Strengths of People with Fragile X Syndrome

Parents and educators agree that people with Fragile X often have untapped potential. Cognitive strengths may include excellent memory skills and imitative abilities.

These skills enable them to learn a wide variety of tasks necessary for independence in daily life. As they are prone to imitate people it is important to consider their role models at home, school and work.

They often have a wonderful sense of humour and are caring, compassionate and helpful. Some have good verbal and reading skills. They learn in a particular way and therefore it is very important to use appropriate methods to teach them.

They often enjoy and do well in activities involving music, arts and sports. They may be quite creative and have excellent memories for songs, films, sporting events, etc. Appropriate interventions are advantageous to their development and well-being as well as helping them to maximise their potential.

You are not alone

The Fragile X Association of Australia

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Our organisation was formed to promote public awareness of Fragile X syndrome with special emphasis on educators and health professionals; provide support for families of people with Fragile X and enable them to meet and share their ideas, concerns and problems; and support scientific research on Fragile X syndrome. We provide specialised Fragile X clinics to our members. The clinics have been established to meet the specific needs of individuals with Fragile X syndrome.

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Fragile What?

Fragile X Syndrome is the most common inherited cause of intellectual disability and cognitive impairment worldwide.

MAY INCLUDE:

- **anxiety**
- **shyness**
- **ADHD**
- **autistic like behaviours**
- **poor eye contact**
- **delayed speech development**
- **tactile defensiveness**
- **repetitive speech**
- **aggression**
- **hyperactivity**
- **poor fine and gross motor skills**

www.fragilex.org.au

What is Fragile X Syndrome?

Behavioural Characteristics

Behaviours range from socially engaging and friendly to extreme shyness, poor eye contact, aggression and anxiety. Individuals with Fragile X syndrome can be easily overwhelmed by sensory stimuli: *crowds, noises, textures, etc.* In addition, they may display an aversion to being touched or held.

Hyperactivity and impulsivity are common in males with Fragile X. Shy and timid behaviour with a tendency toward social withdrawal is common in females. Both experience attention difficulties. Poor eye contact, hand-flapping and hand biting are common autistic-like behaviours. People with Fragile X may display some or all of these characteristics.

Intelligence

Individuals with Fragile X display a wide range of capabilities. Most males with Fragile X will have some degree of intellectual disability ranging from low-normal intelligence to severe impairment. The majority are mildly to moderately intellectually disabled.

Whilst females may appear less affected, over 50% have borderline to severe intellectual disability. The remainder may suffer from a range of emotional, behavioural and learning difficulties.

- Fragile X syndrome is the most common inherited cause of intellectual disability, affecting 1 in 3600 males and 1 in 4000 - 6000 females
- It is estimated that as many as 1 in 280 males and 1 in 125 females are carriers.
- Male carriers may develop FXTAS and female carriers may experience early menopause (FXPOI)
- Fragile X syndrome appears in people of all ethnic, racial and socio-economic backgrounds.
- It is estimated that as many as 100,000 Australians have Fragile X syndrome or are at risk of developing a Fragile X associated disorder.

Speech and Language

Most males will exhibit speech and language delays. As language develops it tends to be dysfunctional; rapid and uneven speech and repetition of words and phrases are common. Speech and language problems tend to be less severe in affected females.

Physical Characteristics

Physical features may be quite subtle or not present and because of this many children and adults with Fragile X syndrome appear to look normal, which may hinder diagnosis.

The physical features associated with Fragile X syndrome include a long narrow face, prominent ears, high palate, and, in postpubescent males, large testicles.

Abnormalities of connective tissue and muscle tone may result in hyper-extensible joints (double-jointed); flat feet; soft velvety skin and mitral valve prolapse (heart murmur). Low muscle tone may cause spinal curvature, strabismus (cross eyed) and slack facial features, particularly in young children.

How is Fragile X Syndrome Inherited?

What Causes Fragile X Syndrome?

Fragile X syndrome is caused by a mutation on the X chromosome. Females have two X chromosomes and males have one X and one Y chromosome. A gene near the end of the X chromosome normally contains between 6 and 50 repetitions of the genetic code CGG.

For reasons which remain unclear, the regulation of the code breaks down in some people causing the number of CGG repeats to increase. An expansion of the sequence from 50 to 200 repeats is called a premutation and may cause few or no symptoms of Fragile X syndrome. People with the premutation are know as carriers.

An increase to over 200 CGG repeats (sometimes into the thousands) is called a full mutation and results in Fragile X syndrome. This full mutation stops production of a protein required for brain development and all brain functioning.

Who Should be Tested?

