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Case Report : Androgen insensitivity syndrome

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ABSTRACT *Androgen insensitivity syndrome or testicular feminization syndrome, is a rare X-linked recessive disorder with an incidence of 1:20,000-64,000 male births, encompasses a male genotype (46XY) with phenotypes ranging from male infertility to variable ambiguity of genitalia to completely normal female external genitalia, that are caused by numerous (>400) different mutations in the androgen receptor (AR) gene located at Xq11-12, with a resulting decrease in binding of androgen to the receptor.*

We report a case in which a 26 yrs old female presented with primary amenorrhoea . She had right sided inguinal hernia repair with probably right gonadectomy in infancy. Subsequent investigations were done which revealed absence of female internal genitalia, a male karyotype and presence of left testes. Left gonadectomy was performed and she was started with hormone replacement therapy.

Full Text:

26-year-old woman presented with primary amenorrhea. There was history of surgery for right inguinal hernia with removal of a lump at the age of 10 months. Her two younger sisters were not affected.

The patient had a female habitus and voice, a normal intellectual function and an average height and built.

On examination, breast development was inadequate (Tanner II), axillary hair absent and had only sparse pubic hair (Tanner II).

There was a linear scar mark seen on right inguinal area.

Systemic and abdominal examination were normal.

She had normal female external genitalia with labia, clitoris, vaginal introitus; her vagina was functional (10 cm in length) but ended in a blind pouch and uterus was absent.

Ultrasonography revealed absent uterus and right ovary; left ovary seen 29x16 mm

Her karyotype revealed 46 XY pattern.

She was posted for diagnostic laparoscopy which revealed no uterus and right gonad, but left ovo-testes was present, which were removed laparoscopically.

Picture: Left ovo testes removed laproscopically.

Histologic examination revealed ovotestes with small seminiferous tubules without spermatogenesis. These were predominantly lined by sertoli cells. At places, the stroma showed clusters of leydig cells. In the wall questionable ovarian stroma was seen.

Reconstructive surgery to external genitalia was not needed.

Proper counselling was done and she was put on hormone replacement therapy.

REVIEW

Androgen insensitivity syndrome (AIS), formerly known as testicular feminization, is an X-linked recessive condition resulting in a failure of normal masculinization of the external genitalia in chromosomally male individuals. This failure of virilization can be either complete androgen insensitivity syndrome (CAIS) or partial androgen insensitivity syndrome (PAIS), depending on the amount of residual receptor function.

The basic etiology of androgen insensitivity syndrome is a loss-of-function mutation in the androgen receptor (*AR*) gene. Loss of AR function means that, despite normal levels of androgen synthesis, the typical postreceptor events that mediate the effects of hormones on tissues do not occur.

In 1953 the first description of the AIS was reported by John Morris.

In 1989, the exact localization of the human Androgen Receptor gene (AR gene) was defined on Xq11-12[5] and in the same year the first proof that AIS was caused by mutations in the AR gene was published by Brown et al.

Subjects with CAIS are born unambiguously female and are not suspected of being abnormal until the onset of puberty, when breast development is normal, external genitalia is normal feminine but pubic and axillary hair is not developed and menses do not occur. The high levels

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of testosterone, a substrate for aromatase activity, result in substantial amounts of estrogens, which are responsible for very good breast development at puberty in CAIS individuals. A number of patients are recognised when they present inguinal hernia.

The phenotype of individuals with partial androgen insensitivity syndrome may range from mildly virilized female external genitalia (clitorimegaly without other external anomalies) to mildly undervirilized male external genitalia (hypospadias and/or diminished penile size).

In either case, affected individuals have 46,XY karyotypes, normal testes with normal production of testosterone and normal conversion to dihydrotestosterone (DHT).

Management:
Psychological support is probably the most important aspect of medical care from the patient's point of view. In a family with an affected infant, the parents are the primary clients. Parents need genetic counseling to understand the nature of the condition and the risk of recurrence (25% for each subsequent pregnancy) as well as to identify other potential carriers.

Reconstructive surgery to external genitilia is not needed in the complete form but gonads need to be removed due to risk of malignancy. For management of incomplete form both gonadectomy and reconstructive surgery of external genitilia is required.

A common practice is to remove the testes after puberty when feminization of the affected individual is complete, since feminization occurs partly by testicular estrogen and partly by peripheral conversion of androgen to estrogen. The reason for the postpubertal gonadectomy is the risk of testicular malignancy, which seldom occurs before puberty.

Orchidectomy and vaginal lengthening procedures may be performed concurrently if surgery is postponed until the patient matures. Ultrasound examination of the gonads can monitor potential tumor development

Prepubertal gonadectomy is indicated if inguinal testes are physically or esthetically uncomfortable and if inguinal herniorrhaphy is necessary. In this case, estrogen replacement therapy is necessary to initiate puberty, maintain feminization and avoid osteoporosis.

Histologic examination of the testes in patients with complete androgen insensitivity syndrome or partial androgen insensitivity syndrome should show fairly normal testicular structure, although the numbers of spermatogonia and/or sperm may be reduced markedly in postpubertal patients.

Osteoporosis and psychological sequelae are the 2 major complications of androgen insensitivity syndrome, and their risk can be decreased significantly by appropriate therapeutic intervention. These interventions involve HRT with estrogen to prevent osteoporosis and early and continuing involvement with an appropriate mental health professional for psychological and emotional support.

Awareness of this entity is important as with early diagonosis such disorder can be managed appropriately and accurate information can be given to parents regarding long term issues of hormone replacement therapy and fertility.