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A Case of Klinefelter Syndrome Presenting with Infertility and Gynaecomastia

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ABSTRACT

The term Klinefelters syndrome refers to a group of chromosomal disorders in which the normal male karyotype 46XY has at least one extra X chromosome. The 47XXY aneuploidy is the most common human male sex chromosomal disorder associated with infertility and hypogonadism .It has prevalence of 1 in 500 to 1 in 800 live births independent of race. The aim of this case report is to document the rare case in the literature with the view of raising awareness about its existence in our environment and beyond.

The Case: We present a case of a 31 year old black male patient with a 7 years history of progressive bilateral painless breast swelling that has rapidly increased in size over the past 3 years. He has no associated breast discharge. He also complained of associated infertility since he married his wife about 7 years ago despite regular unprotected intercourse within the same period. Hormonal assays done showed elevated gonadotropins(Leuteinising hormone and follicle stimulating hormone) , low levels of testosterone and elevated estradiol. Karyotype done revealed 47XXY karyotype.he was couselled on the need for assisted reproductive therapy, which he accepted. Subsequently, his wife was delivered of a baby boy following a successful assisted reproductive therapy by an obstetrician.

Conclusion: Klinefelter syndrome is a rare clinical syndrome but a common chromosomal cause of male infertility. The introduction of assisted reproductive therapy has greatly improved infertility outcomes in terms of achieving pregnancy and live births.

Keywords: Klinefelter syndrome, infertilty, gynaecomastia, North central Nigeria.

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INTRODUCTION.

Klinefelter syndrome (KS) was first reported in 1942 by klinefelter et al[1] when he described nine men with the following features; testicular dysgenesis, micro-orchidism, eunoichidism, gynaecomastia, elevated urinary gonadotropins and azoospermia. It was thought then to be due to an endocrine disorder of unknown origin. However, in 1959, Jacob et al [2] recognized it as a chromosomal disorder in which there is an extra X chromosome resulting in Karyotype 47XXY. The syndrome stems from the acquisition of an additional X chromosome, a product of a nondisjunction event during meiosis. This is not taught to be hereditary.[3]

Klinefelter syndrome occurs in roughly 1:500 to 1:800 live births independent of race [4]. Male patients with KS have low testosterone production which leads to abnormal body built. Phenotypically adult males will be tall, with reduced muscle mass, a sparse body and facial hair, gynaecomastia, weaker bones with broad hips (gynaecoid pelvis). The limited testosterone during puberty results in small penis, testes and prostate gland [5].Additional features include the increasing levels of follicle stimulating hormone(FSH) and Luteinising hormone(LH) with increasing age which are known to be responsible for hyalinisation and fibrosis of the seminiferous tubules [6]. Most cases (>60%) are diagnosed post puberty when symptoms are most obvious [7]. Men with KS often exhibit sexual dysfunction such as impotence, lack of libido and usually produce little or no sperms, with 95% being classified as infertile [8]. Early identification and medical intervention of 47XXY male can greatly improve physical, mental and emotional problems associated with the syndrome.

CASE.

We present a case of a 31 year old black male with a seven years history of progressive bilateral painless breast swelling, which has rapidly increased over the past 3 years. No associated history of galactorrhoea. He complains of associated decrease libido with weak erection, primary infertility and inability of his wife to conceive despite regular unprotected sex in the past 7 years of marriage. No history of trauma to the testes, use of cytotoxic drugs, alcohol or cigarette smoking. History of other drug use was also non contributory.

Examination revealed bilateral non tender gynaecomastia figure 1. Enuchoidism (arm-span187cm -height- 178cm), broad hips (gynaecoid pelvis) And a global decrease in muscle mass.

Hormonal assay revealed levels consistent with hypergonadotropichypogonadism (serum testosterone(8.53nmol/L), Luteinising hormone(40.3I.U/L) and follicle stimulating hormone(37.23I.U/L), estradiol levels was also high (211 pmol/L). Karyotype studies done revealed 47XXY karyotype.

An assessment of Klinefelter syndrome was made based on the above findings. Patient was counseled for assisted reproductive therapy; which he consented to, subsequently his wife was delivered of a baby following a successful assisted reproductive therapy.

DISCUSSION.

Klinefelters syndrome is a rare clinical syndrome but a common genetic cause of male infertility. Our patient, a 31 year old presented with a 7 years history of infertility, gynaecomastia, and a hormonal profile in keeping with hypergonadotropichypogonadism and a karyotype of 47XXY. This patient also seems to have low intellectual capacity as he had only primary level of education a feature also seen in some patients with Klinefelter syndrome. His presenting age of 31 years is in keeping with the fact that most patients with Klinefelters syndrome become increasingly symptomatic after their pubertal years[7].

