HEALTHY MALE
ANDROLOGY AUSTRALIA

Fact Sheet

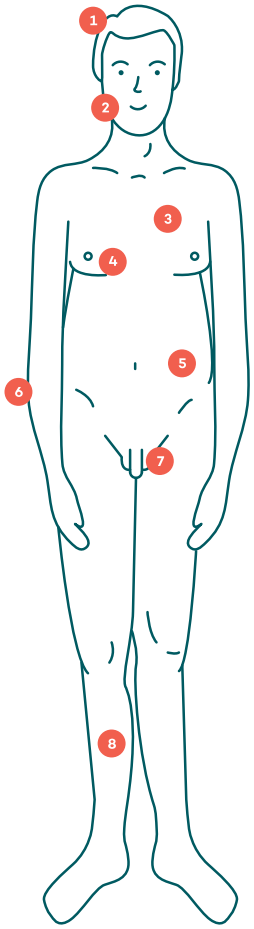
Klinefelter Syndrome

What is Klinefelter syndrome?

Klinefelter syndrome is a genetic (chromosomal) condition that only affects males. It is congenital, which means it is present from birth. Men with Klinefelter syndrome have an extra X chromosome. The normal male chromosome arrangement is 46XY, but for men with Klinefelter syndrome it is 47XXY.

How common is Klinefelter syndrome?

Klinefelter syndrome is the most common chromosomal disorder in men, affecting about 1 in 650 men. However, many men with Klinefelter syndrome are never diagnosed.



1. Taller than average height
2. Reduced facial hair
3. Reduced body hair
4. Breast development (gynecomastia)
5. Feminine fat distribution
6. Osteoperosis
7. Small testes (testicular atrophy)
8. Varicose veins

SYMPTOMS OF KLINFELTER SYNDROME

Childhood

- difficulties with speech and reading
- delayed motor development
- lower attention span
- poor muscle tone
- behavioural problems
- undescended testes at birth (uncommon)

Puberty Adulthood

- small testes
- gynecomastia (breast enlargement)
- taller than average height
- fat accumulation on abdomen and hips
- less facial and body hair/ decreased shaving frequency
- low libido (sex drive)
- poor erections
- fatigue
- infertility
- osteoporosis (thinning of the bones)
- varicose veins
- depression

What are chromosomes?

Chromosomes are found in each cell in the human body. They carry the genetic material that determines all human characteristics, including hair colour, eye colour, height and sex. Each cell in the human body has 23 pairs of chromosomes (a total of 46).

Of the 23 pairs of chromosomes, one pair is called the sex chromosomes because they determine a person's sex. The sex chromosomes in a female are called XX and in a male are called XY. One sex chromosome is inherited from the mother and one from the father. Mothers always pass on an X chromosome, but fathers can pass on an X or a Y chromosome to their children.

What causes Klinefelter syndrome?

The extra X chromosome may come from the egg or sperm, or be 'added' early in the development of the embryo. In either case it is not known why the extra X chromosome happens. The brothers of men with Klinefelter syndrome almost always have a normal 46XY chromosome pattern.

What are the main effects of Klinefelter syndrome?

Klinefelter syndrome is the most common cause of male hypogonadism, a condition where men are unable to produce sperm or enough of the male hormone, testosterone, for the body's needs.

Testosterone is the most important androgen (male sex hormone) in men and it is needed for normal reproductive and sexual function. Testosterone is important for the physical changes that happen during male puberty, such as development of the penis and testes, and for the features typical of adult men such as facial and body hair. Testosterone also acts on cells in the testes to make sperm.

Testosterone is also important for overall good health. It helps the growth of bones and muscles, and it affects mood, libido (sex drive) and certain aspects of mental ability.

The low levels of testosterone in men with Klinefelter syndrome affect the development of male characteristics. The extra X chromosome also affects the ability to produce sperm. Men with this condition are infertile as they almost always have no sperm in their ejaculate (azoospermia).

What are the symptoms of Klinefelter syndrome?

Symptoms of Klinefelter syndrome vary between individuals and include a range of physical features, such as tall stature, breast development (gynecomastia) and behavioural and learning difficulties. Small testes are present in almost all men with Klinefelter syndrome.

How is Klinefelter syndrome diagnosed?

