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Case Report

Nonclassical XX male sex reversal syndrome presenting as male infertility

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Introduction

Intersexuality is defined as a condition of imperfect sexual differentiation, into either male or female. Sexual and physical development of an individual depends on genetic, gonadal, hormonal factors, response to the end organs and psychological factors. SRY gene is a testis determining gene present on Y chromosome. In rare instances phenotypic males can have 46XX karyotype as in XX male sex reversal syndrome known as La Chappelle syndrome – one of the rarest sex chromosomal aberrations[1]. Sex reversal can be further classified as 46XX male sex reversal syndrome and 46XY female sex reversal syndrome. Classical 46XX male sex reversal has apparently normal male phenotype, while nonclassical XX male reversal syndrome has various degrees of sexual ambiguity like micropenis, hypospadias and 46XX true hermaphroditism.

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Case Report

A married couple presented with male factor infertility. The 38-year-old male partner had ambiguous phenotypic male characteristics, 5 feet height normal adrenarcheal hair pattern, unilateral left atrophic testis, right undescended testis, micropenis, underdeveloped scrotal folds and hypospadias (Figure 1 and 2). He could sustain erection for a short time but was not able to ejaculate. Investigations revealed elevated gonadotrophin levels (FSH 18mIU/mL, LH 16mIU/mL), estradiol (20 pg/mL) and testosterones low normal (0.45 pg/mL), progesterone 3mg/mL, OH-progesterone 6 nmol/L, cortisol (8gm) 486 nmol/L, DHES (38 µg/dL) thyroid (TSH 3.6 µIU/L) and prolactin (8 µg/mL). Sonographic evaluation showed small atrophic testis in the left scrotum and intraabdominal right testis measuring 3.1 cm x 2.6 cm. Laparotomy was performed. On opening the abdomen, an infantile uterus was present. Right sided small hydrosalpinx was present with normal ovary. Left ovary was atrophic and tube was normal. Right salpingo oophorectomy was performed, left nodular tissue present in the left scrotal fold was removed, and both the tissues were sent for histopathological examination and karyotyping. The karyotype report was 46XX (Figure 4). Histopathological examination showed that the tissue from the left scrotum had musculoskeletal tissue with no testicular tissue (Figure 3). Right sided specimen

Figure 1. Phenotypic appearance of the male parnter.

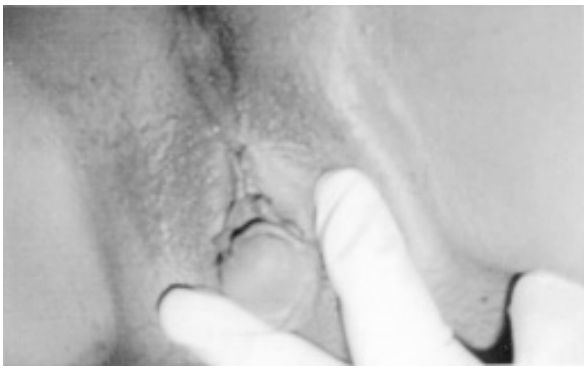

Figure 2. External genitals showing micropenis and underdeveloped scrotum.

revealed mainly ovarian tissue with few seminiferous tubules suggesting it to be ovotestis.

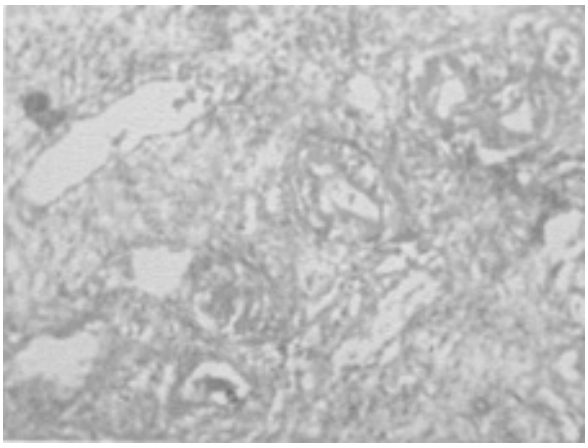

Figure 3. Micro histograph photograph HE stain 10x.

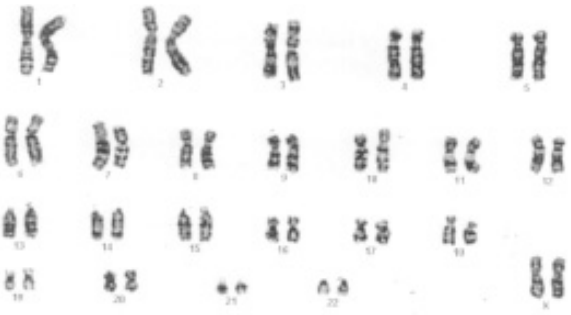

Figure 4. 46 XX, Karyotype

Discussion

This is rare presentation of infertility with the couple having identical sex. Phenotypic males are rarely found to have 46XX karyotype and only about 80 cases have been reported so far in literature and are usually SRY gene positive[2,3]. This couple was advised intrauterine insemination with donor sperm. The male was counseled to continue with the same gender as it was psychologically impractical to change the gender at this stage.

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