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The XY Female: A Rare Disorder with Malignant Potential

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Abstract: A 16 year old female admitted to the hospital with the complaints of primary amenorrhoea and bilateral painless swellings in inguinal region. The clinical examinations and investigations revealed the case as complete androgen insensitivity syndrome. She underwent bilateral gonadectomy with herniotomy. Hormone replacement therapy with psychological counselling was started. The importance of early gonadectomy should be stressed in order to avoid the risk of malignancy.
Keywords: Androgen insensitivity syndrome, The XY female, Primary amenorrhoea, Gonadectomy.

INTRODUCTION

Androgen insensitivity syndrome is inherited as a X linked recessive disorder typically characterized by evidence of feminization (i.e., undermasculinization) of the external genitalia at birth, abnormal secondary sexual development in puberty, and infertility in individuals with a 46, XY karyotype. Genotypically they are male (XY) but phenotypically and psychologically female [1].

This condition occurs when end organs do not respond to androgens either due to lack of androgen cytosol receptor (complete androgen insensitivity syndrome) or defect in the receptor (partial androgen insensitivity syndrome). Affected individuals have male internal organs (testes) that are undescended which means they are abnormally located in the abdomen, inguinal canal or labia. These testes may become cancerous if not removed surgically. Most affected individuals report psychological trauma at diagnosis. We are reporting a case of XY female presented with primary amenorrhoea and bilateral inguinal swellings.

CASE REPORT

A 16 year old phenotypic female presented with primary amenorrhoea and bilateral painless swellings in inguinal region. The patient was 160 cm tall and weighed 48 kg. On physical examination she had normal breast development (Tanner stage III) but no growth of axillary and pubic hairs (Fig.1). Abdomen was soft with no organomegaly. Two non-tender oval swellings of sizes 3x2cm, 3x2.5cm were present in right and left inguinal regions respectively. Right inguinal swelling was felt near the deep ring that become prominent on coughing and apparently reduced in size in the normal standing posture. She had normal looking external genitalia. But the vagina was a blind pouch about 4 cm in length. Uterus was absent on bimanual per rectal examination.

Ultra-sonography showed absence of uterus and ovaries were not visible. An oval structure having homogenous echo-texture is noted in inguinal region on either side. The sizes of oval structures were 29x21 mm on right side and 23x16 mm on left side.

Routine investigations like haemogram, blood urea, sugar were within normal limits. Hormonal evaluation showed serum testosterone level 2.52 ng/ml which is a normal level in males and the level of other hormones were within normal range. Her karyotyping done by GTG banding technique showed 46XY (Fig. 2).

Laparoscopic evaluation showed absence of uterus and adnexae. A sac like structure peeping through the left deep inguinal ring. Right sided gonad could not visualized. She underwent bilateral gonadectomy with herniotomy (Fig. 3). Both testis were submitted for histopathological study which showed similar features and consist of closely spaced seminiferous tubules with marked loss of germ cells and

presence of Sertoli cells and clusters of Leydig cells in the stroma (Fig. 4). Hormone replacement therapy with psychological counselling was started. The diagnosis of complete androgen insensitivity syndrome (CAIS) was made as she presented with primary amenorrhoea with underdeveloped female external genitalia, better developed breast, absence of pubic and axillary hairs and uterus, presence of testes instead of ovaries with XY karyotype.

Fig-1: Phenotype female with normal breast development but no growth of axillary and pubic hairs

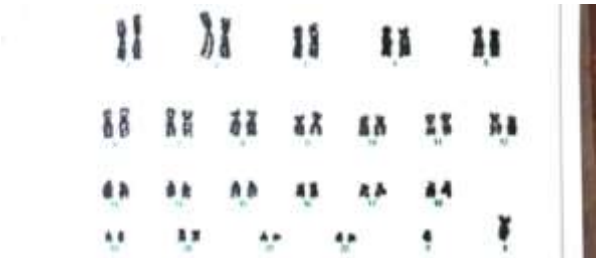

Fig. 2: Karyotyping done by GTG banding technique showed 46XY

Fig. 3: Bilateral Gonadectomy

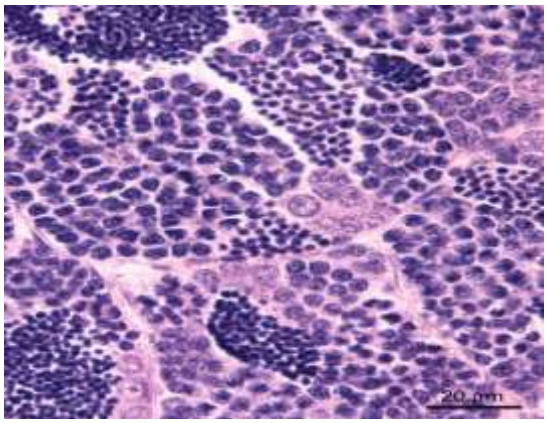

Fig. 4: Presence of Sertoli cells and clusters of Leydig cells in the stroma

DISCUSSION

Androgen insensitivity syndrome (AIS) accounts for about 10% of all cases of primary amenorrhea, the third most common cause after gonadal dysgenesis and congenital absence of vagina [2]. CAIS is estimated to occur in 1 out of every 20,400 male births [3].

The basic etiology of AIS is a loss-of-function mutation in the androgen receptor (*AR*) gene. This *AR* gene has been localized to the long arm of the X chromosome (ie, Xq11-13) [4].

AIS represents a spectrum of defects in androgen action and can be subdivided into three broad phenotypes:

- Complete androgen insensitivity syndrome (CAIS), with typical female external genitalia
- Partial androgen insensitivity syndrome (PAIS) with predominantly female, predominantly male, or ambiguous external genitalia
- Mild androgen insensitivity syndrome (MAIS) with typical male external genitalia

For individuals with AIS, the standard of care is an orchidectomy to prevent possible malignant degeneration of the testes [5]. Untreated patients have a theoretical risk of malignant degeneration and development of gonadoblastoma of the testes. Rather inaccurate risk estimates range from 0-22% in adults with CAIS [6].

Medical care for a patient with AIS has two aspects: hormone replacement therapy (HRT) and psychological support. For patients with CAIS, hormone therapy almost always consists of estrogen replacement.

Phenotypic females who are discovered to be genetic males may have psychosocial problems. Many of these patients have been told that they really are not women but actually are men because of the presence of a Y chromosome and testes. These difficulties and doubts of self-identity often cause shame and self-doubt as well as anger and frustration. These females require sensitive psychological support. Their psychosocial problems range from identity issues to problems dealing with the gender perceptions of the outside world and future family life. In patients who develop virilisation and have an XY karyotype, the testes should be removed immediately to preserve the female phenotype and female gender identity.

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