

# Research into the history of klinefelters syndrome



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Many chromosomal abnormalities occur early in development and involve the sex chromosomes. Klinefelter’s Syndrome falls directly into this category. Klinefelter’s Syndrome is a genetic condition affecting the male population. The following information observes who discovered Klinefelter’s Syndrome and when it was first diagnosed. The etiology or genetic and environmental factors of the condition are discussed as well as complete description of the disability and the clear diagnostic criteria. The affects of Klinefelter’s Syndrome are more recognized in adulthood, when it is typically diagnosed. Many Klinefelter’s patients live out their entire lives without ever knowing they have the condition. Upon conclusion, there should be an adequate amount of information that will give you, the reader, valuable knowledge into the diagnosis and treatment of Klinefelter’s Syndrome.

## History

Klinefelter's Syndrome was first diagnosed in 1942 at the Massachusetts General Hospital in Boston Massachusetts (Schoenstadt, 2006). Dr. Harry Klinefelter was completing his fellowship at the Massachusetts General Hospital when he was assigned to work with Dr. Fuller Albright, also known as the father of endocrinology (Bock, 1993; "Klinefelter Syndrome," 2006). Dr. Klinefelter came to examine nine adult men that had a common set of symptoms during the course of his fellowship ("Klinefelter Syndrome," 2006). Dr. Klinefelter organized a case study that involved these nine men and their similarities and was encouraged by Dr.

Albright to lead the study ("Klinefelter Synrome," 2006). The Journal of Clinical Endocrinology

was published in November of 1942 with the completed case study of these nine men's similar

qualities, which Dr. Klinefelter identified as Klinefelter's Syndrome ("Klinefelter Syndrome,"

2006). The report written by Dr. Klinefelter on these men described them as having testicular dysgenesis, elevated urinary gonadotropins, eunuchoidism, azoospermia, and gynecomastia, all of which have an effect on the underdeveloped size of the testes, the lack of the amount of testosterone produced by the testes, and infertility (Schoenstadt, 2006; Visootsak & Graham, 2006). These adult males also exhibit enlarged breast and sparse facial hair (Schoenstadt, 2006). Two groups found out fourteen years after Dr. Klinefelter's original description of the syndrome, that the buccal mucosal  
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cells contained an extra chromatin mass or that the cells were chromatin positive (Klinefelter, 1966). Although the patients were described as having a positive female sex chromatin, Dr. Klinefelter states that the patients are phenotypic males and should never be considered otherwise (Klinefelter, 1996).

Fourteen years after Dr. Klinefelter first diagnosed Klinefelter's Syndrome, another case study was performed to further understand the characteristics that define the condition (" Klinefelter Syndrome," 2006). In 1956, Dr. Joe Hin Tjio and Dr. Albert Lavan took the research further to determine the factors that made those men with Klinefelter's Syndrome dissimilar from normal adult males through genetic research (" Klinefelter Syndrome," 2006; Schoenstadt, 2006). With the advanced experimental methodology available, Dr. Joe Hin Tjio and Dr. Albert Lavan found that humans had 23 pairs of chromosomes, confirming 46

chromosomes, which prior to this time there was thought to be 48 chromosomes (Bock, 1993;

" Klinefelter Syndrome," 2006). This clarification by Dr. Tjio and Dr. Lavan is the basic

foundation for modern cytogenetics, the study of chromosomes and diseases originating from

numerical or structural abnormalities in chromosomes (" Klinefelter Syndrome," 2006). Dr. Tjio and Dr. Lavan discovered that men with symptoms of Klinefelter's Syndrome had an extra sex chromosome that

created the chromosomal arrangement of XXY, which is distinct to the normal male chromosomal arrangement of XY (“ Klinefelter Syndrome,” 2006; Schoenstadt, 2006,). Klinefelter Syndrome was still believed to be an endocrine disorder of unknown etiology at this time (Visootsak & Graham, 2006).

