



An Adolescent with Klinefelter's Syndrome

Eleje George Uchenna*

Department of Obstetrics and Gynecology, Nnamdi Azikiwe University, Awka, Nigeria

*Corresponding author: Uchenna EG, Department of Obstetrics and Gynecology, Nnamdi Azikiwe University, Awka, Nigeria, E-mail: georgel21@yahoo.com

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Abstract

Klinefelter's syndrome is a common sex chromosomal defect in humans that continues to be a major genetic cause of infertility in men. A 12-year-old student was referred to our hospital because he had small penis and testes from birth, as well as a five-year history of gradual breast augmentation. The height was 1.64 meters and the weight was 75 kilograms, according to the examination. Tanner stage IV and III had well-developed breast and axillary hairs, respectively, while Tanner stage IV had a well-developed hair distribution of external genitalia. The testicles and penis were both tiny but nicely developed. Hypospadias and epispadias were not present.

There were no ovaries or uterus visible on transperineal and trans rectal ultrasounds. Except for a low level of Follicle Stimulating Hormone (FSH), the hormonal profile was normal. Because of the gynecomastia, the parent was referred to a general surgeon for a mastectomy. In our gynaecological clinic, we reported the first occurrence of Klinefelter's syndrome in an adolescent. The numerous types of gynecomastia differential diagnosis were discussed.

Keywords: Klinefelter's syndrome; Hypogonadism; Gynecomastia; Mastectomy

Introduction

In males, Klinefelter's syndrome is a common hereditary cause of infertility [1]. The first instance presented in our gynecological clinic is described. The wide range of physical anomalies, all of which are very minor, may explain why many patients do not seek medical attention until they are adults, when they seek guidance on tiny testes or infertility. The syndrome's low awareness among health professionals may also make diagnosis difficult [2]. The importance of early diagnosis and treatment for improving quality of life and avoiding serious complications cannot be overstated.

Case History

A 12-year-old junior secondary school student was sent to our hospital due to the presence of small penis and testes from birth, as well as a five-year history of gradual breast augmentation. There was

no history of testis descent delay, testis damage, or mump orchids. The patient had had an erection early in the morning, but no sexual activity. Both of the patient's female siblings are alive and healthy. There was no history of a similar condition in the family.

The increase in breast size began at the age of seven and has continued to progress since then. Breast injuries, discomfort, or nipple discharge were not present. The sufferer had been brought up to be a man. The patient had never been admitted to the hospital before and was not diabetic. The mother had no previous history of hormonal medication intake or concussion during pregnancy. Pregnancy, labor, delivery, and the neonatal period went without a hitch. The patient was well immunized and had a normal milestone development. Academically, the patient performed admirably.

Afebrile, anicteric, pallid, and acyanosed, the adolescent was found to be in no apparent distress. There were no palpable lymph nodes in the patient. The person was 1.64 meters tall and weighed 75 kilograms. At Tanner stage III, the axillary hairs were well grown. Tanner stage IV breasts were symmetrical and fully developed on both sides. The nipples were turned inside out. On the breast, there was no pain or palpable lump. The abdomen revealed nothing unusual.

The external genitalia revealed female hair distribution at Tanner stage IV. The penis was well developed with stretched length of two centimeters. There was no hypospadias or epispadias. The patient was circumcised. The testis is measured 1.0 cm to 0.5 cm.

A diagnosis of intersex disorder secondary to Klinefelter's syndrome was made. The packed cell volume was 36%. The urinalysis was normal. The abdominal ultrasound showed right and left testes with homogenous texture and measured 10 mm by 0.8 mm and 11 mm by 7.5 mm respectively. Transperineal and transanal ultrasound showed no ovaries or uterus. The hormone profiles were within normal range except for follicle stimulating hormone that was less than 1.0 mIU/ml (2 mIU/ml-10 mIU/ml).

Buccal smear showed chromatin positive with numerous regular squamous cells with less than 10% showing nuclear bar bodies. The patient and parents were counseled on the diagnosis and they desired to continue to rear the child as male. The problem of sterility was discussed with parents and they consented to mastectomy. He was later referred to the general surgeon for mastectomy. The patient was lost to follow-up.

Discussion

As in the present case report, prepubertal gynecomastia is rare entity and a specific cause is hardly ever identified such that in 90% of patients, it is classified as idiopathic [3]. However, known causes of breast enlargement in children are diverse [4-6]. Therefore, further exploration of the etiology in children is often warranted, particularly to rule out any endocrine or malignant abnormalities. In pre-pubertal period, the following could be responsible, administration of estrogens or estrogenic compounds (androgens or other substrates for aromatase, clomiphene, phytoestrogens, enoestrogens), administration of non-estrogenic drugs (digoxin, high), testicular or adrenal tumors, Human Chorionic Gonadotrophins (HCG) secreting tumors, aromatase excess syndrome (familial hyperestrogenism), central or peripheral precocious puberty, mammary tumors of different cell composition (usually unilateral), and idiopathic gynecomastia [7].

