International Journal of Applied and Advanced Scientific Research (IJAASR)
Impact Factor: 5.255, ISSN (Online): 2456 - 3080
(www.dvpublication.com) Volume 2, Issue 1, 2017

KLINEFELTER SYNDROME

D. Tamilselvi
Associate Professor, Department of Pediatric Nursing, Sree Balaji College of Nursing, Bharath University, Chennai, Tamilnadu

Cite This Article: Tamilselvi, "Klinefelter Syndrome", International Journal of Applied and Advanced Scientific Research, Volume 2, Issue 1, Page Number 79-80, 2017.

Abstract:

Klinefelter syndrome, also known as XXY syndrome, is a fairly common genetic condition found in males only. It occurs when a boy is born with an extra sex chromosome in most or all of his cells. Many boys with Klinefelter syndrome have no signs or symptoms, and some don't even know they have it until later in life. Klinefelter syndrome typically causes a boy's testicles to grow at a slower rate than those of other boys. It also prevents the testicles from producing normal amounts of sperm and the hormone testosterone. Testosterone affects the way a boy develops both physically and sexually. Low hormone levels and problems with sperm production make it difficult or sometimes impossible for a boy with Klinefelter syndrome to father a child later in life.

Key Words: Klinefelter, Syndrome, Chromosome, Meiosis & Aspermatogenesis

Incidence:

Klinefelter syndrome affects 1 in 500 to 1,000 newborn males. Most variants of Klinefelter syndrome are much rarer, occurring in 1 in 50,000 or fewer newborns. Researchers suspect that Klinefelter syndrome is underdiagnosed because the condition may not be identified in people with mild signs and symptoms.

Cause:

Klinefelter syndrome and its variants are not inherited; this would be caused by two non-disjunction events in spermatogenesis, both meiosis I and meiosis II. The duplicated X chromosome in the sperm these chromosomal changes usually occur as random events during the formation of reproductive cells (eggs and sperm) in a parent. An error in cell division called nondisjunction results in a reproductive cell with an abnormal number of chromosomes. For example, an egg or sperm cell may gain one or more extra copies of the X chromosome as a result of nondisjunction. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have one or more extra X chromosomes in each of the body's cells. i.e.48, XXXY.

Signs and Symptoms:

Not all boys with Klinefelter syndrome will have noticeable symptoms. Other boys can have symptoms that are physically apparent or problems with speech, learning, and development. Babies with Klinefelter syndrome typically have weak muscles, reduced strength, and quiet personalities. They also can take longer to do things like sit up, crawl, walk, and speak. Compared with other kids their age, boys with Klinefelter syndrome might have some or all of the following symptoms:

- ✓ a taller, less muscular body
- ✓ broader hips and longer legs and arms
- ✓ larger breasts (a condition called gynecomastia)
- ✓ weaker bones
- ✓ a lower energy level
- ✓ smaller penis and testicles
- ✓ delayed or incomplete puberty (some boys won't go through puberty at all)
- ✓ less facial and body hair following puberty

Diagnosis:

There are three main types of tests used in the diagnosis of Klinefelter syndrome:

- ✓ Complete history and physical examination
- ✓ Hormone testing, which is usually done by taking a blood sample to check for abnormal hormone levels.
- ✓ A chromosome analysis, or karyotype, which is usually done on a blood sample. This test checks the number of chromosomes to see if the XXY syndrome is present.

1. Complete History and Physical Examination: About 10% of Klinefelter cases are found by prenatal diagnosis. The first clinical features may appear in early childhood or, more frequently, during puberty, such as lack of secondary sexual characteristics and aspermatogenesis, while tall stature as a symptom can be hard to diagnose during puberty. Despite the presence of small testes, only a quarter of the affected males are recognized as having Klinefelter syndrome at puberty. Another quarter receives their diagnosis in late adulthood. About 64% of affected individuals are never recognized. Often the diagnosis is made incidentally as a result of examinations and medical visits for reasons not linked to the condition.