In 1959, just three years after Dr. Tjio and Dr. Lavan made their historical discoveries, an English researcher by the name of Dr. Patricia Jacobs and her associate Dr. J. A. Strong published a study supplementing earlier studies (“ Klinefelter Syndrome,” 2006; Noble, 2003). Dr. Jacobs and Dr. Strong found the link between the endocrinal disease and the extra X sex chromosome (Noble, 2003). Dr. Jacobs linked forty-seven chromosomes in Klinefelter’s Syndrome males and determined it to be the X chromosome, which she considered an aneuploidy defined as an unusual number of chromosomes and labeled 47, XXY (Bock, 1993; “ Klinefelter Syndrome,” 2006; Schoenstadt, 2006).

The 1970s brought forth a larger examination of males born with Klinefelter’s Syndrome (Bock, 1993). During this time doctors began screening newborn male babies for the extra chromosome (Bock, 1993; Visootsak & Graham, 2006). The most significant of the studies done at this time was sponsored by the National Institute of Child Health and Human Development (NICHD) whom examined over forty thousand infants for this extra chromosome (Bock, 1993;

Visootsak & Graham, 2006). This study was important for the reason that most studies done

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prior to the 1970s were biased and primarily done on adult males in mental institutions and the

penal system (Visootsak & Graham, 2006). At this time is when the prevalence of Klinefelter's Syndrome was noticed as frequently as one in five hundred to one in one thousand male newborns (Bock, 1993; Visootsak & Graham, 2006). Also observed in this study was the reduction in speech and language abilities as well as decreased reading and spelling achievement (Bock, 1993; Visootsak & Graham, 2006). Along with these disabilities, Klinefelter's patients are characterized by an increased tendency towards fertility, endocrinal, and psychiatric disorders (Noble, 2003). This study demonstrated that most but not all of these males born with the extra chromosome will have these characteristics, and many demonstrate varying degrees of the characteristics (Bock, 1993; Visootsak & Graham, 2006).

Based on this research it has been found that the extra X chromosome that causes Klinefelter's Syndrome is very common, however, the symptoms and characteristics that are most recognizable are quit uncommon (Bock, 1993).

Most males are not diagnosed as having Klinefelter's Syndrome until they reach adulthood, and many that have the syndrome are never diagnosed as having this chromosomal defect at all (Bock, 1993). One pediatrician at the University of Colorado Medical School in Denver and the director of the National Institute of Child Health and Human Development (NICHD) during the major screening research referred to these newborn males as not having Klinefelter's Syndrome because of the possibility that the characteristics may not develop into a syndrome (Bock, 1993; Visootsak & Graham, 2006).

## **Etiology and Genetic Factors**

Every normal human cell has 46 chromosomes that are made up of 23 pairs (Stewart, 2007). Of these 23 pairs, there are 22 that are exactly the same in both males and females called

autosomes (U. S. National Library of Medicine, 2010). The 23rd pair of sex chromosomes is what makes males and females different in that the male will have only one X and Y chromosome whereas the female will have two copies of the X chromosome (Stewart, 2007; U. S. National Library of Medicine, 2010). During the formation of the egg and the sperm, or gametes, the chromosomes are halved through a process called meiosis (Stewart, 2007; The Dorsey, 2009). Cells that carry a single chromosome such as the X or Y chromosome are called haploid cells (The Dorsey, 2009). When the egg and sperm join carrying 23 chromosomes each they create the fertile egg, or zygote, which has two haploid sets of chromosomes (The Dorsey, 2009). Therefore, the baby receives two copies of each chromosome, 46 total chromosomes, just like the parents (Stewart, 2007).

The extra X in Klinefelter Syndrome is caused from either nondisjunction or anaphase lag. Nondisjunction occurs when the chromosome pairs do not separate as they are intended in the meiosis I or meiosis II stage (Pineyard & Zipf, 2003; Stewart, 2007). When this happens there may be a chromosome pair with 24 chromosomes instead of the 23 chromosomes (Stewart, 2007). If this chromosome pair of 24 joins with an egg or sperm with 23 chromosomes then it results in a karyotype with 47 chromosomes (Stewart, 2007). In this case there will be three copies of chromosomes rather than the usual two

copies of chromosomes (Stewart, 2007). The sperm or egg may donate the extra X chromosome at conception causing a chromosomal abnormality

(Mayo Foundation for Medical Education and Research, 2008; Stewart, 2007).

