

# Fragile X ..

Adapted from the Fragile X Association of Australia Website [www.fragilex.org.au](http://www.fragilex.org.au). Accessed 11th January 2012

Fragile X Syndrome is the most common cause of inherited intellectual disability and the most common known genetic cause of autism. It is a condition which can have impact on individuals and families in various ways and degrees of severity. Fragile X is found in all races and at all socio-economic levels. Latest statistics indicate 1 in 3600 males and 1 in 4000- 6000 females are affected and that approximately 1 in 260 females are carriers. Every week in Australia one child is born who is fully affected and twelve children are born who are carriers.

The name of the syndrome comes from its location on the X chromosome. Under particular laboratory conditions, the bottom of the long arm of this chromosome can appear broken or fragile. It has been estimated that, worldwide, 50% of people affected by fragile X have not been correctly diagnosed. Diagnosis of Fragile X is achieved through DNA tests. There is currently no cure for Fragile X. However, special therapies, methods of teaching and medication all provide real benefit to people with the syndrome and help them perform the best they can.

## Behaviour

It is important to understand the behavioural and emotional effects of the syndrome, as they can be as significant as developmental delays. Because people with fragile X syndrome look much the same as others, their behaviour problems can be misinterpreted.

Almost all people with the syndrome have trouble self-regulating their behaviour. This shows up in problems such as:

- Attention deficit disorders and hyperactivity – which can impair learning
- Repetitive behaviours
- Late toilet training
- Disturbed sleep
- The inability to control emotions
- Lack of control of emotional behaviour can result in tantrums or other emotional outbursts.

Both males and females:

- Find concentration difficult.

- They may be quite severely hyperactive and impulsive.
- They may have fixed interests in objects and
- They can strongly dislike any change in routine.

Some behaviours, most obvious with males, is similar to autism. For instance,

- Children can refuse to be touched or held.
- There can be difficulty with making eye contact.
- Hand flapping is often found, which becomes more vigorous when the person is anxious or excited.
- Some people bite their hands or chew their clothing. This can be a way to calm themselves.

Females with fragile X:

- May be shy and moody
- Lack social skills
- May avoid social contact
- They can suffer from depression and severe anxiety.
- Refusing to talk in certain situations (selective mutism) can be exhibited.

Despite the behavioural challenges outlined, people with fragile X often enjoy being social, have pleasant personalities and a good sense of humour.

## Development

Intellectual disability is the most significant characteristic of those with Fragile X. Most males and approximately two thirds of females exhibit some intellectual disability. This is usually accompanied by poor fine and gross motor skills. The range of learning problems is wide. Some are affected by minor developmental delays. At the other end of the scale there can be severe intellectual disability. Most affected males fall somewhere in the middle. Males typically appear more severely affected than females. However, parents and educators are often surprised at their achievements.

Females appear less affected. However, they may experience difficulty with math and tend to suffer from a range of anxiety disorders including social anxiety.

## Weaknesses

- Short-term memory
- Auditory-only processing
- Abstract concepts
- Sequencing, praxis and

planning

- Fine and gross motor
- Perceptual, visual motor
- Social, language, semantic-pragmatic
- Attention and initiation

## Strengths

- Learn visually e.g. pictures, computers
- Whole word, number and pattern recognition, 'gestalt' learning
- Long term and incidental memory
- Concrete, relevant tasks
- Strong imitation skills, drama
- Good functional life skills
- Friendly, good sense of humour

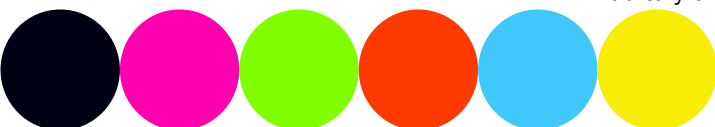
## Appearance

There are some of the physical features common to many, but not all, people with Fragile X syndrome. Even if present, the degree of visible effect varies from person to person. The features are more common in males than in females and are seen in approximately 50% of individuals

- The ears may be prominent.
- There may be a high forehead and/or long jaw, making the face look long, although this is often not apparent until puberty.
- Eye & vision impairments may cause a squint or turn in the eye which may only be apparent when the eye is stressed, for example in strong sunlight.
- Eyelids can tend to puffiness
- The chest may have a "hollow" look
- There may be a single palmar crease
- Flat feet (pes planus) may be evident
- A high arched mouth palate may be present
- Low muscle tone and hyper-extensible joints can cause awkwardness of movement and a "double jointed" look
- Large testicles (may be evident after puberty)
- The skin can be especially smooth and soft.
- Growth patterns indicate that children with Fragile X, especially males, are of average height or tall while young, but tend to be average once they are adults. These features are more common in males than in females and are seen in approximately 50% of individuals.

