Klinefelter Syndrome Essay, Research Paper

Klinefelter syndrome, also called testicular dysgenesis, is the phenotype of the 47,XXY genotype and is characterized by male hypogonadism and infertility. Klinefelter Syndrome is probably the most common chromosomal variation found in humans. In random surveys, it is found to appear about once in every 500 to one in every 1,000 live born males. Since the largest percentage of these men would have never been diagnosed otherwise, it shows that in many cases affected individuals lead healthy, normal lives with no particular medical or social questions. Klinefelter Syndrome is diagnosed through a karyotype; a chromosome analysis usually done on a blood sample.

Klinefelter Syndrome is caused by a chromosome variation involving the sex chromosomes. The person with Klinefelter Syndrome is a male who, because of this chromosome variation, has a hormone imbalance. While Dr. Harry Klinefelter accurately described this condition in 1942, it was not until 1956 that other researchers reported that many boys with this description had 47 chromosomes in each cell of their bodies instead of the usual number of 46. This extra sex (X) chromosome causes the distinctive make-up of these boys. All men have one X chromosome and one Y chromosome, but sometimes a variation will result in a male with an extra X. This is Klinefelter Syndrome and is often written as 47,XXY. There are other, less common variations such as 48,XXYY; 48,XXXY; 49,XXXXY; and XY/XXY mosaic. All of these are considered Klinefelter Syndrome variants.

The major effect of the extra X chromosome in boys with Klinefelter Syndrome seems to be the function of the testes. The testes produce the major male sex hormone testosterone and the amount of this hormone may be decreased in boys with Klinefelter Syndrome. When these boys reach 10-12 years of age, it is helpful to measure their blood hormone levels periodically (e.g., yearly) to see if they are normal. If the testosterone level is low, resulting in diminished sexual changes which boys otherwise undergo during puberty, or if other characteristics are present which seem to point to poor metabolism of the existing hormone levels, then treatment with male sex hormones is usually very beneficial.

The most common form of treatment involves administering depotestosterone, a synthetic form of testosterone, by injection once a month. The dose gradually needs to be increased and given more frequently as the boys get older. Treatment should result in normal progression of physical and sexual development, including pubic hair growth, an increase in the size of the penis and scrotum (but not the testes), beard growth, deepening of the voice, and increase in muscle bulk and strength.

Because they often don’t appear any different from anyone else, many XXY males probably never learn of their extra chromosome. However, if they are to be diagnosed, chances are greatest at one of the following times in life: before or shortly after birth, early childhood, adolescence, and in adulthood (as a result of testing for infertility).

In recent years, many XXY males have been diagnosed before birth, through amniocentesis or chorionic villus sampling (CVS). In amniocentesis, a sample of the fluid surrounding the fetus is withdrawn. Fetal cells in the fluid are then examined for chromosomal abnormalities. CVS is similar to amniocentesis, except that the procedure is done in the first trimester, and the fetal cells needed for examination are taken from the placenta. Neither procedure is used routinely, except when there is a family history of genetic defects, the pregnant woman is older than 35, or when other medical indications are present.

The next most likely opportunity for diagnosis is when the child begins school. A physician may suspect a boy is an XXY male if he is delayed in learning to talk and has difficulty with reading and writing. XXY boys may also be tall and thin and somewhat passive and shy. Again, however, there are no guarantees. Some of the boys who fit this description will have the XXY chromosome count, but many others will not.

A few XXY males are diagnosed at adolescence, when excessive breast development forces them to seek medical attention. Like some chromosomally normal males, many XXY males undergo slight breast enlargement at puberty. Of these, only about a third-10 percent of XXY males in all-will develop breasts large enough to embarrass them.

The final chance for diagnosis is at adulthood, as a result of testing for infertility. At this time, an examining physician may note the undersized testes characteristic of an XXY male. In addition to infertility tests, the physician may order tests to detect increased levels of hormones known as gonadotropins, common in XXY males.

A karyotype is used to confirm the diagnosis. In this procedure, a small blood sample is drawn. White blood cells are then separated from the sample, mixed with tissue culture medium, incubated, and checked for chromosomal abnormalities, such as an extra X chromosome.

Dr. Alan D. Rogol, of the University of Virginia, Charlottesville, and also a member of the Klinefelter Syndrome and Associates Scientific Advisory Committee has let us know that University of Virginia is up and running for a comprehensive pediatric Klinefelter Syndrome Clinic at the Kluge Children’s Rehabilitation Center. Working closely with him on this project is Dr. Sharon Hostler, the Medical Director of the Kluge Children’s Rehabilitation Center.

Doctors Rogol and Hostler have asked for volunteers to help the University of Virginia in an advisory capacity. The parents and young adults would be a part of a process in advising the steering committee from the “user/consumer” perspective. They have asked Klinefelter Syndrome and Associates to let local member families know of their needs.