This forms the XXY chromosomal formation, which is diagnosed as Klinefelter's Syndrome. At least half of 47, XXY conceptions are spontaneously aborted (Pineyard & Zipf, 2003). The chromosomal

abnormality is random and not known to be caused by any environmental factors (Genetic Science Learning Center, 2010; Mayo Foundation for Medical Education and Research, 2008; National Institute of Health, 2007).

This anomaly happens entirely by chance and is unrelated to family history prior to the male child's birth (Mayo Foundation for Medical Education and Research, 2008). This is to say that the male embryo's likelihood of being born with Klinefelter's Syndrome is not increased or decreased by what the parent does or does not do (Mayo Foundation for Medical Education and Research, 2008). Klinefelter's Syndrome is not affected by race (Chen, 2010). This is a completely random occurrence of the sex chromosomes not successfully separating during the formation of the egg or the sperm (Genetic Science Learning Center, 2010). Once this occurs the extra chromosome is then copied into every cell of the embryo (Genetics Science Learning Center, 2008).

There are extremely rare cases when there may be three or four extra X chromosomes in all copies of the cells known as 48, XXXY or 49, XXXXY (Stewart, 2007). The 49, XXXXY mosaic is also known as Fraccaro's Syndrome and is the most rare form of Klinefelter's Syndrome (Duenas et al., <https://assignbuster.com/research-into-the-history-of-klinefelters-syndrome/>



2007). This rare chromosomal abnormality results in more exaggerated features of Klinefelter's Syndrome (Stewart, 2007). There are instances where an extra X chromosome is found in only some of the cells (Stewart, 2007). This can be found as two different chromosomal patterns (Stewart, 2007). One pattern occurs when some cells have 46 chromosomes and some have 47 chromosomes (Stewart, 2007). The other pattern is called the mosaic XXY syndrome, or chromosomal mosaicism, and affects approximately six percent of these cases, with the most rare cases being the 48, XXXY or the 49, XXXXY, or other arrangements of X chromosomes (Stewart, 2007).

The mosaic XXY syndrome occurs only after conception from a mistake in cell division (Stewart, 2007). Anaphase lag is a result of a gamete lacking a sex chromosome (Klinefelter, 1966). When this chromosome lags it is not incorporated into the new cell during the mitosis stage (Klinefelter, 1996). Anaphase lag is thought to be a reason for the mosaic variations of Klinefelter's Syndrome (Klinefelter, 1966).

Although the chromosomal abnormality of 49, XXXXY is considered to be a variant form of Klinefelter's Syndrome, it appears to have a very independent, distinct phenotype (Duenas et al., 2007). Males that show the 49, XXXXY chromosomal structure have much more severe clinical features than that of a Klinefelter's Syndrome male (Duenas et al., 2007). This is the most rare of the Klinefelter's Syndrome variants and has been reported in over one hundred cases with the frequency being approximately 1 in 85, 000 newborn males (Duenas et al., 2007).

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There have been reports of an even more extreme variant of Klinefelter's Syndrome mosaic in newborn males (Duenas et al., 2007). This variant is a 47, XXY/48, XXXY/49, XXXXY mosaicism and has only been reported in three cases according to a researcher in Mexico (Duenas et al., 2007). This means that the male newborn would have the whole spectrum of XY variations.

Another variant that affects only males is the 46, XX chromosomal variation (Bock, 1993). This condition occurs when individuals have two X chromosomes in each cell, but are male in appearance. These individuals have male external genitalia. These individuals also have

small, undescended testes possibly along with an urethra opening on the underside of the penis. A small amount of 46, XX Males have external genitalia that don't clearly resemble either male or female genitalia. These individuals are typically raised male. Phenotypically, there are three groups of these sex-reversed individuals. The first group includes phenotypically normal XX Males, the second group includes the males with genital ambiguities, and the third group is the true hermaphrodites (Bock, 1993).