Small testes (1–4 mL, about the size of a sultana grape) after puberty are an indication of Klinefelter syndrome in most cases.

A diagnosis of Klinefelter syndrome is confirmed using a blood test called a karyotype that checks the number and structure of chromosomes in cells.

A blood test is also carried out to look at levels of testosterone, luteinizing hormone (LH) and follicle stimulating hormone (FSH). LH is needed for the cells in the testes to make testosterone. Testosterone and FSH act together on the seminiferous tubules (sperm-producing tubes) in the testes to make sperm. In many men with Klinefelter syndrome, levels of LH are raised, but testosterone levels are borderline or below normal. FSH levels are markedly raised as a sign of damage to the seminiferous (sperm producing) tubules in the testes.

When is Klinefelter syndrome diagnosed?

With the increase in use of prenatal (before birth) testing (such as maternal blood testing in early pregnancy), Klinefelter syndrome may be diagnosed before birth. If not, a paediatrician may diagnose Klinefelter syndrome immediately after birth (postnatally). In other cases, Klinefelter syndrome is identified during childhood when learning or behavioural difficulties develop, or around the time of puberty when expected physical changes are delayed or do not happen. Because the symptoms are not always obvious, the diagnosis of Klinefelter syndrome might not be made until the man seeks medical help for infertility, a loss of sex drive or a bone fracture, or is not diagnosed at all.

Why is Klinefelter syndrome under-diagnosed?

It is suspected that as many as three quarters of the men with Klinefelter syndrome are not diagnosed and remain untreated for life. This could be because doctors do not routinely check testes size.

Some symptoms of Klinefelter syndrome during childhood and puberty, such as learning difficulties and behavioural problems, can be due to other conditions and so doctors may not think about Klinefelter syndrome.

A lack of knowledge about their own body is another reason that men with undiagnosed Klinefelter syndrome may not visit a doctor. These men may be unaware of how small their testes are and they may not think anything is wrong. Other men may be too shy or embarrassed to approach a doctor if they are concerned about the size of their testes.

How is Klinefelter syndrome treated?

Klinefelter syndrome cannot be cured, but men with the condition need lifelong testosterone therapy to maintain general wellbeing.

What are the main forms of testosterone therapy?

Testosterone therapy is available in Australia in the form of injections, gels, creams, patches and tablets, and works very well for men with confirmed androgen (testosterone) deficiency. The type of treatment prescribed can depend on patient convenience, familiarity and cost. Commercial testosterone products contain only the natural testosterone molecule that is chemically produced from plant materials.

Will I need to see a specialist for Klinefelter syndrome?

A general practitioner or endocrinologist can supervise testosterone therapy in men with Klinefelter syndrome.

When should testosterone therapy start for Klinefelter syndrome?

Testosterone therapy in males with Klinefelter syndrome should be started from puberty. Teenage boys with the condition should start off on a lower dose of testosterone than adult men, and build up to the full dose as puberty progresses. Management of Klinefelter syndrome may need involvement from the school because these boys may have learning difficulties and benefit from extra assistance in the classroom.

How is infertility treated in men with Klinefelter syndrome?

Infertility is a major issue for men with Klinefelter syndrome.

It is rare for men with Klinefelter syndrome to have any sperm in their ejaculate; however, in around four in 10 men, sperm can be found in testicular tissue. If sperm can be retrieved from testicular tissue, assisted reproductive technology such as intracytoplasmic sperm injection (ICSI) can be used to achieve pregnancy.

ICSI is a form of in vitro fertilisation (IVF) where a single sperm is placed directly into each egg by piercing the outer covering of the egg.

For many men wishing to have children with their partner, the best option is donor insemination. Donor insemination involves implanting donated sperm into a woman to achieve pregnancy.

Counselling is available for men coming to terms with the diagnosis of Klinefelter syndrome and issues such as infertility.

Visit **healthymale.org.au**
or speak to your doctor for more info.

HEALTHY MALE
ANDROLOGY AUSTRALIA

EXPERT REVIEWER

Prof Rob McLachlan AM
MBBS FRACP PhD
Healthy Male (Medical Director)

DATE REVIEWED: FEBRUARY 2018

healthymale.org.au

Australian Government
Department of Health