat words or phrases that are spoken to them (echolalia) or repeat many times words or phrases that they themselves say (verbal perseveration). There may also be a tendency to leave sentences incomplete.

Girls with Fragile X may also have a high-pitched voice with repetitions.

Some children with Fragile X experience articulation difficulties which may be due to a large jaw and high arched palate. Hypotonia (low muscle tone) which occurs in some children with Fragile X may affect their muscles at the back of the mouth, the lips, nose and tongue which may in turn affect their ability to produce speech sounds.

Delay in the development of speech and language or language dysfunction as described above is often an indicator that a child with learning difficulties has Fragile X syndrome.

### References

- Davies, K (Ed.)** (1991), *The Fragile X syndrome*. Oxford: Oxford University Press
- Dixon, J** (2000), *Children with Fragile X syndrome*. Bethesda, MD: Woodbine House
- Dykens, E, Hodapp, R, & Leckman, J** (1994), *Behaviour and development in Fragile X syndrome*. London: Sage Publications
- Saunders, S** (2001), *Fragile X syndrome*. London; David Fulton
- Schopmeyer, B, & Lowe, F (Eds.)** (1992), *The Fragile X child*. London: Singular Publishing
- Turk, J, & Zwink, L** (1993), *Autism and the Fragile X syndrome*. *Communication*, 27(1), 10–11

***Please note: Afasic does not hold copies of any referenced material. However, it may be obtained via academic libraries.***

## **Other organisations which can help**

The Fragile X Society  
Rood End House  
6 Stortford Road  
Great Dunmow  
Essex  
CM6 1DA  
Tel: 01371 875100

*Written by Lynne Zwink, The Fragile X Society, and Katy Price, speech and language therapist*



Telephone  
Helplines  
Association  
quality and confidence for callers to helplines

© Afasic 2004

**Afasic**  
**1st Floor**  
**20 Bowling Green Lane**  
**London EC1R 0BD**  
**Phone 020 7490 9410**  
**Fax 020 7251 2834**  
**Email [info@afasic.org.uk](mailto:info@afasic.org.uk)**  
**[www.afasic.org.uk](http://www.afasic.org.uk)**  
**Helpline 0845 3 55 55 77**

**Registered charity no. 1045617**