CASE REPORTS

A Klinefelter’s female?!

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Introduction

Human sex development is the result of a complex interaction of an individual’s genes, hormones and the environment. Disorders of sex development (DSD) or intersex disorders occur when an individual’s sex development takes a different path from the typical female or male [1].

Case Report

A 24 year old female netball player noticed a painless, left inguinal lump with a cough impulse which was constant in size for 2 months. It was initially diagnosed as an inguinal hernia and underwent exploration which revealed an undescended testis with an indirect sac. Orchidopexy was done with indirect sac transfixation and further investigation was prompted.

She has a brother who is married with children whilst her sister is infertile. She is phenotypically female with normal small-sized breasts, tall stature and a normal vulva (Figure 1) with clitoromegaly (Figure 2).

Ultrasonography revealed bilateral inguinal testes and a blind-ended vagina with absent uterus and ovaries. Her genetic assessment revealed 47, XXY Klinefelter syndrome, and androgen insensitivity syndrome (AIS) was suspected by hormone assay. Her testosterone level was 43.061 nmol/l, well above the male level. LH (36.1 U/L) was normal whilst the FSH (51.6 U/L) was above the female range in the luteal phase. The DEXA scan was normal.

After counselling, she wished to continue as a female and hormone replacement was commenced. Plastic surgical gender reassignment was done by reducing the rudimentary phallus whilst preserving the dorsal neurovascular bundle of the penis and reconstructing a normal sized clitoris and labia minora (Figure 3 & 4). Inguinal testes were excised and sent for histology. The presence of Leydig cell hyperplasia on testicular histology supported the diagnosis of AIS.

Discussion

Klinefelter syndrome is the most common chromosomal disorder in men, affecting about 1-2/1000 men; usually due to non-disjunction of sex chromosome. Males with Klinefelter syndrome are commonly infertile since only a single normal Y chromosome is present. However, a few cases of female Klinefelter syndrome are reported.

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syndrome may demonstrate: small firm testes, a small penis, sparse pubic, armpit and facial hair, enlarged breasts, tall stature, and abnormal body proportions [1,6,7].

Androgen insensitivity syndrome (AIS) is x-linked recessive, causing failure of normal masculinization of external genitalia in chromosomally normal males. The basic etiology is due to mutations in the androgen receptor (AR) gene of the X chromosome. Thus, they look like females with female external genitalia, but do not possess ovaries, an uterus or an upper vagina. Their testosterone level is either normal or elevated [1,8,9].

Our patient was phenotypically female, with narrow shoulders, sparse pubic, armpit and facial hair, enlarged breasts (gynaecomastia) and tall stature. As Klinefelter syndrome presents as males, her female presentation raised the suspicion of co-existant AIS. Her grade 3 external genitalia, absent uterus, fallopian tubes and ovaries with undescended testes favoured AIS. In contrast, those with Klinefelter syndrome have malfunctioning testes in the normal position. Her DEXA scan was normal due to high testosterone in contrast with Klinefelter syndrome patients who have low testosterone levels.

A typical patient with Klinefelter syndrome presents with low serum testosterone, high LH and FSH, and often elevated estradiol [2,3,6,7]. However in this patient, pre-operative testosterone levels were high likely due to coexisting AIS. Following an orchidectomy, her testosterone level dropped to 0.34nmol/l.

**Conclusion**

Although our patient is 47, XXY (Klinefelter Syndrome), she is a phenotypic female due to the co-existance of AIS. Further genetic studies are needed to identify the presence of a uniparental disomy of the X chromosome leading to a replication of the mutation on the androgen receptor gene [10].

**References**


**Key Point:**

- As the prevalence of Klinefelter syndrome is considerably high it should be actively sought when investigating subfertility.
- With the development and availability of endocrinology, plastic, reconstructive and microsurgical facilities in Sri Lanka, the complete management of such a scenario is possible.