

What is Fragile X Syndrome?

Fragile X syndrome is a genetic disorder and is the most common form of inherited learning disability. It is a sex-linked genetic abnormality in which the mother passes it on to her sons.

How many people have this condition?

It affects approximately 1 in every 1,000 to 2,000 male individuals, and the female carrier frequency may be substantially higher. Males with this syndrome typically have a moderate to severe form of learning difficulties. Females may also be affected but generally have a milder form.

Approximately 15% to 20% of those with Fragile X Syndrome exhibit autistic-type behaviours, such as poor eye contact, hand-flapping or odd gesture movements, hand-biting, and poor sensory skills.

Behaviour problems and speech/language delay are common features of Fragile X Syndrome.

Are their typical features of Fragile X?

Yes. These include a high arched palate, squints, large ears, long face, large testicles in males, low muscle tone, flat feet, and sometimes mild, heart valve abnormalities.

Although most individuals with Fragile X syndrome have a characteristic 'look' (long face and large ears), some individuals do not have this appearance.

How do you diagnose it?

This is usually by a paediatrician with support from a geneticist. Counselling will be offered and advice on management usually takes place in an educational setting.

Organisations:

Useful Links

UK

The Fragile X Society,

53 Winchelsea Lane

Hastings, East Sussex, TN35 4LG

THE DYSCOVERY CENTRE

Telephone: 44 – (0) 1424 813147

www.FragileX.org.uk

In US

The Fragile X Foundation
P.O. Box 30023
Denver, CO 80203