Klinefelters syndrome is rare in our environment; Sabir et al[9] reported a case of a 40 year old whose presenting feature was gynaecomastia in Sokoto, Northern Nigeria. The finding of hypergonadotropichypogonadism

seen in our this case was also reported by Smyth et al [10] and Pinyerd et al [11] who in a review, reported that 95% of patient with KS had elevated gonadotropins, infertility and 75% of them had decrease testosterone level.

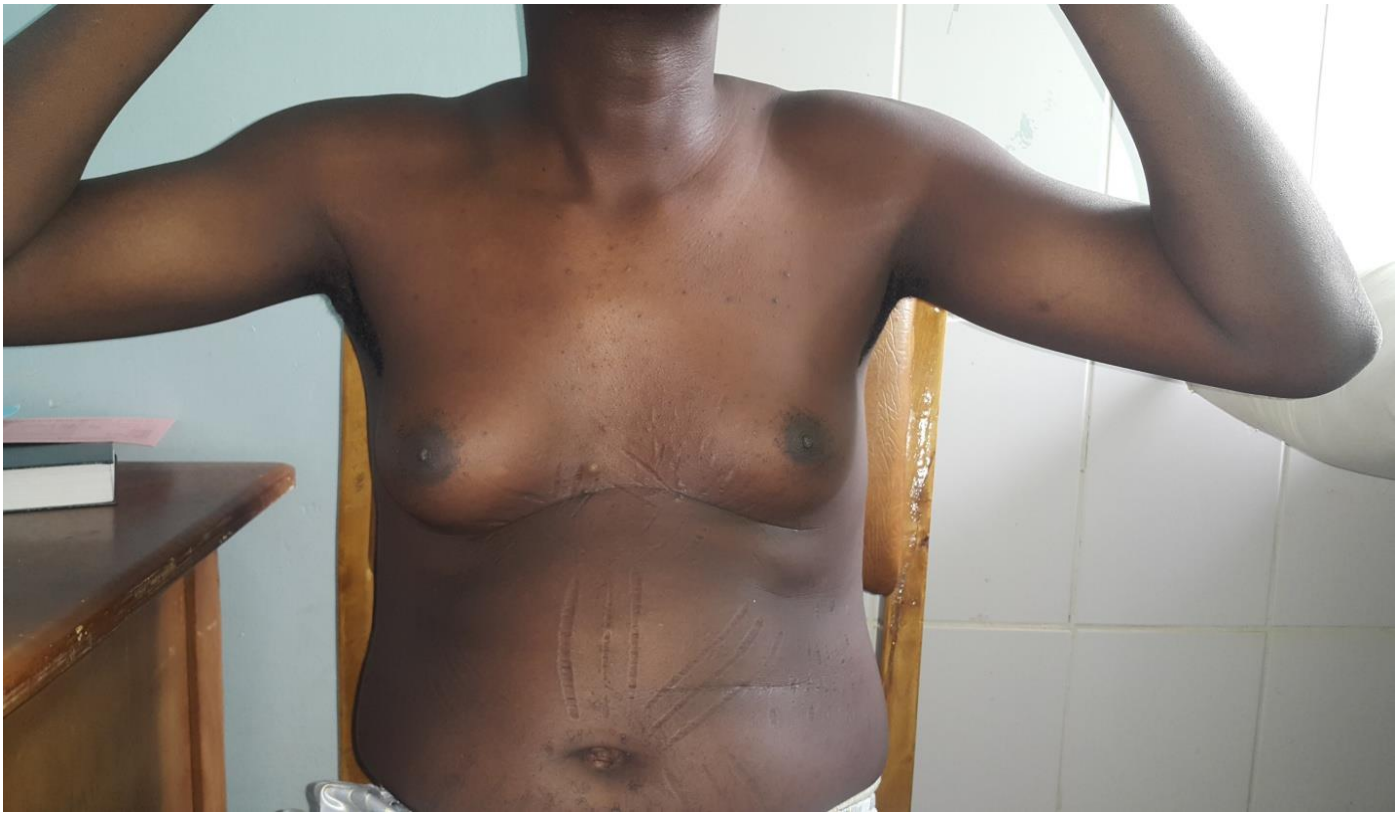

Figure 1 Showing bilateral Gynaecomastia

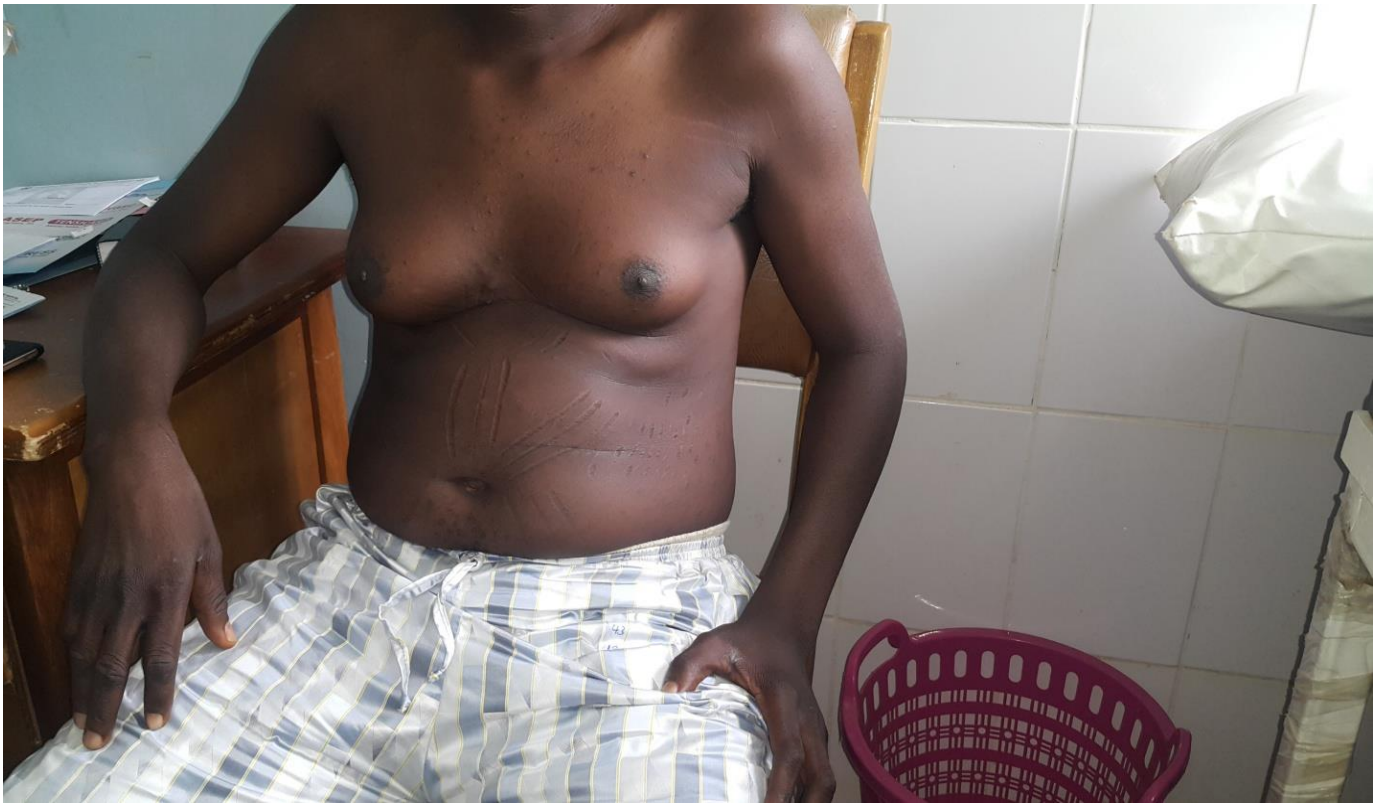

Figure 2 Same patient with muscle wasting,gynaecoid pelvis and gynaecomastia

Infertility was a major presenting feature of our patient. This is consistent with a previous report which indicated that 95% of patients with KS are infertile [6]. Before the introduction of Intracytoplasmic sperm Injection (ICSI) technique, the outcome of infertility in terms of achieving pregnancy and life birth were near absent. However, with the advent of ICSI, pregnancies and life birth have been reported by several sources[12,13,14] .

CONCLUSION.

Klinefelters syndrome is a rare clinical syndrome but a common genetic cause of male infertility.

Early identification and treatment with the use of testosterone especially during puberty can tremendously improve physical, mental and emotional problems associated with the syndrome.

Patients presenting with infertility should be referred early to specialised gynaecologic centres that offer assisted reproductive therapy. This is because outcomes in terms of achieving pregnancy and live births are promising.

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