Klinefelter Syndrom Essay, Research Paper

KLINEFELTER SYNDROME

THE XXY BOY IN THE CLASSROOM

Although there are exceptions, XXY boys are usually well behaved in the classroom. Most are shy, quiet, and eager to please the teacher. But when faced with material they find difficult, they tend to withdraw into quiet daydreaming. Teachers sometimes fail to realize they have a language problem, and dismiss them as lazy, saying they could do the work if they would only try. Many become so quiet that teachers forget they’re even in the room. As a result, they fall farther and farther behind, and eventually may be held back a grade.

CAUSES

No one knows what puts a couple at risk for conceiving an XXY child. Advanced maternal age increases the risk for the XXY chromosome count, but only slightly. Furthermore, recent studies conducted by NICHD grantee Terry Hassold, a geneticist at Case Western Reserve University in Cleveland, OH, show that half the time, the extra chromosome comes from the father.

Dr. Hassold explained that cells destined to become sperm or eggs undergo a process known as meiosis. In this process, the 46 chromosomes in the cell separate, ultimately producing two new cells having 23 chromosomes each. Before meiosis is completed, however, chromosomes pair with their corresponding chromosomes and exchange bits of genetic material. In women, X chromosomes pair; in men, the X and Y chromosome pair. After the exchange, the chromosomes separate, and meiosis continues.

In some cases, the Xs or the X chromosome and Y chromosome fail to pair and fail to exchange genetic material. Occasionally, this results in their moving independently to the same cell, producing either an egg with two Xs, or a sperm having both an X and a Y chromosome. When a sperm having both an X and a Y chromosome fertilizes an egg having a single X chromosome, or a normal Y- bearing sperm fertilizes an egg having two X chromosomes, an XXY male is conceived.

DIAGNOSIS

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.

“If I were going to say something to parents who have had a prenatal diagnosis, it would be ‘You are so lucky that you know,” said Melissa, the mother of one XXY boy. “Because there are parents who don’t know that their sons have this problem. And they will never be able to help them lead a normal life. But you can.”

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.

TESTOSTERONE TREATMENT

Ideally, XXY males should begin testosterone treatment as they enter puberty. XXY males diagnosed in adulthood are also likely to benefit from the hormone. A regular schedule of testosterone injections will increase strength and muscle size, and promote the growth of facial and body hair.

In addition to these physical changes, testosterone injections often bring on psychological changes as well. As they begin to develop a more masculine appearance, the self-confidence of XXY males tends to increase. Many become more energetic and stop having sudden, angry changes in moods. What is not clear is whether these psychological changes are a direct result of testosterone treatment or are a side benefit of the increased self confidence that the treatment may bring. As a group, XXY boys tend to suffer from depression, principally because of their scholastic difficulties and problems fitting in with other males their age. Sudden, angry changes in mood are typical of depressed people.

Other benefits of testosterone treatment may include decreased need for sleep, an enhanced ability to concentrate, and improved relations with others. But to obtain these benefits an XXY male must decide, on his own, that he is ready to stick to a regular schedule of injections.

Sometimes, younger adolescents, who may be somewhat immature, seem not quite ready to take the shots. It is an inconvenience, and many don’t like needles.

Most physicians do not push the young-men to take the injections. Instead, they usually recommend informing XXY adolescents and their parents about the benefits of testosterone injections and letting them take as much time as they need to make their decision.

Individuals may respond to testosterone treatment in different ways. Although the majority of XXY males ultimately will benefit from testosterone, a few will not.

To ensure that the injections will provide the maximum benefit, XXY males who are ready to begin testosterone injections should consult a qualified endocrinologist (a specialist in hormonal interactions) who has experience treating XXY males.

Side effects of the injections are few. Some individuals may develop a minor allergic reaction at the injection site, resulting in an itchy welt resembling a mosquito bite. Applying a non-prescription hydrocortisone cream to the area will reduce swelling and itching.

In addition, testosterone injections may result in a condition known as benign prostatic hyperplasia (BPH). This condition is common in chromosomally normal males as well, affecting more than 50 percent of men in their sixties, and as many as 90 percent in their seventies and eighties. In XXY males receiving testosterone injections, this condition may begin sometime after age 40.

The prostate is a small gland about the size of a walnut, which helps to manufacture semen. The gland is located just beneath the bladder and surrounds the urethra, the tube through which urine passes out of the body.

In BPH, the prostate increases in size, sometimes squeezing the bladder and urethra and causing difficulty urinating, “dribbling” after urination, and the need to urinate frequently.

XXY males receiving testosterone injections should consult their physicians about a regular schedule of prostate examinations. BPH can often be detected early by a rectal exam. If the prostate greatly interferes with the flow of urine, excess prostate tissue can be trimmed away by a surgical instrument that is inserted in the penis, through the urethra.

CHROMOSOMAL VARIATIONS

Occasionally, variations of the XXY chromosome count may occur, the most common being the XY/XXY mosaic. In this variation, some of the cells in the male’s body have an additional X chromosome, and the rest have the normal XY chromosome count. The percentage of cells containing the extra chromosome varies from case to case. In some instances, XY/XXY mosaics may have enough normally functioning cells in the testes to allow them to father children.

A few instances of males having two or even three additional X chromosomes have also been reported in the medical literature. In these individuals, the classic features of Klinefelter syndrome may be exaggerated, with low I.Q. or moderate to severe mental retardation also occurring.

In rare instances, an individual may possess both an additional X and an additional Y chromosome. The medical literature describes XXYY males as having slight to moderate mental retardation. They may sometimes be aggressive or even violent. Although they may have a rounded body type and decreased sex drive, experts disagree whether testosterone injections are appropriate for all of them.

One group of researchers reported that after receiving testosterone injections, an XXYY male stopped having violent sexual fantasies and ceased his assaults on teenaged girls. in contrast, Dr. Robinson found that testosterone injections seemed to make an XXYY boy he had been treating more aggressive.

Scientists admit, however, that because these cases are so rare, not much is known about them. Most of the XXYY males who have been studied were referred to treatment because they were violent and got into trouble with the law. It is not known whether XXYY males are inherently aggressive by nature, or whether only a few extreme individuals come to the attention of researchers precisely because they are aggressive

INFERTILITY

The vast majority of XXY males do not produce enough sperm to allow them to become fathers. If these men and their wives wish to become parents, they should seek counseling from their family physician regarding adoption and infertility.

However, no XXY male should automatically assume he is infertile without further testing. In a very small number of cases, XXY males have been able to father children.

In addition, a few individuals who believe themse to be XXY males may actually be XY/XXY mosaics. Along with having cells with the XXY chromosome count, these males may also have cells with the normal XY chromosome count. If the number of XY cells in the testes is great enough, the individual should be able to father children.

Karyotyping, the method traditionally used to identify an individual’s chromosome count, may sometimes fail to identify XY/ XXY mosaics. For this reason, a karyotype should never be used to predict whether an individual will be infertile or not.

Resources

http://www.nih.gov/health/chip/nichd/klinefelter/#xwhat