

## Testicular Feminization Syndrome - A Rare Case Report.

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### ABSTRACT

Testicular feminization syndrome is a genetic disorder that makes XY fetuses insensitive/unresponsive to the actions of the androgen hormones. Instead, they are born looking externally like normal girls and internally, there is a short blind pouch vagina and no uterus, fallopian tubes or ovaries. There are testis in the abdomen or in the inguinal canal. The androgen insensitivity syndrome occurs in one out of 20000 births. It can be incomplete (various sexual ambiguities) or complete (the person appears to be a woman). In this case report, a married nulliparous female reported to Gynecology & obstetric dept. for treatment of primary infertility and had history of primary amenorrhea. Physical examination revealed normal external female genitalia with normal breast development (Tanner Stage- 5) and absence of axillary and pubic hair. Bilateral adnexal masses were removed and sent for histopathological examination. On microscopic examination both the ovaries revealed seminiferous tubules lined only by the sertoli cells with no evidence of spermatogenesis. No ovarian tissue seen.

**Keywords:** Ambiguous genitalia, Testicular feminization syndrome, Androgen Insensitivity Syndrome.

### INTRODUCTION

Androgen insensitivity syndrome could be considered as disease that causes resistance to androgens actions, influencing both the morphogenesis and differentiation of the body structures and systems in which this hormone exerts its effects. The first who described this syndrome was John Morris.<sup>[1]</sup>

The clinical phenotypes of androgen insensitivity syndrome could vary and be classified into three categories, as complete (CAIS) and partial (PAIS) and mild (MAIS) forms and in seven grades in order of androgen resistance one evidenced by Quigley et al.<sup>[2]</sup>

CAIS is characterized by short blind ending vagina, absence of wolffian duct derived structures i.e epididymis, vas deferens, seminal vesicles and absence of prostate. One important feature that helps to address correct diagnosis is that at puberty breast develops regularly while there is scarce or absent development of pubic and axillary hair.<sup>[2]</sup>

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### CASE REPORT

30 year old nulliparous female married for 5 years reported to gynaecology & obstetric dept. for treatment of primary infertility and with the history of primary amenorrhea.

Physical examination revealed normal external female genitalia with normal breast development (Tanner Stage- 5) and absence of axillary and pubic hair. On per vaginal examination uterus is not palpable but bilateral adnexal masses palpated. On ultrasonography, uterus is hypoplastic, fibrotic, band-like and connecting the 2 ovaries, in right ovary a heterogeneous mass 90x81x86 mm. - ? teratoma ovary, left ovary appears normal.

Rest of the organs were normal. Bilateral adnexal masses were removed and sent for histopathological examination. On gross examination: We received two solid soft tissue pieces- first -an already cut open, encapsulated, well circumscribed globular grey-white soft tissue piece with smooth glistening surface measuring 6.5 x 5 x 2.5 cms. Cut surface was solid & homogenous grey-white to grey-brown. Second- another globular grey-white soft tissue piece - received in the same container measuring 4 x 3 x 2 cms. Cut surface was smooth with glistening surface.

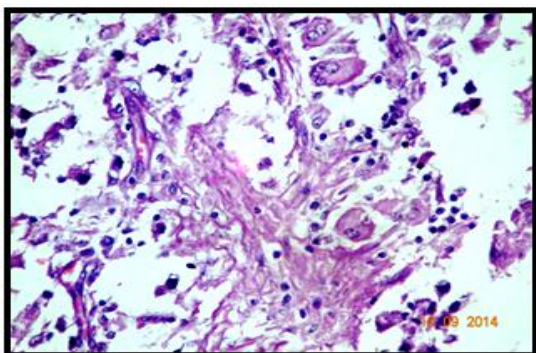


**Figure 1 & 2: Grey white to grey brown masses showing smooth glistening surface.**

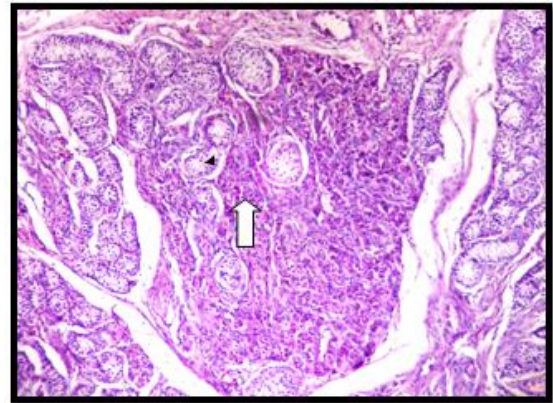
On microscopic examination

Larger mass showed structures resembling seminiferous tubules bounded by basement membrane & lined exclusively by tall columnar sertoli cells having uniform ovoid nuclei & abundant cytoplasm in the background of mesenchymal tissue, congested blood vessels with clusters of inflammatory cells (lymphocytes & a few giant cells). No evidence of spermatogenesis seen in the tubules. No ovarian tissue seen in the sections.

Smaller mass showed seminiferous tubules lined only by the sertoli cells. No evidence of spermatogenesis seen. Interstitial cells of leydig were also seen in between the tubules. No ovarian tissue seen.



**Figure 3: H and E (400x) Photomicrograph showing structures resembling seminiferous tubules and few giant cells (arrow).**



**Figure 4: H and E (100X) Photomicrograph showing seminiferous tubules (black arrow) with interspersed Leydig cells (empty arrow).**

## DISCUSSION

Androgen insensitivity syndrome (AIS) is the most frequent X-linked recessive disorders of sex development. These conditions are characterized by partial or complete inability of the cell to respond to androgen secondary to mutation in the androgen receptor gene located on long arm of the X chromosome (locus X q 11-12) in the form of complete or partial deletions, point mutations or small insertions. The unresponsiveness of the cell to the presence of androgen hormones can impair or prevent the masculinization of male genitalia, development of male secondary sexual characters with no significant impairment of female sexual development.<sup>[3]</sup> As in the normal male the testes appear to liberate the Mullerian inhibiting substance with the result that uterus and fallopian tubes are absent. The testes continue to develop but usually remain either in the abdomen (21%), the groin (60%),<sup>[4]</sup> or labia majora. Testosterone is formed normally during intrauterine life but due to inability of the target organs to respond to it, male characteristics fail to develop and the result is somatic female without uterus and tubes since the posterior vagina is also of Mullerian origin its development, too, is inhibited and a short blind vagina results. In our case the external genitalia was of female type with normal breast development. The chromosome make-up of patients with testicular feminization is well established and is usually 46 XY. Exceptions are the mosaics XO/XY/XX and XY/XYY/XXY.<sup>[5,6]</sup> Grossly testes is usually slightly smaller than normal, with cystic structures attached to one pole and a mass of fibromuscular tissue to the other. The tunica albuginea is thickened and the cut surface is of a light brown colour and often contains one or more circumscribed cream coloured nodules.<sup>[7]</sup> In our case also cut surface was glistening grey white to grey brown colour. On microscopic examination small seminiferous tubules without lumina composed of sertoli cells only, usually

immature, with sparse spermatogonia, marked Leydig cell hyperplasia (often without Reinke crystals), ovarian type stroma; nodules are probably hamartomas of Sertoli cells.<sup>[8]</sup> In our case similar findings were noted on microscopy.

## CONCLUSION

Testicular feminization is a rare disease that must be diagnosed and treated through close work between gynecologists and pathologists. Bilateral laparoscopic orchiectomy is the best procedure to remove intra-abdominal testis in order to avoid the malignant transformation.

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