International Journal of Pharmaceutical Research and Applications
Volume 7, Issue 3 May-June 2022, pp: 320-322 www.ijprajournal.com ISSN: 2456-4494

title:xxx

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Submitted: 05-05-2022 Accepted: 15-05-2022

ABSTRACT
Klinefelter syndrome is a group disorder in which there is an extra X chromose occur in male karyotype. Differentiation between normal child and KS child is very difficult in age below 7yr. 47,XXY aneuploidy is the most commonly seen in this disorder with a prevelance rate of one in 500 male child. Other sex chromosomal aneuploidy like 48,XXYY and 48,XXXY have also been described but they are much less frequent.Occurance of this syndrome is due to error in thechromosome non-disjunction during parenteral gametogenesis as a results a sperm or an egg carries extra X chromosome in addition to the normal single sex chromosome. School age child is associated with language delay, learning disabilities or behavioral problems, adults are often associated with low level of androgen, small testes, gynecomastia and many other symptom. Androgen replacement therapy is used to control or reduce the symptom, the most cruical time to initate this therapy is before puberty at the age of 10-11 years.
Keywords: Androgen, Azoospermia, chromosome, gynecomastia, hypogonadism, meiosis

I. INTRODUCTION

Klinefelter's syndrome is a genetic disorder/or chromosomal disorder in which a male individual born with an extra X chromosome which is abnormal when compared it to normal individual born with 23 pair of chromosome where XY chromosome is present but in klinefeltersyndromic person it is associated with (47, XXY) chromosome. This condition is also called as XXY syndrome. This type of abnormality is not detected during childhood because it is not possible to distinguish the normal child with hypogonadism child. Diagnosis of this type of condition is possible only during puberty or adolescence or the age of maturity. It is found that men with this type of syndrome is infertile by birth.It is found that klinefelter syndrome affect 1over 500 male people, it results in testicular failure, androgen deficiency, impaired spermatogenesis[(1)]. In chromosomal study of suspected individual, it is found that other extra chromosome is also present like 48,XXYY; 48,XXXY; and 49,XXXXY.

History of Klinefelter syndrome

This syndrome wad named after american endocrinologist Harry klinefelter. Klinefelter syndrome was firstly described when jacobes et al. recognised that syndrome occure due to the chromosomal disorder in which an extra X chromosome is present. Until 1959, it is thought that it can be occur due to endocrine disorder of unknown etiology. In 1942 Dr klinefelter recognized 9 male individual with symptom like gynecomastia, small testis, azoospermia and increased gonadotropin levels. They belived that failure in the sertoli cell of the testis cause hypogonadism. They also suggest that these patient had low or absent level of a putative second testicular hormone, which regulate the level of pitutary gonadotropins by feedback inhibition and which they labeled X hormone or inhibin[(2)].

Synonyms

Ks, 47,XXY, XXY Syndrome, XXY Triosomy, 3+X Chromosome with Y, Triosomic disorder.

Epidemiology

Klinefelter syndrome is the most common chromosomal disorder seen only in male with an estimated frequency of 1:500- 1:1000 male child birth[(3)]. It is characterized by X chromosome polysomy with X disomy, triosomy. 47,XXY is the most common varient[(4)]. Almost all men with a 47,XXY karyotype will be infertile;klinefelter syndrome posses 3 percent of male infertility[(5)]

Sign & Symptom

Oligospermia and azoospermia are most commonly in klinefelter syndrome infertile patient[(6)]. Affected men also have small testes, decreased facial hair, gynecomastia, decreased pubic hair, small penis, Tall, slender body habitus with long bones and shorter torso, motor delay and dysfunction, speech and language difficulty, attention difficults, learning disabilities, Dyslexia, Psychosocial or behavioral problems.

Androgen deficiency cause loss of libido, decreased muscle bulk and tone, decreased bone mineral density etc[(7)].

Etiology

The extra chromosome found in klienfelter syndrome is mainly occur due to an error of non-

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disjunction during parenteral gametogenesis as a results a sperm or an egg carries extra X chromosome in addition to the normal single sex chromosome[8]. It also may results due to error in the division during mitosis in the zygote[9]. A study using DNA probes found that paternal non-disjunction posses 53% of case, maternal meioticI error posses 34.5% of case, maternal meiotic II error posses 9.5% of case and postzygotic mitotic error posses only 3% case[10]. There is a positive coorelation between maternal age and maternal meiotic I error. The error in the gametogenesis of both maternal and paternal is equally responsible for the causing the 47,XXY disorder[11].

Diagnosis

Testicular biopsy specimen of infants with this disorder describe only a reduced number of germ cell. After the puberty pathologic changes like hyalinization, fibrosis of the seminiferous tubules small shrunken testes and azoospermia are major characteristics[12]. There is also decreased in level of inhibin B observed due to loss of functional seminiferous tubule and sertoli cell. It is the hormone which regulate the level of FSH[13]. The presence of increased serum LH level despite low-normal testosterone level indicate that patients with KS have an altered hypothalamic pitutary-gonadal axis[14].

Treatment

There are no cure for KS but most of the problem arised like decreased libido, weakness, decreased bone density, and androgen deficiency can be reduced by the use of testosterone replacement therapy.

Testosterone replacement therapy is used in this syndrome, despite of it has no effect on infertility but it correct the androgen deficiency[15]. This therapy results in increase in facial and pubic hair, a more masculine distribution of body fat, increased libido, and bone mineral density[16]. At the time of begining of puberty is the optimal time for the initiation of testosterone replacement therapy because this timing boys with KS experience a pubertak changes similar to their peers and allows testosterone to have it most marked effect on bone mineral density[17]. This treatment therap has no effect on testicular size or spermatogenesis,and it usually does not reduce gynecomastia, which can be very well treated with plastic surgery.[18]

II. CONCLUSION

Klinefelter syndrome is a an extra X chromosomal syndrome in which a male child posses an extra X chromosome which may leads to differentiation among normal child after the puberty. Better understanding of inactivation of X chromosome, regulation of meiosis as well as pathophysiology related to spermatogonia allow the researcher to develop better therapeutic management approach for this syndrome.

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