## Genetics

A change in a gene is called a mutation. Some mutations have no effect, but others, such as in the FMR-1 gene, cause changes in the structure or function of our bodies.





In front of the FMR-1 gene is the DNA sequence 'CGG'. This sequence is normally repeated between 6 and 50 times. In some people, this sequence is repeated between 50 to 200 times and is called a premutation, which generally causes few or no symptoms of fragile X syndrome.

In a woman with the premutation, the size can expand to over 200 repeats when that X chromosome is passed on in her egg. This is called a full mutation. In the full mutation, the gene for FMR-1 switches off and no protein is produced. Fragile X syndrome is the result. The expansion does not occur from males to their children. Males with the fragile X gene will pass that X chromosome without a change to all of their daughters.

### Medical Issues

People affected by Fragile X syndrome have a normal life expectancy. They do not suffer from major medical problems because of the syndrome and thus are generally healthy. However, there are some medical factors which may be more common in those with fragile X syndrome.

#### Common Medical Issues:

- Problems with connective tissue can lead to flat feet and low muscle tone.
- Eyelids that tend to puffiness and chests with a 'hollow' look are features which probably also relate to this problem.
- The joints are often extremely flexible and may be subject to dislocation.
- Hernias are sometimes present.
- Children, especially boys, are particularly prone to ear infections (otitis media). It is very important that every occurrence of ear infection is treated quickly, as problems in speech and language can be increased if there is even a slight temporary hearing loss.
- Eye problems can include squint (strabismus), long sightedness and visual perceptual problems.
- About one in four people with fragile X have epilepsy which can be generalised

or focal (grand mal, petit mal or absences, or complex partial seizures). Seizures usually begin in childhood or adolescence and are not frequent, often being outgrown before adulthood.

- A few people have heart valve problems (mitral valve prolapse), which may show up in late adolescence to early adulthood. Hence annual medical review is recommended.
- Many boys develop enlarged testicles (macroorchidism) after puberty, but this does not seem to pose any medical problems.
- Women with the premutation can have premature menopause. In extreme cases this can occur in the early twenties.
- FXTAS: Some male carriers, very often grandfathers of children with Fragile X Syndrome, may suffer from FXTAS (Fragile X-associated tremor/ataxia syndrome). This is a neurological disorder which can cause progressively worsening tremors as well as balance and co-ordination problems, cognitive and psychological changes.

### Sensory Issues

People with fragile X have difficulty in sorting out and screening all the messages that come through their senses. They are extremely hypersensitive to their environment. The problems they have in sorting sensory information are called 'sensory integration deficits'. These characteristics can increase learning and behavioural problems, as all sensations are received in an extreme form. They may have a negative response to situations that seem normal to others. Such situations include reactions to unfamiliar or strong smells, refusing to wear tight clothing, or clothing made of a rough fabric and being extra sensitive to bright or flashing lights. They are often disturbed in areas of high ambient noise, such as shopping centres and other crowded locations.

People with fragile X may not be able to communicate what is upsetting them. It is important for carers to know about this sensory integration problem. This response to sensory overstimulation has been called 'sensory defensiveness'. Individual programs, usually involving commonsense ways of organising and reducing environmental stimulation, can be planned to lessen the reaction. For instance, noise which is sensed as too loud, lighting which seems too bright and hypersensitivity to certain types of clothing can all be changed.

Occupational therapy with a sensory integration focus can lessen a person's sensory defensiveness.

### Speech

Most males exhibit delayed speech as children and poor command of spoken language. Some are inhibited when they do begin to speak.

#### Common speech characteristics:

- The rhythm of speech may be uneven or be fixed in a chant-like pattern.
- The automatic use of known phrases is common.
- Speech can be very fast and others may find it difficult to understand.
- Males with fragile X often continually repeat words and phrases (perseveration).
- Echoing the words of others (echolalia) is another problem.
- Males can speak constantly and inappropriately about a single topic.
- There are often problems in the use of expressive language, that is, in conveying thoughts to others.

On the positive side, vocabulary can be within the normal range. Some children verbally imitate others extremely well; this can become a helpful learning tool. Speech and language pathologists can assist in encouraging people to communi-

cate, and in improving the speech of older children and adults. For non-verbal individuals, augmentative communication systems, such as visual aids and signing, may be introduced. As with all helping techniques, the earlier the therapy begins the better.

