Klinefelter’s syndrome

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

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

What causes Klinefelter’s 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’s syndrome almost always have a normal 46XY chromosome pattern.

What are the main effects of Klinefelter’s syndrome?
Klinefelter’s 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.

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.
syndrome are not diagnosed and remain untreated for life. This could be because doctors do not routinely check testes size. Some symptoms of Klinefelter's 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's syndrome. A lack of knowledge about their own body is another reason that men with undiagnosed Klinefelter's 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’s syndrome treated?
Klinefelter's 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’s syndrome?
A general practitioner or endocrinologist can supervise testosterone therapy in men with Klinefelter's syndrome.

When should testosterone therapy start for Klinefelter’s syndrome?
Testosterone therapy in males with Klinefelter's 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’s 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’s syndrome?
Infertility is a major issue for men with Klinefelter's syndrome. It is rare for men with Klinefelter's syndrome to have any sperm in their ejaculate; however, in about 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's syndrome and issues such as infertility.

For more information visit www.andrologyaustralia.org, call 1300 303 878, or speak to your doctor.