## **Description of Characteristics or Traits**

Klinefelter's Syndrome has only one constant physical description and that is the small testicular size (Visootsak & Graham, 2003). Boys with Klinefelter's Syndrome have variable

phenotypic characteristics with no obvious facial dysmorphology (Visootsak & Graham, 2003). The presence of gynecomastia, or enlarged breast, and other findings of eunuchoid body habits and sparse body hair vary (Visootsak & Graham, 2003). Eunuchoid or eunuchoidism is defined as an abnormal

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condition in males, characterized by underdeveloped reproductive organs with some female characteristics, such as a higher voice or the lack of facial and body hair that results in the lack of male sex hormones (Eunuchoidism, n. d.). Gonadotropins are produced by glands, such as the pituitary, and can result in sparse body hair when not produced adequately (Gonadotropin, 2010). The medical dictionary states that eunuchoidism is marked by a deficiency of sexual development with the persistence of prepubertal characteristics, and often

has the presence of characteristics that are typical of the opposite sex (Eunuchoidism, n. d.). Another likely characteristic is azoospermia (Schoenstadt, 2006; Visootsak & Graham, 2006). Azoospermia is defined as having little or no sperm count (Azoospermia, 2010). Testicular

dysgenesis, or gonadal dysgenesis, is another characteristic of Klinefelter's Syndrome (Schoenstadt, 2006; Visootsak & Graham, 2006). Testicular dysgenesis is considered a reproductive system developmental disorder that causes a progressive loss of primordial germ cells, or cells that create gametes, in the developing gonads of an embryo (Gonadal dysgenesis, 2010). This gonadal dysgenesis can lead to the extremely hypoplastic, or underdeveloped, and disfunctioning gonads mainly composed of fibrous tissues (Gonadal dysgenesis, 2010).

Most infants and children with the 47, XXXY chromosomal abnormalities go through normal growth stages. It is not until puberty that the Klinefelter's Syndrome characteristics or traits become more prevalent and noticeable (Visootsak & Graham, 2003). There is a significant increase in height

between the ages of five and eight (Visootsak & Graham, 2003). Another characteristic of Klinefelter's Syndrome is the elongated length of arms and legs (Klinefelter, 1966). There is a decrease in androgen production that causes the secondary sexual characteristics to not fully develop (Visootsak & Graham, 2003). An androgen is any substance such as androsterone or testosterone that supports male characteristics (Androgen, n. d.). Typically Klinefelter's males are infertile (Visootsak & Graham, 2003). However, there have been cases of impregnation without the assistance of medical technology (Visootsak & Graham, 2003).

Autoimmune diseases such as juvenile arthritis can also be present in Klinefelter's adolescents. Whereas boys with Klinefelter's Syndrome are generally tall with long limbs and

remain thin until puberty, they tend to suffer from obesity latter in life.

Neurocognitive effects of Klinefelter's Syndrome may be more subtle than that of the physical stigmata. Klinefelter's

males have been found to have relative deficits on verbal IQ subtests and have verbal IQ scores around 20 points lower than those of unaffected siblings. There are also deficits in articulation, word finding, phonemic processing, verbal memory, language comprehension, oral expression problems, as well as linguistic processing speed. It seems that the speech/language problems and some motor deficits are most common in Klinefelter's males that have an extra X chromosome. Ninety-two percent of individuals with Klinefelter's Syndrome confirm difficulty learning to read. Seventy percent had reading achievement discrepancies or absolute reading

deficits on standardized testing. A group of boys with mental retardation and suspicion of fragile X were subject to a genetic screening and the results showed that eight of these boys had Klinefelter's Syndrome. Most of the more extreme verbal, visuospatial, and motor skills, such as found in mental retardation and fragile X syndrome are typically spared. However, some boys with Klinefelter's Syndrome suffer from poor manual dexterity and are commonly found to be clumsy and below average in sports (Wodrich & Tarbox, 2008).