2. Hormone Testing, Which is Usually Done by Taking a Blood Sample to Check for Abnormal Hormone Levels: This method may be research of high serum levels of gonadotropins (follicle-stimulating hormone and luteinizing hormone), presence of azoospermia, determination of the sex chromatin, and prenatally via chorionic villus sampling or amniocentesis. A 2002 literature review of elective abortion rates found that approximately 58% of pregnancies in the United States with a diagnosis of Klinefelter syndrome were terminated.

3. A Chromosome Analysis, or Karyotype, Which is Usually Done on a Blood Sample. This Test Checks the Number of Chromosomes to See if the XXY Syndrome is Present: The standard diagnostic method is the analysis of the chromosomes' karyotype on lymphocytes. In the past, the observation of the Barr body was common practice as well. To confirm mosaicism, it is also possible to analyze the karyotype using dermal fibroblasts or testicular tissue.

Management:

There's no way to change the XXY condition if a boy is born with it, but treatments can help relieve some of the symptoms. As with many conditions, beginning treatment early can greatly increase its effectiveness.

Testosterone replacement therapy (TRT): Works by increasing a boy's testosterone levels into the normal range. Additional testosterone can help a boy with Klinefelter syndrome develop bigger muscles and a deeper voice, as well as promote growth of the penis and facial and body hair. It can also help improve bone density and reduce the growth of a boy's breasts. Testosterone therapy cannot increase the size of a boy's testicles or prevent or reverse infertility, however.

Educational Support Services Can help boys and teens with Klinefelter syndrome keep pace in school. Many benefit from extra assistance when it comes to schoolwork. If your son has Klinefelter syndrome, let his teachers and school nurse know about his condition and see what kind of support is available. He may be eligible for an individualized education plan (IEP) or 504 education plan, which both can provide accommodations for kids with special needs.

Speech Therapy and Physical Therapy Can help boys with Klinefelter syndrome learn to speak, read, and write better, or improve muscle strength and coordination.

Other Forms of Therapy Include Behavioral, Mental Health, and These can help improve low self-confidence, shyness, and delayed social development.

Often individuals that have noticeable breast tissue or hypogonadism experience depression and/or social anxiety because they are outside of social norms. An academic term for this is psychosocial morbidity. At least one study indicates that planned and timed support should be provided for young men with Klinefelter syndrome to ameliorate current poor psychosocial outcomes.

Surgical Management:

The surgical removal of the breasts may be considered for both the psychological reasons and to reduce the risk of breast cancer.

References:

1. Gropman A, Samango-Sprouse CA. 2013. Neurocognitive variance and neurological underpinnings of the X and Y chromosomal variations. Am J Med Genet Part C Semin Med Genet 163C: 35–43.
2. Greenstein, Robert M.; Harris, David J.; Luzzatti, Luigi; Cann, Howard M. (1970-04-01). "Cytogenetic Analysis of a Boy with the Xxxy Syndrome: Origin of the X-Chromosomes". Pediatrics. 45 (4): 677–686. ISSN 0031-4005. PMID 5438170.
3. Hultborn R, Hanson C, Köpf I, Verbiené I, Warnhammar E, Weimarck A; Hanson, C; Kopf, I; Verbiene, I; Warnha:mmar, E; Weimarck, A (November–December 1997). "Prevalence of Klinefelter syndrome in male breast cancer patients". Anticancer Res. 17(6D): 4293–7. PMID 9494523.
4. Graham JM, Bashir AS, Stark RE, Silbert A, Walzer S; Bashir, AS; Stark, RE; Silbert, A; Walzer, S (June 1988). "Oral and written language abilities of XXY boys: implications for anticipatory guidance". Pediatrics. 81 (6): 795–806. PMID 3368277.
5. Boone KB, Swerdloff RS, Miller BL, Geschwind DH, Razani J, Lee A, Gonzalo IG, Haddal A, Rankin K, Lu P, Paul L (May 2001). "Neuropsychological profiles of adults with Klinefelter syndrome". J Int Neuropsychol Soc. 7 (4): 446–56. PMID 11396547.
6. Gabriele R, Borghese M, Conte M, Egidi F (2002). "[Clinical-therapeutic features of gynecomastia]". G Chir (in Italian). 23 (6-7): 250–2. PMID 12422780.
7. Harold Chen. "Klinefelter Syndrome - Treatment". medscape.com. Retrieved4 September 2012.