- All people with an intellectual disability or ASD (Autism Spectrum Disorder)
- People with significant learning problems, mild cognitive deficits, ADHD and anxiety.
- People with any of the physical or behavioural features of Fragile X syndrome, regardless of gender or family history.
- Any individual who has a family member with a diagnosis of Fragile X syndrome or a family history of intellectual disability or developmental delay.
- Women who experience early menopause and people with symptoms of Parkinson's type diseases.

A DNA blood test for Fragile X can be organised through your GP or paediatrician or any medical doctor and may be covered by Medicare. It can also be used for prenatal diagnosis, and foetal testing can be accomplished via either amniocentesis or earlier in pregnancy through chorionic villus sampling (CVS). Testing prior to pregnancy allows full choice of family planning options. Genetic counselling should be part of any testing procedure.

Because Fragile X is an X-linked disorder, a father who carries the premutation will pass it on to all of his daughters but none of his sons.

A carrier mother, because she has two X chromosomes, has a 50% chance with each pregnancy of passing the mutation to her children.

The Fragile X mutation often increases when passed from mother to child. This means that children born with Fragile X syndrome frequently appear in families with no previous history of Intellectual disability.

Mother	Father	
	Y	X fragile
X	Son	Daughter
	XY	X X fragile
	Son	Daughter
X	XY	X X fragile

Father	Mother	
	X	X fragile
X	Daughter	Daughter
	XX	X X fragile
	Son	Son
Y	XY	Y X fragile

Majority Undiagnosed

Although Fragile X has been found in all parts of the world, it is estimated that the majority of individuals with Fragile X have not yet been diagnosed.

Treatment

Currently there is no cure for Fragile X syndrome, but there are effective treatments that can improve life quality. A multi -disciplinary therapeutic approach incorporating educational, medical, and behavioural management techniques is most beneficial. Pyschological, occupational, physical and speech therapies can be incorporated as appropriate.

Many people with Fragile X syndrome have sensory and perceptual processing problems which distort the way they receive information and their ability to use it to learn and behave appropriately.

Occupational therapists using sensory integration techniques in collaboration with the child's teacher and other educational team members have markedly improved these children's abilities to manage their behaviour, learn new information and increase their potential for a productive life. These strategies are especially effective when incorporated into an early intervention programme.

A doctor who is well-informed about the symptoms and implications of a Fragile X diagnosis is a critical member of any treatment team. He/she may recommend some medications.

Medical Treatment

Medical treatment focuses on ADHD (hyperactivity) symptoms, tantrums, anxiety, mood instability and obsessive-compulsive behaviours. Stimulants may be prescribed alone or in combination with other medications. Serotonin agents target anxiety and obsessive-compulsive behaviours and often improve social behaviours and language. Atypical antipsychotic medications may help to stabilise mood or decrease aggression.

Autism and Fragile X

Fragile X syndrome is the most common cause of inherited intellectual disability and the most common known genetic cause of autism.

Around 30% of children with Fragile X also have some degree of autism and conversely 2% to 6% of children diagnosed with autism have Fragile X syndrome. Therefore a child diagnosed with autism, or Autism Spectrum Disorder (ASD), should be tested for Fragile X syndrome. Also a child diagnosed with Fragile X syndrome should be evaluated for autism or ASD.

Premutation Disorders

People with the premutation (carriers) may have Fragile X associated disorders including intellectual disability, behavioural disorders, developmental delays, social and anxiety disorders that are often less obvious than for Fragile X syndrome. In addition male carriers can develop Fragile X associated Tremor/Ataxia Syndrome (FXTAS) and female carriers can experience early menopause or primary ovarian insufficiency (FXPOI).

FXTAS

A progressive, neurological condition affects some predominantly male carriers of Fragile X (up to 40%) over the age of 50. It is characterised by tremors, balance problems and dementia that become worse with age. The carriers are generally healthy until they reach their middle years, and have normal to above average intelligence. Initial signs of the disorder may include difficulty in writing, using utensils, pouring and walking. The symptoms progress over years or decades until many daily tasks become extremely difficult. There may be short-term memory loss, anxiety, decreased sensation to touch in the lower extremities and rigidity in movement. It is common to find these carriers misdiagnosed as having Parkinson's disease, senile dementia or Alzheimer's disease.

FXPOI

Approximately 20% - 25% of women who are Fragile X carriers will experience early menopause. This occurs on average 5 years earlier than non-carriers. In extreme cases, it can occur in the early 20s. Due to the decrease in hormone levels that accompanies this condition, women with FXPOI are at risk of osteoporosis at an earlier age than noncarriers.