There are many different factors that may underlie linguistic and reading problems. One possibility is a dysfunction of the left hemisphere that may be related to diminished gray matter or a lack of hemispheric asymmetry, or both. It is also possible that executive and frontal deficits may be a cause (Wodrich & Tarbox). There is evidence that language is a fundamental issue for Klinefelter's children and this can result in further scholastic issues. This problem seems to manifest as dyslexia as defined by poor reading in the setting of normal intelligence.

Klinefelter's males have also been observed to have difficulties with arithmetical functions. The deficits in auditory processing and verbal memory are the two key cognitive processes that

underlie these difficulties. These deficits are also true for normal chromosomal children with dyslexia. The findings are supportive of the concept that defects in frontal systems seem to be caused by a language-based, left frontal-systems problem (Geschwind & Dykens, 2004). Adult Klinefelter's males have reported to have difficulties with mental flexibility

(Wodrich & Dykens, 2004). Even with these studies, it should be noted that not all adults that have Klinefelter's Syndrome show these classic patterns of verbal deficits that are observed in children (Geschwind & Dykens, 2004). However, these findings are not appropriate for all Klinefelter's males, many of which complete high school and move on to post-secondary education successfully (Wodrich & Tarbox, 2008).

Two characteristics that has been falsely associated with Klinefelter's males in the past, is sociopathy and criminal behavior. There is, contrary to this belief, fewer psychiatric problems reported among these individuals. However, there are commonly traits of introversion, unassertiveness, and a paucity or lack of ambition. There are also possible traits of impulsivity and social inappropriateness (Wodrich & Tarbox, 2008). A Reiss Profile of Fundamental Goals measurement was used to assess the degrees of which Klinefelter's males were motivated in 15 domains (Geschwind & Dykens, 2004). The Reiss Profile generates a profile that is based on the motivational sensitivities across the domains of aversive sensations, citizenship, family, curiosity, honor, independence, food, order, physical exercise, rejection, power, sex, social contact, vengeance, and social prestige. The Reiss Profile is a well-established psychometric measure that is being used more and more to assess people with and without mental retardation

(Geschwind & Dykes, 2004). The results suggested that the Klinefelter's male group was not

particularly motivated by the need for social prestige, independence, or the desire to seek vengeance. This group was also not motivated to avoid physical pain. The general motivator for all the Klinefelter's males in this group was curiosity. There were no age effects to this study (Geschwind & Dykes, 2004).

### **Characteristics in Adulthood**

There is a persistent deficiency of androgen in adulthood that can result in the loss of libido, decreased muscle bulk and tone, decreased bone density, a propensity for thromboembolism (an obstruction in a vein or artery from a blood clot), and an increased risk of mortality from cardiovascular and diabetic complications. A common characteristic for Klinefelter's adults is gynecomastia (Wattendorf & Muenke, 2005). Gynecomastia involves the risk of developing breast carcinoma. There is 200 times more of a risk for Klinefelter's males to develop breast carcinoma than other karyotypically normal individuals. This may be a result of the estradiol (the prominent sex hormone in females) to testosterone ratio being so much higher than karyotypically normal men. Another possibility is that it is caused by the increase of peripheral conversion of testosterone to estradiol (Visootsak & Graham, 2006).

There are different views as to whether Klinefelter's adult males are more aggressive or have a greater chance of psychological issues depending on the resource. One study describes the differences as relative to individual

testosterone levels and the age at which they received the diagnosis (Morris, Jackson, & Hancock, 2009). Equally, there is an impact from the way the diagnosis is reacted to by the Klinefelter's male, the family, and friends or peers. The seven major themes that emerged from this study were the diagnosis, the testosterone treatments, health

care problems, appearance, self-identity, relationships, and school and education. Of the Klinefelter's adults studied, 60 % reported clinical levels of anxiety and 34% had clinical levels of depression. The results of this study show that a prolonged lack of testosterone can have far reaching negative effects on the Klinefelter's adult (Morris, Jackson, & Hancock, 2009). The historical studies show a disturbingly increased risk for psychiatric disturbance, criminality, and mental retardation. However, these results are outdated and extremely questionable given the initial examinations were given to institutionalized populations (Chen, 2010).

