CASE REPORT

Testicular Feminization Syndrome: A Case Report

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Abstract

Testicular feminization syndrome or androgen insensitivity syndrome is a rare disorder. 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 etiology of this syndrome is congenital insensitivity to androgens transmitted by means of a maternal X-linked recessive gene responsible for androgen intracellular receptors. We have found an un-usual presentation of female phenotype with androgen insensitivity syndrome, which is discussed.

Keywords: *Androgen insensitivity, CIAS, PIAS, Testicular feminization.*

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Introduction

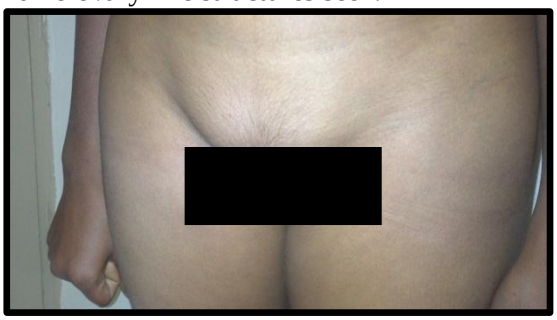
Testicular feminization syndrome or androgen insensitivity syndrome is a rare disorder with an incidence of 1:20,000-64,000 male births. The individual with complete form of this syndrome (CIAS) have female external genitalia while those with partial form (PIAS) have variable ambiguity of genitalia and often need extensive reconstructive surgery [(1)]. This syndrome was 1st described by John Morris in 1953. 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 [(2)]. We are reporting the un-usual presentation of female phenotype with androgen insensitivity syndrome (Testicular feminization syndrome).

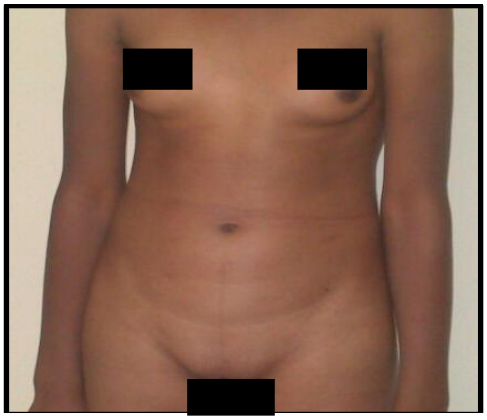
Case Report

A 14 year female came in OPD with complaints of swelling on both inguinal regions from 7 to 8 years and pain off and on in that area from last 2 years. On Examination breast normal, pubic hair and axillary hair absent. On examination bilateral inguinal region testes were palpable.

Sonographic Findings

On USG pelvis two well defined ovoid soft tissue structure, seen at the level of superficial inguinal rings on both sides with epididymis (indistinct). No uterus seen but two ovaries like structures seen with multiple tiny anechoic areas. On transrectal USG, there was no uterus and no ovary like structures seen.





Other Investigations

Serum testosterone level was found to be 0.81ng/ml, serum LH was 7.2m IU/ml while serum FSH level was 15m IU/ml. Barr body examination was found to be negative. Karyotyping was 46XY.

Discussion

Testicular feminization syndrome may present as complete form Complete Androgenic Insensitivity (CAIS) and incomplete form Partial Androgenic Insensitivity (PAIS) [(2)]. Variable phenotypic expression has allowed the classification of AIS into complete (CAIS) and partial forms (PAIS) [(4)]. 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 [(3)]. People with CAIS are normal appearing females, despite the presence of testes and a 46XY chromosome constitution [(5)]. 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 [(3)]. The common associated genitalia abnormalities are hypospadias, micropenis, bifid scrotum and bilateral cryptorchidism predominantly in male phenotype. CAIS is caused by mutations in the androgen receptor gene, resulting in impaired embryonic sex differentiation and producing a female external phenotype [(6)].

The differential diagnosis includes 17-Hydroxylase Deficiency Syndrome, 3-Beta-Hydroxysteroid Dehydrogenase Deficiency, Congenital Hyperplasia, Mayer-Rokitansky Syndrome and 5 alfareductase deficiencies [(7)].

Androgen insensitivity syndrome (complete or partial), has modest morbidity or mortality. Though, un-treated patients have a risk of progression to seminoma and gonadoblastoma of the testes as documented in the medical theory. The psychological morbidity in form of psychological trauma is very frequent. Phenotypic females who are discovered to be genetic males may have psychosocial problems. The diagnostic work up includes ultrasound, CT scan, karyotyping, hormonal profile (testosterone, LH, FSH), biopsy and diagnostic laparoscopy. Management includes of genetic counselling of parents and patient, in addition to gonadectomy and reconstructive surgery [(7)].

Conclusion

Testicular feminization syndrome or androgen insensitivity syndrome is a rare disorder. Androgen insensitivity syndrome (complete or partial), has modest morbidity or mortality. Un-treated patients have a risk of progression to seminoma and gonadoblastoma of the testes. The psychological morbidity in form of psychological trauma is very frequent. Such cases can be diagnosed with wide variety of diagnostic aids like USG, karyotyping and hormonal profile. Management options available are genetic counseling of parents and patient, in addition to gonadectomy and reconstructive surgery.

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