Fragile X syndrome
Summary

- Fragile X syndrome is the most common inherited cause of intellectual disability.
- About 1 in 3,600 boys and 1 in 4,000–6,000 girls have Fragile X syndrome.
- The effects of Fragile X syndrome vary widely but most people experience lifelong difficulties.
- Both men and women can be carriers of the Fragile X gene. Female carriers of the Fragile X gene are at risk of having one or more children with Fragile X syndrome.

Fragile X syndrome is a genetic disorder caused by a change to one of the genes on the X chromosome. It is the most common inherited cause of intellectual disability. Fragile X syndrome is also linked to features of autism spectrum disorder.

Fragile X syndrome affects around 1 in 3,600 boys and between 1 in 4,000 – 6,000 girls. Fragile X syndrome causes a wide range of physical, developmental, behavioural and emotional difficulties although the level of severity between people with the condition varies enormously.

Effects of Fragile X syndrome

Fragile X syndrome can cause a range of physical, developmental, behavioural and emotional difficulties in people. Although there is no typical presentation, some boys with Fragile X syndrome have a long narrow face, prominent forehead and large testicles.

The most significant effects of Fragile X syndrome are:

- global developmental delay, including speech, language and communication difficulties
- intellectual disability and learning problems
- anxiety
- autistic-like behaviours such as hand flapping, repeating words and sentences, and difficulty with social interactions
- attention deficit hyperactivity disorder (ADHD)
- poor eye contact
- difficulty processing sensory information.

The level of severity of these effects varies considerably and not every person with Fragile X syndrome will have every one of these characteristics.

Cause of Fragile X syndrome

Fragile X syndrome is caused by a change to a gene on the X-chromosome called the FMR1 gene. This gene produces a protein that helps the brain to function normally. If this gene is changed or altered in any way, it cannot produce its normal protein, which can result in Fragile X syndrome.

Fragile X syndrome is inherited in a way that is known as 'X-linked', as the changed gene is on the X chromosome. This means that men with Fragile X syndrome are often more severely affected than women. This is because men only have one X chromosome, whereas women have two X chromosomes, only one of which is changed.

Pre-mutation carriers of the Fragile X gene

Although Fragile X syndrome is not that common, affecting around 1 in 3,600 boys and between 1 in 4,000 – 6,000 girls, the number of men and women who are carriers of the Fragile X gene is significantly higher.

It is estimated that 1 in 150 women and 1 in 800 men are carriers of the gene. These people are known as 'Fragile X pre-mutation carriers'. Pre-mutation carriers may not have any symptoms of Fragile X syndrome but they are at risk of passing on the changed FMR1 gene and having a child or grandchild with Fragile X syndrome.

People who are Fragile X pre-mutation carriers may also be at risk of developing health problems later in life. Around 20 per cent of female Fragile X pre-mutation carriers experience reduced fertility or early menopause. This is called Fragile X-associated Primary Ovarian Insufficiency, or FXPOI.

Older male and female Fragile X pre-mutation carriers are also at risk of developing a neurological condition called FXTAS, or Fragile X-associated Tremor Ataxia syndrome. Problems with balance and gait, tremors and gradual intellectual decline are common in FXTAS. These conditions are called Fragile X-associated Disorders.

Testing and diagnosis of Fragile X syndrome

Fragile X syndrome (and Fragile X-associated disorders) can only be diagnosed by DNA testing – usually by a blood test but sometimes via cheek swab or mouthwash. If possible, testing should be done at a recognised genetics service [http://www.genetics.edu.au/Genetics-Services/genetic-testing-services].

If there is a diagnosis of Fragile X syndrome, it is important that immediate and extended family members are also tested to identify others who may have the changed
FMR1 gene. This is called cascade testing.

DNA testing is recommended for:

- people with a family history of Fragile X syndrome or intellectual disability
- people with intellectual disability, developmental delay or learning disability together with features of Fragile X syndrome such as anxiety, ADHD or characteristics of autism spectrum disorder
- men or women over 50 with balance or gait problems, tremor or dementia
- any woman with problems with fertility or early menopause (under 40)
- women with family history of primary ovarian insufficiency (loss of function of the ovaries before age 40) for testing before or during pregnancy.

