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...
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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.