## Education

Education is available for people with Fragile X at all stages of their development, from pre-school to the end of high school. Post-secondary education is an option for some. Children with developmental delay can be helped with early intervention even before a diagnosis is made. The earlier that this intervention can be included into the child's learning pattern, the better the results later on.

### Teaching Recommendations:

- Establishing structured routines
- Making careful preparation for changes
- The use of calming techniques and the elimination of distractions.
- Being able to see 'the whole picture' of what is going to happen, for example knowing in advance the full schedule for the day's schooling.
- Placing them with good role models in the classroom, wherever possible, as they are excellent imitators.

Pre-school and school teachers should be encouraged to work with the particular strengths of those with Fragile X.

### Strengths:

- They generally have very good long-term memories.
- They tend to learn visually. Use of pictures

can be very helpful, and illustrated social stories can assist with behaviour.

- A particular strength of many students with fragile X is their computer skills.

## Therapy

### Sensory Issues

Sensory problems are common particularly in boys affected by Fragile X. An excess of stimuli coming in through the senses can cause stress, anxiety and problems with concentration and behaviour. This sensory defensiveness can, in turn, result in a lowering in performance in other areas of development and learning. There are many strategies that can be used to alleviate such sensory challenges. Occupational therapists with sensory integration knowledge can work with the child and advise parents and carers of methods they can use themselves on an ongoing basis.

Sensory integration involves the ways we process and make sense of information coming in through each of our 5 senses. People with Fragile X, especially males, may be hypersensitive to sounds and noises, smells and tastes may be too strong for them, touch too intense and visual stimuli too much for them to process efficiently. In some cases the opposite may be true: they are hyposensitive and not getting enough stimuli.

A number of calming techniques can be used to reduce anxiety, to improve concentration and learning, to prevent inappropriate behaviour and enable the child to get used to his surroundings. Methods include massage, skin brushing, deep pressure, wearing weighted vests, the use

of equipment such as swings, mini trampolines and therapy balls and specially made objects to chew on.

Reducing strong stimuli that causes distress or preparing children in advance are other techniques that can be used. For example: removing fluorescent flickering lights, keeping noise levels down, using earplugs, giving the child a break or time out can reduce their stress. Explaining in advance in a way they understand can help prepare them; the use of homemade photo books is often recommended. Physical activity can improve sensory integration skills by giving a child practice. ●

You can learn more about Fragile X on the Fragile X Association of Australia Website. Visit [www.fragilex.org.au](http://www.fragilex.org.au)

You will find information about

- Clinics, Testing, Treatment and Counseling around Australia
- Resources
- Support for families
- Links to more information about Fragile X and related issues
- Membership
- Fundraising
- Forums



## resources...

- **The Nest** is a national plan for child and youth wellbeing by the Australian Research Alliance for Children and Youth. The Nest will involve an evidence review, national public consultation and a national action plan. You can find out more [www.thenestproject.org.au](http://www.thenestproject.org.au).
- **Book in Hand** is an Australian bookseller that focuses on books and resources for the ASD community. You can sign up to their e-list at [www.bookinhand.com.au](http://www.bookinhand.com.au).
- A new web resource for indigenous social and emotional wellbeing workers is now available on the **Australian Indigenous Health/InfoNet** website. The web resource provides free online access to Indigenous publications, resources and mental health screening and assessment tools. Visit [www.healthinfor.net.ecu.edu.au](http://www.healthinfor.net.ecu.edu.au) for more resources.
- The **Intellectual Disability Rights Service** has a great tool on their website which is a step by step guide to making a section 32 Application for a person with intellectual disability. Go to [www.idrs.org.au](http://www.idrs.org.au) and download the guide.
- **ABC Ramp Up** is a website on disability media. This site is where you will find ABC stories, interviews and videos on the subject on disability. You can visit the website at [www.abc.net.au/rampup](http://www.abc.net.au/rampup) or keep connected by RSS feed, following on Facebook or twitter @ABCRampUp.
- Visit [www.child-encyclopaedia.com/en-ca/home.html](http://www.child-encyclopaedia.com/en-ca/home.html) for an online child encyclopaedia of early childhood development. The encyclopaedia is free and has over 47 topics related to the psychosocial development of the child.
- **Network Autism** is a place where professionals can come together, share good practice and achieve better outcomes. Visit [www.autism.org.uk/networkautism](http://www.autism.org.uk/networkautism)
- The School Counselling Workforce in NSW Government schools and the literature review: Meeting the psychological and emotional wellbeing of children and young people: models of effective practice are available on the webpage <http://www.det.nsw.edu.au/about-us/statistics-and-research/public-reviews-and-enquiries/school-counselling-services-review>