Unfortunately diagnosis of Fragile X syndrome is often delayed and it is not uncommon for families to have had a second child born with Fragile X syndrome before the first child has been diagnosed. A prompt diagnosis of Fragile X syndrome is important so that families can get access to appropriate early intervention services to help their child reach their full potential.

**Genetic counselling services and Fragile X syndrome**

The facts about Fragile X syndrome are complicated and the ramifications for families can be serious. It is recommended that parents and family members ask their doctor to refer them to a genetics service, both for testing and follow-up counselling. The genetics counsellor can also provide information about the range of reproductive options that are now available.

Clinics in Victoria are located in metropolitan Melbourne and major regional centres. Contact Victorian Clinical Genetics Services for more information about Fragile X syndrome or to organise an appointment.

**Where to get help**

- Your doctor
- Paediatrician
- Fragile X Association of Australia Tel: 1300 394 636 (freecall)
- Victorian Clinical Genetics Services Tel: (03) 8341 6212 or 1300 11 8247
- Fragile X Alliance Clinic Tel: (03) 9528 1910
- Genetic Support Network Victoria Tel: (03) 8341 6315
- Carers Victoria Tel: 1800 242 636 (freecall)

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More information

Genes and genetics

The following content is displayed as Tabs. Once you have activated a link navigate to the end of the list to view its associated content. The activated link is defined as Active Tab

- A-Z of genetic conditions
- Genes and genetics explained
- Genetic testing

A-Z of genetic conditions

- Ambiguous genitalia
  The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals...
- Angelman syndrome
  The characteristic features of Angelman syndrome are not always obvious at birth, but develop during childhood...
- Ankylosing spondylitis
  Ankylosing spondylitis (AS) is a type of inflammatory arthritis that targets the joints of the spine...
- Barrett's oesophagus
  Symptoms of Barrett's oesophagus are similar to regular heartburn, which means many people don't seek treatment until their condition is quite advanced...
- Bipolar disorder
  Bipolar disorder is a type of psychosis, which means the person's perception of reality is altered. It is characterised by extreme mood swings...
- Central nervous system birth defects
  Folic acid taken before conception, and during at least the first four weeks of pregnancy, can prevent around seven out of 10 cases of neural tube defects...
- Charcot-Marie-Tooth disease (CMT)
  Charcot-Marie-Tooth disease is the most common inherited disorder affecting the peripheral nervous system...
• Cleft palate and cleft lip
  Most cleft palates and cleft lips can be repaired so that appearance and speech develop normally...

• Congenital adrenal hyperplasia (CAH)
  CAH is a rare genetic disorder, but it is well understood and treatment is readily available...

• Creutzfeldt-Jakob disease (CJD)
  Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, dementia and walking difficulties...

• Cri du chat syndrome
  Most children born with cri du chat syndrome have moderate intellectual disability, with varying degrees of speech delay and some health problems...

• Cystic fibrosis (CF)
  When a person has cystic fibrosis, their mucus glands secrete very thick sticky mucus that clogs the tiny air passages in the lungs and traps bacteria...

• Digestive tract birth defects
  Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of defects of the digestive tract...

• Down syndrome
  With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community...

• Dwarfism
  Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth...

• Eczema (atopic dermatitis)
  Eczema can vary in severity, and symptoms may flare up or subside from day to day...

• Essential tremor
  Essential tremor causes involuntary shaking or trembling of particular parts of the body, usually the head and hands, but it is not Parkinson's disease...

• Fragile X syndrome
  The facts about fragile X syndrome are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic...

• Friedreich's ataxia
  To the casual observer, a person with Friedreich ataxia may seem to be drunk...

• Genetic factors and cholesterol
  Familial hypercholesterolemia is an inherited condition characterised by higher than normal levels of blood cholesterol...

• Haemochromatosis
  Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses...

• Haemophilia
  All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed...

• Hair
  Human hair grows one centimetre every month...

• Hearing problems in children
  The earlier that hearing loss is identified in children, the better for the child's language, learning and overall development...

