Klinefelter’s Syndrome Essay, Research Paper

KLINEFELTER’S SYNDROME

Klinefelter’s syndrome is a genetic disorder that is only found in males. Until recently you could not detect this disorder until after puberty, but now they can detect it before birth using amniocentesis. Amniocentesis is the use of a long needle to withdraw amniotic fluid surrounding the fetus. Klinefelter’s syndrome was first discovered by H.F. Klinefelter in 1942. This disease affects about one in one thousand males that are born.

Klinefelter’s syndrome is caused by an extra X chromosome, which is caused by an error in meiosis, to make a combination of XXY, also known as a trisomal karyotype. Although the extra X chromosome is inactive, also known as a Barr body, it still interferes with the development of some male characteristics. Other causes of this disease are excessive alcohol intake, hormone disturbance, and urinary tract infections. Some STD’s such as syphilis and non specific urethritis can cause a male to get this disease also. The older a woman is when she gives birth to her child the more likely the son is to have Klinefelter’s syndrome. Even so, it is not a hereditary disease.

Along with the interference of the development of male characteristics, Klinefelter’s can cause someone to have some mental retardation; the condition does not worsen, nor does it get better, after birth. As the number of X chromosomes increase the severity of mental retardation and malformation will also increase. Some other effects of this disease are the plasma concentration of the male hormone testosterone is often low and they are unable to impregnate a female, in other words they are sterile. Someone with Klinefelter’s syndrome often has trouble learning to read and write this will often times cause them to speak later than others. Klinefelter’s syndrome can cause problems with a males genitals and his endocrine system. Some physical effects of this disease are that a person tends to be taller, have longer limbs, and their testes are small and firm. In some cases a male may form breast tissue. Many males infected with this disease will also have reduced muscle development. There is also excessive loss of sexual stimulating hormones in the urine.. It is said that a male with Klinfelter’s syndrome is most often very talented in arts and mechanics.

One of the main ways of diagnosing a patient with Klinefelter’s syndrome is through karyotyping the patient. This is done by examining the arrangement of chromosomes on DNA from samples of blood, bone marrow, amniotic fluids, and placental tissue. Another way that doctors are using to diagnose a patient is through the presence of Barr bodies taken form oral swabs. Doctors have now found that Klinefelter’s syndrome, and many other genetic disorders, can be detected before birth. This process is called amniocentesis. In this process the doctor will use an ultra sound, ultra sonic waves produce an image of the fetus on a screen, to monitor the position of the fetus while inserting a long hollow needle through the mother’s abdominal wall and into the uterus. The doctor will then withdraw a small amount of amniotic fluid, it contains cells shed by the fetus. Then they will let the cells grow in a culture for a few weeks.

After the cells have had a chance to grow the doctors will test them for certain genetic disorders.

Some ways that a person with Klinefelter’s syndrome can be treated is through language therapy. Language therapy works best if it is started at two years of age. It can help lessen speech problems when they get older. They can also get testosterone therapy. This type of therapy can be oral, injections, or through a patch. Testosterone therapy works best when started at eleven to twelve years of age. A doctor could prescribe a patient with Vitamin B and Vitamin E supplements to take. Along with the vitamin supplements a doctor could also prescribe human chorionic gonadotrophins, a hormone replacement. If the patient feels the need to, he can get surgery to correct anatomical abnormalities of the reproductive system.

In rare cases males with Klinefelter’s syndrome will be more aggressive than others. Because of this studies say that they are more probable to be incarcerated. If some one with Klinefelter’s syndrome is aggressive this could cause his friends to act differently towards him. In some cases males with this disease may tend to be more shy and more of an introvert. Often times if a person’s acquaintances knew that he had Klinefelter’s syndrome they might tease him. If not tease him they will treat him as if he is a special case and he should be treated differently when in reality that is probably the last thing anyone would want. He is still a person. If someone has this disease it would be better to treat them like you would anyone else. Many of these social implications can be lessened with a loving and caring family, caring friends, and in some cases therapy might help.

Klinefelter’s syndrome is a very common disease even though it goes undetected in many males. Not many people know about this and many other genetic diseases which could be affecting someone in their family or just someone you know. Researching a disease that someone a person knows could help many people in better understanding what that he or she is going through and also it will help them realize better ways to help the person..