



Fragile X Syndrome (FXS)

Fragile X syndrome is the most common inherited cause of intellectual disability; it affects around 1 in 4000 boys and men, as well as 1 in 8000 girls and women. Boys with the condition are typically more severely affected than girls, some girls may not have any noticeable learning difficulties.

Fragile X can be detected by a simple blood test, and medical practitioners are increasingly recommending this if signs of ASD and learning difficulties are present in a child or young person. A significant number of people with Fragile X Syndrome have a dual diagnosis of autism.

Physical features of Fragile X Syndrome can be very subtle and include, to varying degrees:

- Low muscle tone resulting in hyper-extensible joints
- Flat feet
- Unusually soft skin
- Large and prominent ears
- A long, narrow face
- (The last two characteristics aren't always obvious before puberty).



Other key characteristics include:

- Learning difficulties across all areas
- High sensitivity to sensory stimuli (e.g., sounds, smells, bright lights)
- Social withdrawal (due to anxiety)
- Preference for 'sameness'
- Higher anxiety levels than usual for their peers, increases with changes
- Hand flapping and/or hand biting
- Hyperactivity
- Inattention

Fragile X Syndrome



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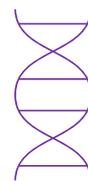


Positive features are that individuals with FXS often have good imitation skills, a good long-term memory and are often described as having likeable personalities and a good sense of humour.

Young people with Fragile X will usually respond better to an approach that is good-natured and humorous, with limited direct eye contact (finding this particularly difficult). Because they can be so sensitive and reactive to sensory input, a calm quiet environment will help to relieve anxiety and stress.

They also, as with individuals with other neurodevelopmental conditions, will benefit from early intervention in speech and language, occupational therapy, behavioural interventions and learning support.

Because any physical characteristics are subtle, especially in younger children, a diagnosis of Fragile X can be missed. A genetic test may be carried out if there is a reason to think a child may be affected (usually due to hereditary causes, but also for otherwise unexplained developmental delay). An unusual presentation of autistic characteristics with a happy/friendly personality may also indicate FXS.



For further information: the Fragile X Society has a number of downloadable booklets available on their website for parents and professionals:

www.fragilex.org.uk

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