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CASE REPORT

Clinical & Cytological Study on Klinefelter Syndrome

Siddanagouda M Biradar[1], Rudragouda S Bulagouda [2], Gurushantappa Kadakol[3]

Abstract

Klinefelter Syndrome is a chromosomal disorder with addition of X sex chromosome (47XXY) in males. A patient attended to our hospital with loss of secondary sexual characteristics and male infertility. Physical examination revealed thin built, hypogonadism and absence of pubic hairs. Karyotype and biochemical analysis were performed to detect chromosomal abnormality as well as hormonal level to confirm the diagnosis of Klinefelter syndrome.

Key Words

Klinefelter Syndrome, 47 XXY, Gynecomastia

Introduction

Klinefelter Syndrome is chromosomal disorder with presence of two or more X Chromosomes in a male. The clinical phenotype of KS was first described in 1959 in males with tall stature, small testes and gynaecomastia with the genetic etiology of extra X chromosomes. The incidence of KS (ranging from 0.1 to 0.2 % in newborn male infants) rises up to 3-4 % among infertile males and 10-12 % in azoospermic patients. [1, 2]

Hypogonadism, gynecomastia, and infertility are common symptoms that lead to the diagnostic evaluation of males for Klinefelter syndrome. Lacking in clinical care does exist as there are gaps in diagnosis, lack of standardization of care and access to treatments are not always affordable in India. [3, 4] We report such rare case with KS in our hospital Vijayapur District Karnataka.

Case Report

A 20-year-old married male was brought to our superspeciality Centre, Sri B M Patil Medical College and Research Institute, Vijayapur Karnataka, with the history of generalized weakness since five days along with pain in the left lower part of thigh. He was born of non-consanguineous parents. He was neither on any medications nor did he have history of diabetes/ hypertension. There was no family history of same. He had in past recurrent fractures of long bones of leg (*Fig 1*).On physical examination, this male had a height of 155 cm, weight of 58kg with gynaecomastia, wide gap in toes, gynecomastia, female pattern of hair line with grey colored hair, puffiness of face (Cheek) & pot belly (abdomen) with pigmentation & straie over the abdomen with testicular volume bilaterally 4 to 5ml, firm consistency, non tender. Also on examination left leg deformity with swelling was present (*Fig 2*).

These findings were suggestive of testosterone deficiency syndrome with male infertility. This patient was referred for karyotype and relevant biochemical tests

Department of Medicine[1], Anatomy[2], Human Genetics[3], BLDE (DU) Shri B M Patil Medical College Hospital & Research Centre Vijayapur

Correspondence to:Dr G S Kadakol, Deptt of Anatomy,Genetics Laboratory, BLDE (DU) Shri B M Patil Medical College Hospital & Research Centre Vijayapur

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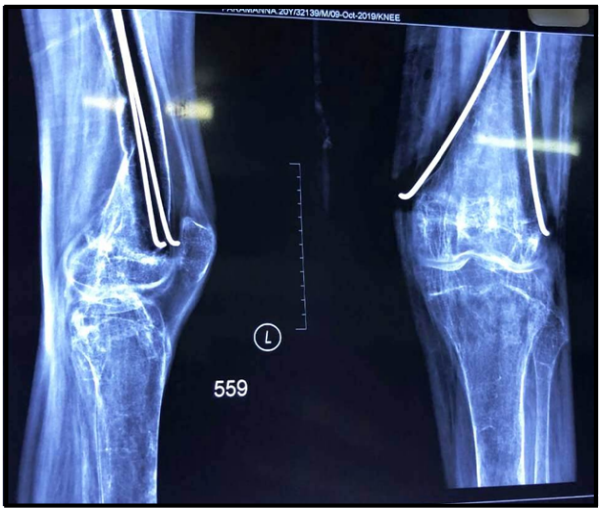

Fig. 1 Fractures of Long Bones of Leg

Fig.2 left Leg Deformity with Swelling

to confirm the diagnosis and know the cause of this infertility syndrome.