## **Differential Diagnosis**

Classic Klinefelter's Syndrome, 47, XXY, cases make up approximately 80-90% of all Klinefelter's diagnosis. There are approximately 6-10% of these cases that are mosaics, which are the cells with 46, XY/47, XXY; 46, XY/48, XXXY; and 47, XXXY/48, XXXY (Chen, 2010; Visootsak & Graham, 2003; Visootsak & Graham, 2006). In 5% of the cases there are two X chromosomes without a Y chromosome or 46, XX (Visootsak & Graham, 2006). The other cases were karyotypes 48, XXXY, 48, XXYY, 49, XXXXY, and 49, XXXYY (Visootsak & Graham, 2003). Approximately 1% of these cases are due to a structurally abnormal X with a normal X and Y chromosome described as karyotypes 47, X, i(Xq)Y and 47, X, del(X)Y (Chen, 2010).

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Klinefelter's Syndrome variants occur much less frequently than the classic 47, XXY chromosomal abnormality (Bock, 1993; Visootsak & Graham, 2006).

Klinefelter variant 48, XXXY is characterized by being average or tall stature with ocular hypertelorism, which are widely spaced or deep set eyes; flat nasal bridge; curving of the fifth finger, or clinodactyly. Other characteristics are small penis and testicles with hypergonadotropic

hypogonadism, which is the absence or decrease in function of the male testes. These individual's intelligence quotients range from 40-60. Variant 48, XXYY is characterized by having a tall stature, an eunuchoid habitus with long legs, sparse body hair, small testicles and penis, hypergonadotropic hypogonadism and gynecomastia. These individual's intelligence quotients range from 60-80.

Males with variant 49, XXXXY are severely affected. They have smaller than average

head circumference also known as microcephaly, short stature with ocular hypertelorism, flat nasal bridge, and upslanting palpebral fissures. Cleft palates are present along with small genitalia and a heart defect known as patent ductus arteriosus. These individual's intelligence quotients range from 20-60. (Visootsak and Graham, 2003).

Klinefelter's Syndrome 47, XXY, has no major physical signs, which explains why it may go undiagnosed or misdiagnosed throughout an individual's life. Also with no physical signs, it is truly only diagnosed when genetic testing occurs for a variety of unrelated reasons. Klinefelter's Syndrome may be

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diagnosed prenatally or during early childhood, as an adolescent during puberty, or as an adult when there are recognized fertility problems (Bock, 1993). Klinefelter's Syndrome can be diagnosed prenatally through amniocentesis or chorionic villus sampling (Bock, 1993). These tests are normally done if the pregnant woman is older than 35, if there is a family history of genetic defects, or when other medical indications exist (Bock, 1993). A pediatrician may suspect a male child as having Klinefelter's Syndrome if there are delays in learning to talk or difficulties in reading and writing as well as physical abnormalities during adolescence (Bock, 1993).

## **Treatments and Interventions**

All hope is not lost when it comes to the treatment and interventions of the undesirable traits and characteristics that males diagnosed with Klinefelter's Syndrome may display or develop. It is recommended that Klinefelter's males have a comprehensive neurodevelopmental

evaluation as soon as they have been diagnosed. A multidisciplinary developmental evaluation can determine the appropriate treatments during infancy and early childhood. These treatments may include physical therapy, infant simulation programs, and speech therapy (Wattendorf, 2005). If the language difficulties are detected in childhood, then there is more of a possibility for intervention.

