

What is Fragile X Syndrome?

Fragile X Syndrome is the leading cause of inherited intellectual disability. It can range from learning disabilities to severe cognitive or intellectual disabilities. Symptoms can include characteristic high anxiety and behavioural features, delays in speech and language development, ADHD and some physical features.

Fragile X 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. **All children exhibiting developmental delay or autistic behaviours, should be tested for fragile X.**

Fragile X is a genetic disorder that affects about one in 4000 males and one in 6000 females. As many as one in 130 women and one in 800 men may be carriers of the FMR1 gene mutation on the X chromosome. Carriers are at risk of developing fragile X- associated disorders like Fragile X-associated Tremor/Ataxia Syndrome (FXTAS) and Fragile X-associated primary ovarian insufficiency (FXPOI), early menopause.

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 gene mutation to all their daughters. For New Zealand's population of just under 5 million people, this means that around 12,000 New Zealanders are affected by fragile X.

Although there is currently no cure for fragile X syndrome, a recognised diagnosis can lead to appropriate support, intervention and treatment 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.

Fragile X can be passed on in a family by individuals who have no apparent signs of this genetic condition. In some families a number of family members appear to be affected, whereas in other families a newly diagnosed individual may be the first family member to exhibit symptoms.

About the Fragile X New Zealand Trust (FXNZ)

FXNZ is a charitable organisation that supports families affected by fragile X syndrome and fragile X-associated disorders and raises awareness about this genetic disorder. FXNZ offers a toll-free support line, a website and social media sites, publishes regular eNews and a newsletter, and runs annual family gatherings and workshops where information can be shared in a supportive and family atmosphere. We employ a part-time executive director to deliver our services and organise a team of volunteers; coordinate a nationwide family support network; run workshops and conferences; run education workshops in schools; and run a fragile X clinic where families can have their FX child assessed and given specialist support.