Klinefelter syndrome
Klinefelter syndrome is a chromosome disorder that affects males. It is not an inherited disorder.

A male with Klinefelter syndrome has an additional X chromosome, which causes infertility, low testosterone and other characteristics such as development of breast tissue.

The chromosomes are present in every cell of the body and the extra X chromosome cannot be removed.

Treatment includes lifelong testosterone therapy and cosmetic surgery to remove enlarged breast tissue. Options for men with Klinefelter syndrome to have their own children are becoming more widely available.

Klinefelter syndrome is a chromosome disorder that affects males. Usually, a male has two chromosomes that determine his sex: an X inherited from his mother and a Y inherited from his father. A male with Klinefelter syndrome has an additional X chromosome.

The effects can vary greatly and may include:

- infertility – the condition may be diagnosed when fertility problems are being investigated
- reduced testicle size – in all males with Klinefelter syndrome, the testicles do not develop properly and are noticeably smaller from early adolescence
- incomplete puberty – not enough of the male hormone testosterone is produced and puberty may appear to be delayed. These boys may be given testosterone treatment so they develop usual male physical characteristics like facial hair and a deeper voice
- language and learning problems – in many males with Klinefelter syndrome, difficulties with speech, writing, or understanding and processing speech may be noticed.

Klinefelter syndrome occurs in around one in every 450 male babies, which makes it one of the most common variations of the chromosomes. However, only around a quarter of these males have a diagnosis of Klinefelter syndrome, also known as XXY syndrome. The additional X chromosome does not influence sexual orientation.

Signs of Klinefelter syndrome at birth

The condition isn’t usually diagnosed at birth, because the baby boy looks healthy and unaffected. However, certain physical characteristics sometimes associated with Klinefelter syndrome may be apparent, including:

- small penis
- undescended testicles
- hypospadias (the urethra is located on the underside of the penis instead of the tip).

Signs of Klinefelter syndrome at childhood

The appearance of signs of Klinefelter syndrome in childhood can vary greatly and range from mild to those that are more obvious. Many of the signs of Klinefelter syndrome also occur in children with other conditions, or children who do not have an underlying diagnosis, which makes detecting Klinefelter syndrome in childhood difficult.

Furthermore, many boys show no signs of Klinefelter syndrome during childhood. However, for those that do, early intervention provides the opportunity to overcome difficulties and for each boy to reach their individual potential.

Signs that may be apparent in childhood include:

- delays in starting to talk or walk
- learning difficulties, including trouble listening or concentrating in class
- poor motor development or coordination (sometimes leading to shyness or an avoidance of rough sports)
- tiredness and fatigue
- reduced muscle strength or tone (sometimes leading to other complications such as sleep apnoea or chronic constipation).

Signs of Klinefelter syndrome at puberty

Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don’t occur. Some of the signs and symptoms of the condition include:

- small testicles (hypogonadism) – males with Klinefelter syndrome always have small testicles from puberty

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• lack of facial, pubic and underarm hair
• poor muscle development
• breast tissue development (gynaecomastia)
• unexplained weight gain, especially on the stomach or trunk
• tiredness and fatigue
• small penis
• disproportionately long arms and legs compared to the length of the body
• single crease in the palm (simian crease)
• difficulties with sexual functioning
• speech and learning difficulties – in some cases this can lead to behavioural problems
• sadness, lowered mood or depression.

Boys may become particularly self-conscious in puberty when they notice that their body is not undergoing the same changes as their peers. Testosterone replacement therapy can help the body to undergo these changes.

Most males with Klinefelter syndrome are not diagnosed until they are well into adulthood, when diagnosis can come as a major shock. An earlier diagnosis provides opportunities for the child and his family to access treatments and interventions that can help overcome any difficulties associated with the condition.

**Cause of Klinefelter syndrome**

While the cause of Klinefelter syndrome is genetic, it is not inherited. Humans have 46 paired chromosomes, made up of two sex chromosomes that determine gender and 44 chromosomes that determine other factors. The mother always passes on an X chromosome. If the father provides an X chromosome, the child will be female, while a Y chromosome makes the child male.

A boy with Klinefelter syndrome has an additional X chromosome. This is thought to be caused by an error within the fertilised egg or the dividing cells as the baby develops.

The presence of the Y chromosome ensures male sexual characteristics but, because the testicles are underdeveloped, there may not be enough testosterone production. This is why the penis and testicles may be smaller than average, and why most men with Klinefelter syndrome do not produce sperm. Some researchers suspect that advanced parental age may be a risk factor.

**Complications of Klinefelter syndrome**

Compared with males in the general population, Klinefelter syndrome is associated with an increased risk of certain diseases and conditions, including:

• osteoporosis
• diabetes mellitus
• lung disease
• breast cancer
• testicular cancer
• anaemia
• infertility
• depression or other psychiatric conditions
• leukaemia
• non-Hodgkin lymphoma
• thyroid disease
• sleep apnoea
• high cholesterol
• tooth decay
• varicose veins.

It is currently thought that testosterone treatment from puberty, which helps with bone and muscle development among other things, may reduce the risk of many of the above conditions.

**Diagnosis of Klinefelter syndrome**

Klinefelter syndrome is diagnosed using a number of tests, including:

• physical examination – all males with Klinefelter syndrome have small testicles, which can be detected by genital examination
• chromosome analysis – will confirm the diagnosis
• blood tests – can check for hormone levels
• semen examination – checks fertility.

**Treatment of Klinefelter syndrome**

There is no cure for Klinefelter syndrome. Treatment options can improve some aspects of the condition and provide emotional support. Sample treatments include:

• hormone therapy
• reproductive technologies to aid fertility
• educational support (including: speech therapy, physical or occupational therapy) – if necessary
• cosmetic surgery
• counselling
• frequent screening tests – to ensure the early diagnosis of any associated complications.
Further information about Klinefelter syndrome can be found on the Andrology Australia website.

**Where to get help**

- Your doctor
- Endocrinologist
- Genetic Support Network Victoria Tel. (03) 8341 6315
- Andrology Australia Tel. 1300 303 878

**References**


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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.

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- **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 hypercholesterolaemia 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**
  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.

- **Tuberous sclerosis**
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity.

- **Turner's syndrome**
  Turner's syndrome is a random genetic disorder that affects females, causing short stature and infertility.

- **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**

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

• **Genetic disorders**
  A 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 Genetics Education NSW
Support Groups

- Genetic Support Network Victoria (GSNV)

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