The language barriers that Klinefelter's males may have to cope with can not only affect their academics, it can obstruct their building of social relationships and learning social skills necessary for these relationships. Here is where the Klinefelter's child could benefit from a social skills training

program. In a social skills training program, the Klinefelter's child will be able to practice talking and listening, observing children's making friends processes, sharing of information, attitudes, and beliefs. This will also assist them in proper classroom behavior and playground behavior. Language disabilities and barriers can prevent Klinefelter's males from fitting in socially, so this kind of intervention and assistance can benefit the child greatly. Hearing can be an issue if frequent ear infections occur. Hearing test and screens should be done to ensure that a hearing impairment is not a part of the language difficulties. If the Klinefelter's child is not communicating effectively with single words by the ages of 18 to 24 months, then consultation with a speech and language pathologist will be very beneficial (" Klinefelter Syndrome Information," 2002).

Teachers should be informed of the difficulties that a Klinefelter's child may be dealing

with in the classroom. A teacher may consider the Klinefelter's child to be lazy and daydreaming and a teacher may even forget the child is even in the room. This can result in the Klinefelter's

child falling behind and eventually being held back a grade. Under the Public Law 94-142, the

Individuals with Disabilities Education Act, adopted by Congress in 1975, all children with

disabilities have a right to a free, and appropriate public education (“

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Information,” 2002).

Once the Klinefelter’s male reaches puberty there is usually an inability to produce a normal amount of testosterone. This along with hypogonadism can result in impaired bone mineral density and skeletal muscle development. Also associated with testosterone deficiency is a decrease in libido and energy (Wattendorf & Muenke, 2005). Androgen therapy or Testosterone Treatment should begin by time the Klinefelter’s male reaches middle school, approximately 12 to 14 years of age, based on the level of pubertal development (“ Klinefelter Syndrome Information,” 2002; Wattendorf & Muenke, 2005). Testosterone Treatment will ultimately increase the muscle size and strength, as well as, promoting the growth of body and facial hair. It must be noted that Testosterone Treatments can also bring on psychological changes. It is important to adequately inform the parent(s) and the child of these changes so that they can make the most informed decision (“ Klinefelter Syndrome Information,” 2002). There are different ways to receive Testosterone Treatment and that is through injections, transdermal (patches, gels, or creams), orally, or implantation. The kind of testosterone injection will depend

mainly on the dosage used and the country in which you receive the injections. Some injectable testosterone esters are Testosterone enanthate, Testosterone cypionate, Sustanon, Testosterone propionate, Testosterone phenylpropionate, Omnadren, and Aqueous testosterone suspension.

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Types of transdermal patches are Androderm and Testosterone TTS. Two different kinds of

testosterone gels and creams are Androgel, and Testim. A few oral supplements include

Methyltestosterone and Testosterone undecanoate. The last form of Testosterone Treatment is the Subcutaneous testosterone pellet, which is delivered by implanting a pellet of pure, crystalline testosterone under the skin of the buttocks or abdomen (“ Testosterone Types and Delivery, n. d.).

Adult males with Klinefelter’s Syndrome usually develop gynecomastia which predisposes men to breast cancer. Therefore, it is important that Klinefelter’s males do monthly breast examinations. If gynecomastia causes psychological or physical problems, then possible treatment would be cosmetic surgery to remove the breast tissue (Wattendorf & Muenke, 2005). Swerdlow et. al (2005) stated that men with Klinefelter Syndrome have elevated risks of several cancers. Prostate cancer, along with breast cancer was more prevalent. Men with Klinefelter Syndrome are also at a substantially higher risk for non-Hodgkin lymphoma, and possibly lung cancer. Breast cancer risk is higher in 47, XXY mosaics. Adult males may face possible infertility issues due to the lack of testosterone production, but if diagnosed early on, this can be minimized and they will be able to reproduce without outside assistance.

## **Summary**

Klinefelter Syndrome is one of the more recently discovered medical syndromes. Klinefelter Syndrome is not one that causes major dysfunctions  
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and is usually only discovered during genetic testing for infertility or during prenatal testing due to maternal age or prior genetic issues within the family. Because Klinefelter Syndrome has not had a lot of research until the last few years, there is no federal funding set aside for this syndrome. Families with sons that are found to be affected by it have no real support system that is knowledgeable of this syndrome and have to research on their own and create resources to fit their situation as none are available in most areas.