

Fragile X Syndrome is the leading cause of inherited mental impairment. It is also the leading known cause of autism. Approximately 5% of people with autism turn out to have fragile X and 30% of children with fragile X meet the diagnostic criteria for autism.

Fragile X is a genetic disorder that affects about one in 4000 individuals. As many as one in 130 women and one in 800 men may be carriers of the faulty gene. For New Zealand's population of just over 4 million people, this means that around 1000 families are affected by fragile X and over 8000 New Zealanders are carriers of the gene.

This gene is located near the bottom of the X chromosome and gives it a "fragile" appearance. Because it is an X-linked disorder, female carriers have a 50% chance of passing the syndrome on to their children. Male carriers never pass on the syndrome to their sons, but will pass on the faulty gene to all their daughters.



Fragile X is characterized by a wide range of cognitive, behavioural and physical features.

The main cognitive features:

- × Mild learning difficulties to severe mental impairment
- × Developmental delay
- × Delayed and repetitive speech
- × Difficulties with mathematics and sequencing

The main behavioural features:

- × Attention deficit and hyperactivity
- × Autistic behaviours
- × Shyness, social anxiety
- × Difficulty adjusting to change
- × Need for routines
- × Sensory sensitivities (aversion to touch and noise)
- × Poor eye contact
- × Hand biting and hand flapping
- × Impulsivity, tantrums/meltdowns
- × Obsessive-compulsive disorder
- × Talkativeness
- × Good imitation skills

The main physical features:

- × Long face and large or prominent ears
- × Low muscle tone, double-jointed fingers, flat feet and heart murmurs
- × Frequent ear infections
- × Epilepsy (25% of people with fragile X)
- × Premature menopause in some female carriers
- × A Parkinson's-like tremor or ataxia syndrome (FXTAS) in some older adult carrier males

Note that few people with fragile X will display all these features

Testing for fragile X

Although there is currently no cure for fragile X syndrome, a recognised diagnosis can lead to appropriate management and intervention that helps affected individuals reach their full potential. The information obtained from the test can help identify other family members at risk of carrying the gene change and having children with fragile X. Fragile X is diagnosed from a simple blood test. This test is recommended for:

- × Anyone with an intellectual disability (borderline to severe) or autism of unknown cause
- × Anyone with learning difficulties of unknown cause and behavioural features of fragile X (including autistic features, anxiety and ADHD)
- × Anyone with a family history of fragile X who may have inherited the faulty gene
- × Intellectually disabled individuals with a previous fragile X cytogenetic test that was negative or inconclusive
- × Women who experience premature menopause (menopause before the age of 40 years)
- × Adult males (>50 years of age) who present with unexplained ataxia with or without essential tremor, parkinsonism and dementia

Fragile X testing is carried out by
Genetic Services, which has offices in Auckland (Tel: 0800 476 123), Wellington and Christchurch. (Tel: 0508 364436)



Intervention and Education

There are many types of intervention that can improve the lives of those affected by fragile X and their families. Everyone with fragile X can be helped and the proper education, therapy, and support can maximize their potential.

Speech and language, behaviour, cognitive development, sensory integration, gross motor development, daily living, and social skills are areas that often need to be addressed for someone with fragile X. All these areas require physical and behavioural intervention but medication is often an important component of the treatment.

Because of their talent for imitation, most fragile X children perform best in a mainstream classroom with adequate one-on-one support and curriculum adaptation. Visual timetables and cues are important to overcome difficulties with sequencing and following verbal instructions. For similar reasons, visually-based whole language approaches to reading are more effective than phonics or other auditory approaches. Fragile X children can be highly proficient with computers, which help to overcome difficulties with writing and provide a form of non-threatening learning.



Message to fragile X families

There is a global network of researchers, clinicians and parents working to understand, treat and eventually cure fragile X. If you have access to the internet, you are one click away from a vast information source, support system, and professional advice.

You or your child with fragile X have attributes that are rare in the wider population, but those of us who live with fragile X have learned to treasure these gifts: an engaging smile and brilliant sense of humour, an eagerness to please, a great imagination, an incredible eye for detail, a terrific memory for hard facts, and a warm, loving heart. The trick is to find the strategies that maximise these positive attributes and minimize others.

Newly diagnosed families are encouraged to contact their local support group. For contact information visit www.fragilex.org.nz

WEB RESOURCES

- × Fragile X Trust (NZ): www.fragilex.org.nz
- × Fragile X Association of Australia: www.fragilex.org.au
- × US National Fragile X Foundation: www.fragilex.org
- × US FRAXA Research Foundation: www.fraxa.org
- × UK Fragile X Society: www.fragilex.org.uk
- × NZ organisation for Rare Disorders (NZORD): www.nzord.org.nz

For more information please contact

Fragile X Trust (NZ)

Post: 196 Taita Drive, Lower Hutt 5011

Email: fragilex.info@nzord.org.nz

Phone toll-free: 0508 938 0552

Have you heard about Fragile X?



www.fragilex.org.nz