Serum testosterone levels were very low [0.05 ng/ml] (Reference range for adult males more than 19years of age: 0.05-13.50 ng/ml). Serum ultra-sensitive TSH levels were 0.75 ng/ml (Reference range: 0.40-4.00ng/ml). Karyotype result showed 47, XXY chromosome complement (*Fig 3*) in all screened metaphase spreads. The subject was advised genetic counseling by the genetic counselor thereafter with androgen replacement and neuropsychological / adaptive therapies.

Discussion

Kinefelter'syndrome classically, it is characterized by very small firm testis, azoospermia and infertility with uniformly elevated gonadotropis levels.

In infertility sex chromosomal abnormalities are common. Klinefelter Syndrome is a rare chromosomal disorder with addition of one X chromosomes (47 XXY) seen in males. Extra X chromosomes lead to testicular hyalinization, fibrosis and testicular hypo function which may result in genital abnormalities, and usually hypogonadism and infertility. Incident rate of KS is 1: 600 in male births, approximately 64% of which remain undiagnosed throughout life. [5]

Although the damage to testis initiated in utera in patients with klinefelter syndrome, there is accelerated damage to the testis during mid puberty. The cause for accelerated testicular damage during mid puberty is not clear. However, the rise in levels of gonadotropis and alteration in intratesticular testosterone to strong in ratio have been implicated. Hence, our patient has two children from his early marriage but has now become infertile

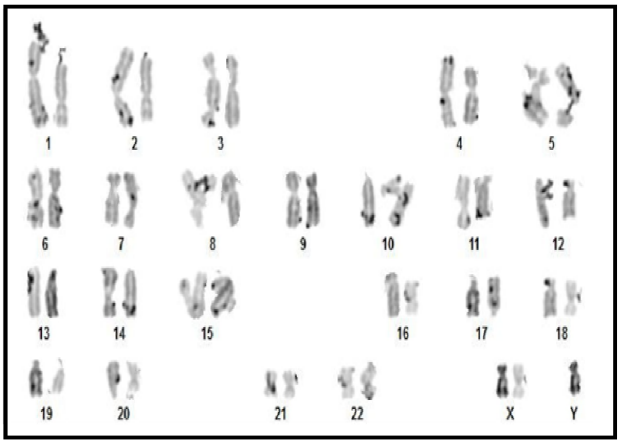

Fig 3. Karyotype result showing 47, XXY chromosome complement

due to accelerated testicular damage. Gynecomastia is seen in adult men, associated with psychological distress. Adolescents with gynecomastia are frequently affected emotionally and psychologically, regardless of graded severity of disease. [6] There are increased feelings of loneliness, restlessness, and tension. There has been a higher association of depression, anxiety, adjustment disorders, low self-esteem, and suicidal ideation. [7]

It is extremely rare to encounter acne in individuals with hypergonadotropic hy pogonadism typical of KFS as in our case. [8] Some individuals have the extra chromosome in only few cells and are described as mosaic KFS. Mosaicism results in clinical variations in KFS as some of the cells have normal karyotype.

Klinefelter syndrome cases were confirmed by cytogenetic analysis using GTG-banding technique revealed the karyotype 47, XXY. Peripheral blood is used for karyotyping without harming the fetus the frequency of prenatal diagnosis of KS is expected to increase worldwide, resulting in early diagnosis and management.[9] KFS mosaic 46, XY/47, XXY is also not inherited. It occurs as a random event during cell division early in fetal development. As a result, some of the body's cells have one X chromosome and one Y chromosome (46, XY), and other cells have an extra copy of the X chromosome (47, XXY). Such individuals would manifest acne and also would be able to father children following successful sperm retrieval. [10]

Conclusion

Klinefelter syndrome is a rare chromosomal disorder with addition of one X chromosome. KS is also with insulin resistance such as type-2 diabetes, dyslipidemia, poor bone mineral density as well as peripheral vascular disease, thromboembolic disease. KFS was diagnosed only after 18 years of age in two-thirds of patients. Adults predominantly presented with hypogonadism. In our study, we observed the infertile male with 47, XXY karyotype. For mosaicism, further advised for FISH/ chromosomal microarray/ molecular studies but he is not able to go for all the tests due to high cost.

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Nil.

Conflicts of Interest

There are no conflicts of interest.

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