• Huntington's disease
  The symptoms of Huntington's disease usually, but not always, first appear when the person is approaching middle age...

• Kabuki syndrome
  Kabuki syndrome affects males and females equally and there is no cure...

• Kennedy's disease
  Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs...

• Kidneys - medullary cystic kidney disease

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Medullary cystic kidney disease causes the growth of abnormal cysts in the kidneys.

- **Kidneys - polycystic kidney disease (PKD)**
  Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds.

- **Klinefelter syndrome**
  Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don't occur.

- **Leukodystrophy**
  Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain, which causes loss of normal brain functions.

- **Long QT syndrome**
  You should be investigated for long QT syndrome if you faint for no apparent reason, during or after exercise or emotional excitement.

- **Marfan syndrome**
  Some people may not realise they have Marfan syndrome, because their features are either mild or not obvious.

- **McCune-Albright syndrome**
  The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict.

- **Muscular dystrophy**
  People affected by muscular dystrophy have different degrees of independence, mobility and carer needs.

- **Neurofibromatosis**
  Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

- **Noonan syndrome**
  Noonan syndrome is a genetic condition that usually includes heart abnormalities and characteristic facial features.

- **Osteoporosis in children**
  Osteoporosis in children is rare and usually caused by an underlying medical condition.

- **Phenylketonuria (PKU)**
  PKU is an inherited disorder that prevents the normal breakdown of a protein found in some foods.

- **Prader-Willi syndrome**
  A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

- **Premature and early menopause**
  The symptoms of premature or early menopause are the same as for menopause at any age.

- **Rett syndrome**
  People with Rett syndrome have a keen desire to communicate.

- **Spinal muscular atrophy (SMA)**
  A child with spinal muscular atrophy type 1 rarely lives beyond three years of age.

- **Tay-Sachs disease**
  Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians.

- **Thalassaemia**
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated.

- **Tourette syndrome**
  Milder forms of Tourette syndrome can be misdiagnosed, as it often occurs at the same time as attention deficit hyperactivity disorder (ADHD) and other disorders.

- **Treacher Collins syndrome**
  Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial defects and hearing loss.

- **Trisomy disorders**
  Children affected by trisomy usually have a range of birth defects, including delayed development and intellectual disabilities.
• Tuberous sclerosis
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity.

• Usher syndrome
  Services aim to help a person with Usher syndrome prepare for the dual loss of sight and hearing.

• Von Willebrand disease
  A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth.

• Williams syndrome
  Williams syndrome often goes undiagnosed, which means that some people with the disorder fail to get the support and treatment they need until later in life.

• Wilson disease
  In Wilson's disease, a build-up of copper damages organs including the liver, nervous system, brain, kidneys and eyes.

Genes and genetics explained

• Genes and genetics explained
  Children inherit physical characteristics such as eye colour from their parents through their genes.

• Gene therapy
  Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases.

• Genetic disorders
  Genetic disorder is caused by an altered or faulty gene or set of genes.

• Genetic services in Victoria
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare.

Genetic testing

• DNA profiling
  DNA profiling is a way of establishing identity and is used in a variety of ways.

• Egg freezing
  You can freeze your eggs for medical reasons or for reasons that are more to do with your life circumstances.

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• Genetic testing for inherited cancer
  A predisposition to certain cancers can be inherited via altered genes.

• Newborn bloodspot screening
  Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk of rare, but serious, medical conditions.

• Pregnancy tests – chorionic villus sampling
  Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities.

• Pregnancy tests - maternal serum screening
  Maternal serum screening can indicate increased risk of abnormalities in the unborn child, but is not a diagnosis.

Related Information

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**Related information on other websites**

- Centre for Developmental Disability Health (CDDHV)
- Fragile X Alliance Clinic
- Murdoch Childrens Research Institute
- NSW Council for Intellectual Disability
- The Fragile X Association of Australia
- Victorian Clinical Genetics Services

**Support Groups**

- The Fragile X Association of Australia

**Content Partner**

This page has been produced in consultation with and approved by: Victorian Clinical Genetics Services (VCGS)

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