A variety of endocrinopathies, mostly as a result of an increased ratio of circulating estrogens to androgens, induce stimulation of breast tissue leading to gynecomastia. Calzada et al. showed that the presence of hormone receptors in gynecomastia may provide a setting favorable for mammary glands to develop gynecomastia [8].

In pubertal gynecomastia, the following are differential diagnosis. These include: Physiologic gynecomastia of adolescence, feminizing tumors, drugs, familiar gynecomastia, primary hypergonadotropic gonadal dysfunction, Klinefelter's syndrome (47, variants), males, androgen insensitivity syndrome (with ambiguous external genitalia), defects in testosterone biosynthesis (with ambiguous external genitalia), true hermaphroditism (with ambiguous external genitalia). The other causes include hypergonadotrophic hypogonadism (infections, chemotherapy external radiation), secondary hypogonadotrophic gonadal dysfunction, hyperthyroidism, hepatic damage and idiopathic causes.

In 1998, Sher et al. analyzed the etiologies of 60 adolescent boys with gynecomastia greater than 4 cm in diameter, aged 10 years-20 years of age. Endocrine anomalies were detected in 7 subjects, Klinefelters syndrome, XX male, primary testicular failure, and hepatocarcinoma. Different pathological processes were also diagnosed. In another 8 patients, and 45 subjects were labeled as idiopathic.

Although the presence of low FSH may appear that the gynecomastia seen in the present report may be pre-pubertal in type but we strongly believe that this is pubertal gynecomastia. This is because the increase in breast size was noticed at about seven years of age which is the time of onset of puberty and has remained progressive since then. Additionally, axillary hairs were well developed at Tanner stage III and hair distribution of external genitalia was at Tanner stage IV. Klinefelter's syndrome is a well-recognized, yet rare occurring clinical entity. It is the most common male chromosomal disorder associated with hypogonadism [9,10]. Klinefelter's syndrome does not have any racial predilection. Because the syndrome is caused by an additional X chromosome on an XY background, this condition affects only the males. It may go undiagnosed in most affected males. However, among males with known Klinefelter's syndrome, many do not receive the diagnosis until they are adults as was seen in our patient. Although some readers and gynecologists may view this differently, the case under consideration is a typical of Klinefelter's syndrome. This is because; it associates strongly with hyper gonadotrophic hypogonadism, small testes with azoospermia and intellectual deficit. Behavioral and school difficulties are frequent. Since gonadal dysfunction becomes evident at puberty, diagnosis is seldom made before adolescence.

Moreover, sometimes, signs are mild and diagnosis is not made, even in adults. Unilateral or bilateral cryptorchidism is frequent among boys with Klinefelter syndrome. Puberty onset is usually at the right age, with pubic hair and genital development but testis remain small and of higher consistency. Hypogonadism or micro penis might be present. Gynecomastia is frequent. Even though serum testosterone might reach normally low values, serum estradiol is relatively high, and it is assumed that an abnormal estradiol testosterone ratio favors mammary development seen in the syndrome. Seminiferous tubules deterioration is progressive, ending in hyalinization and a further decrease in serum testosterone. In these conditions, replacement testosterone treatment improves hypogonadism, but not infertility. When gynecomastia disturbs patient's everyday life, surgical removal of the breasts is indicated.

Klinefelters syndrome is characterized by gynecomastia, hypogonadism, infertility and psychological problems [11-13]. The other symptoms include fatigue, body weakness, erectile dysfunction, osteoporosis, language impairment, academic difficulty, poor self-esteem, subnormal libido, and behavioral problems. The latter symptoms were not observed in our patient. Children may be anxious, immature, excessively shy, aggressive, may engage in antisocial acts. Early diagnosis improves patient's quality of life and enables better medical treatment. To achieve this, it is crucial to increase both medical and general awareness of the disease, including through use of the mass media as well as the associations of the patients.

As was seen in the index case, the development of gynecomastia and eunuchoidism could be one of the most common signs leading boys and their parents to consult a doctor. This is caused by imbalance in the normal circulating levels of testosterone and a normal estrogen-testosterone ratio eunuchoidism. Gynecomastia may also be caused by an imbalance between estrogen receptors and androgen receptors, leading to excessive estrogen action, deficient androgen action or a combination of these effects.

Conclusion

We reported a first purported case of a Klinefelter's syndrome occurring in an adolescent in our environment. Although reports regarding the rare entity of Klinefelter's syndrome have been published, reviews of literature and the clinical implications of this case are discussed including potential differential diagnosis for prepubertal and pubertal gynecomastia.

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