

Karyotype revealed 47, xxy chromosome (klinefelter syndrome): a case report

Chandra Bhushan Jha,¹ Shaligram Dhungel¹ and Dilip Rai²

Department of Anatomy ¹BP Koirala Institute of Health Sciences, Dharan, Nepal and ²Nepal Medical College Jorpati, Kathmandu, Nepal

Correspondence author: Dr. Chandra Bhushan Jha, Additional Professor, Department of Anatomy
B P Koirala Institute of Health Sciences, Dharan, Nepal, e-mail: chandraj_2000@yahoo.com

ABSTRACT

The Klinefelter syndrome is most common chromosomal cause of male infertility. However, the many cases of the syndrome remain undiagnosed due to variations in clinical presentation. A patient attended to surgical OPD with complaints of loss of secondary sexual characteristics and infertility. Physical examination revealed tall stature, thin built, small testes size, and absence of beard and pubic hairs. Karyotype and biochemical tests were performed to detect chromosomal abnormality as well hormonal level to confirm the diagnosis of androgen deficiency syndrome. Chromosomal complement confirmed the case of Klinefelter syndrome (47, XXY) causing androgen deficiency. Timely detection of Klinefelter syndrome is important to formulate further treatment modalities for the benefit of the patient.

Keywords: Hypogonadism, infertility, Klinefelter syndrome.

Klinefelter syndrome, the most common cause of male infertility is first described in 1942. The syndrome is usually detected during the development of secondary sexual characteristics. In adulthood, small firm testes and symptoms of androgen deficiency associated with azoospermia, tall stature, and bilateral gynaecomastia characterize it.¹ However, the many cases of this syndrome remain undiagnosed due to substantial variations in clinical presentation and insufficient professional awareness of the syndrome. Early diagnosis and androgen replacement treatment of the disorder can improve quality of life.²

Prevalence of the syndrome is 1: 500 among males. Barr bodies examination, a simple diagnostic method, was used for rapid screening in past in suspected Klinefelter syndrome.³ However, the diagnostic accuracy of this screening test has never been evaluated and cannot stand alone as a diagnostic tool. Nowadays Karyotype is the definite test for the syndrome in which sex chromosome aneuploidy or structurally abnormal X chromosome is present.⁴ This case of Klinefelter syndrome is being reported considering its importance in the diagnosis and management of male infertility.

CASE

A 24-year young married male attended the surgical OPD with complaint of loss of moustache, beard and pubic hairs, infertility, and decreased libido of two-year duration. Physical examination revealed tall (6 ft.) and thin general body texture. Both testes were small and secondary sexual hairs were absent (fig.1). These findings were suggestive of androgen deficiency syndrome and the patient was referred for karyotype and biochemical tests to confirm the diagnosis. Follicular stimulating (FSH) and luteinizing (LH) hormones were found to be elevated [FSH: 25.99 mIU/ml (Reference range: 1.0-15.0 mIU/ml), LH 14.56 mIU/ml (Reference range: 0.7-7.4 mIU/ml) respectively]. Estradiol was estimated to be 15.0 pg/ml (Reference range: 12-34 pg/ml). However, testosterone was estimated to be 0.49 nmol/L below the normal values (Reference range: 0.52-38.17 nmol/ml).⁵

Peripheral blood lymphocyte culture was done and karyotype was prepared by using standard protocol (ISCN, 1995).⁶ Karyotype showed 47, XXY chromosome complement (fig. 2) in all screened metaphase spreads. The diagnosis of Klinefelter syndrome has been confirmed and this patient was referred to an endocrinologist for further management.

DISCUSSION

Small firm testes and symptoms of androgen deficiency are the characteristic features of the Klinefelter syndrome and it is the most frequent form of hypogonadism in the male population first described in 1942.⁷ In this case, small testes, infertility, decreased libido, loss of moustache, beard, and pubic hairs were the main symptoms. These features were suggestive of hypogonadism. The endocrine profile showed androgen deficiency. Hypogonadism associated with androgen deficiency directed us towards Klinefelter syndrome for

which chromosome analysis is a definite diagnostic test.⁸ Karyotype showed 47, XXY chromosome complement and proved the case as Klinefelter syndrome.

The patient was referred to an endocrinologist for further evaluation and testosterone replacement therapy for hormonal treatment. It can improve the quality of life and correct the symptoms of androgen deficiency.⁹ Intra-cytoplasmic sperm injection (ICSI) is another mode of treatment of Klinefelter syndrome patients which provides them an opportunity to father a child.¹⁰ However, the patient has to be counseled about concern of chromosomal normality of an embryo generated through ICSI.

Klinefelter syndrome should be confirmed by karyotyping which is being the main cause of male infertility. Timely detection of Klinefelter syndrome is important to formulate further treatment modalities for the benefit of the patient.

REFERENCES

1. Kamischke A, Baumgardt A, Horst J, Nieschlag E. Clinical and diagnostic features of Patients with suspected Klinefelter syndrome. *J Androl* 2003; 24: 41-7.
2. Amory JK, Anabalt BD, Paulsen CA, Bremner WJ. Klinefelter's syndrome. *Lancet* 2000; 356: 333-5.
3. Moore KL, Barr ML. Smears from the oral mucosa in the detection of chromosomal sex. *Lancet* 1955; 269: 57-8.
4. Jacobs PA, Strong JA. A case of human intersexuality having a possible XXY sex determining mechanism. *Nature* 1959; 183: 302-3.
5. Robert KM, Daryl KG, Peter AM, Victor WR. Harper's Biochemistry. 24th Edition, 1996: 833.
6. Mitelman F. Ed. ISCN 1995: An International System for Human Cytogenetic Nomenclature (1995). Karger 1995.
7. Klinefelter HF, Reifenstein EC, Albright F. Syndrome characterized by gynaecomastia, aspermatogenesis without Leydigism, increased excretion of follicle stimulating hormone. *J Clin Endocrinol* 1942: 615-27.
8. Smyth CM and Bremner WJ. Klinefelter syndrome. *Arch Intern Med*.1998; 158: 1309-14.
9. Lanfranco F, Kamischke A, Zitzmann M, Neischlag E. Klinefelter's syndrome. *Lancet* 2004; 364: 273-83.
10. Staessen C, Tournave H, Van Assche E *et al*. PGD in 47,XXY Klinefelter's syndrome patients. *Hum Reprod Update* 2003; 9: 319-30.



Fig.1. Tall male without secondary sexual hairs



Fig. 2. Metaphase spread and karyotype showing 47, XXY chromosome.