

Testicular Feminization Syndrome

Vaneet Kour, Ajay Abrol*

Abstract

Testicular feminization syndrome or androgen insensitivity syndrome is a rare disorder with an incidence of 1:20,000-64,000 male births. The individual with complete form of this syndrome (CIAS) have female external genitalia while those with partial form (PIAS) have variable ambiguity of genitalia and often need extensive reconstructive surgery. The diagnosis should be suspected in female child with inguinal hernia or presenting with primary amenorrhea and on examination there is no vagina with absent axillary or pubic hair. Awareness of this entity is important as with early diagnosis such disorder can be managed appropriately and accurate information can be given to parents regarding long term issues of hormone replacement therapy and fertility.

Key Words

CAIS, PAIS, Androgen

Introduction

This syndrome also known as androgen insensitivity syndrome was 1st described by Morris at Yale. The phenotype is a female despite the normal male karyotyping 46 XY. The etiology of this syndrome is congenital insensitivity to androgens transmitted by means of a maternal X-linked recessive gene responsible for androgen intracellular receptors (1). The incidence of androgen insensitivity syndrome is estimated to be 1:20,000-64,000 male births and with variable phenotype expression, this syndrome can present as complete or partial forms (2).

Case Report

37 years old school teacher, Miss M. presented in Outpatient's Department of Obstetrics and Gynecology with complaints of primary amenorrhea. There was past history of some surgery done in the inguinal area 14 years back. On examination, she was tall, thin built, height was 64 inches and weight was 55 kgs. Pubic and axillary hair

were absent. Breasts were poorly developed. There were two linear scar marks seen on both inguinal areas. Systemic and abdominal examination were normal. External genitalia showed no definitive labia. External urethral meatus was patulous. A small hypoplastic phallus was seen. A blind pouch about 1.5 cms was seen below the urethra (Fig.1).

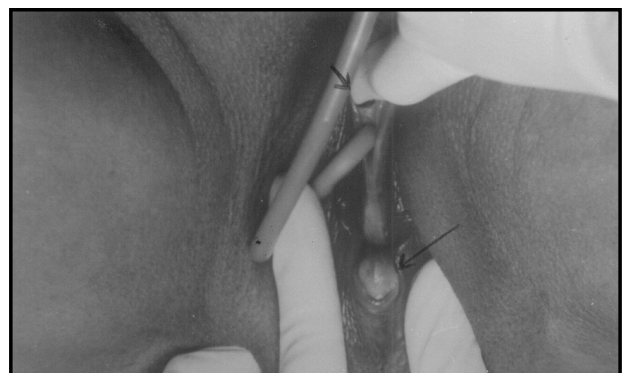


Fig. 1. Showing a blind pouch below the urethra

From The Deptt. of Obstetrics & Gynae and *Plastic & Reconstructive Surgery, Christian Medical College & Hospital, Ludhiana.
Correspondence to: Dr. Vaneet Kaur, Reader, Department of Obst. & Gynae, Christian Medical College & Hospital, Ludhiana, Punjab.

Ultrasonography of pelvis showed absent uterus and ovaries (Fig.2). Karyotyping was 46 XY. IVP showed normal kidneys, ureter and bladder. FSH was 81 and LH was 19 mIU/ml respectively. Diagnostic Laparoscopy showed absent uterus and ovaries. Vaginoplasty using split skin thickness graft was done.



Fig. 2. Ultrasonography of pelvis showing absent uterus and ovaries.

Discussion

Testicular feminization syndrome may present as complete form (CAIS) and incomplete form (PAIS) (2). In the complete form, there is no androgen response, therefore normal external female genitalia develop and these infants are reared as females. There may be labial or inguinal swellings which contain testis. These patients most often present in late adolescence with primary amenorrhoea. There is absence of uterus and ovaries on ultrasound scan or laparoscopy. Vagina is short, develops from urogenital sinus only and ends blindly.

The partial or incomplete form of testicular feminization syndrome is associated with wide range of genital abnormalities and typically present at birth with genital ambiguity. Severe hypospadias associated with micropenis, bifid scrotum and bilateral cryptorchidism are common. In some patients, the external genital phenotype may be predominantly female with partial labial fusion and clitoromegaly (3). Ultrasonography or laparoscopy should be done in all such patients to examine internal genital organs. Measurement of serum 17-hydroxyprogesterone and its sulphate can be done to detect testosterone biosynthetic defects (2). Management consist of appropriate counseling of parents regarding fertility and long term use of HRT needs to be discussed. Reconstructive surgery to external genitalia 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 genitalia is required (4).

References

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