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

How is Klinefelter’s syndrome diagnosed?
Small testes (1–4 mL, about the size of a sultana grape) after puberty are an indication of Klinefelter’s syndrome in most cases. A diagnosis of Klinefelter’s syndrome is confirmed using a blood test called a karyotype that checks the number and type of chromosomes.

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.
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’s 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’s syndrome diagnosed?

With the increase in use of prenatal (before birth) testing (such as maternal blood testing in early pregnancy), Klinefelter’s syndrome may be diagnosed before birth. If not, a paediatrician may diagnose Klinefelter’s syndrome immediately after birth (postnatally). In other cases, Klinefelter’s 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’s syndrome might not be made until the man seeks medical help for infertility, a loss of general wellbeing 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 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’s syndrome and issues such as infertility.

Males with Klinefelter’s syndrome need lifelong testosterone therapy to maintain general wellbeing