Volume-4, Issue-6, June-2015 • ISSN No 2277 - 8160

Research Paper | Medical Science

Complete Androgen Insensitivity Syndrome; A Rare Case Report.

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ABSTRACT

Androgen insensitivity syndrome, also known as testicular feminization syndrome is an X linked recessive disorder, a condition resulting from an end organ resistance to androgens (particularly testosterone). Affected individuals are genotypically male with 46XY karyotype but with a female phenotype. Incidence being 1in 20000-60000 live births. It is most common cause of male psuedohermophroditism and 3rd most common cause of amonorrhoea.2We are reporting a case of 47 years female patient, presented with left inguinal hernia at the age of 47yrs. Patient had primary amennorrhoea. Physical examination showed absence of axillary hair and sparse pubic hair. Per vaginal examination revealed blind vagina, uterus and cervix could not be made out. USG pelvis showed absence of uterus and ovaries and well defined isoechoic round to oval structures in right and left inguinal regions. Histopathology of excised specimen of bilateral inguinal swellings showed testes with atrophied and hyalinised seminiferous tubules with dense leydig cell hyperplasia. Left testes showed a sertoli cell nodule. The diagnosis of Complete Androgen Insensitivity Syndrome was made based on gynecological examination, ultrasonography and histopathology. Patients can be helped to achieve an excellent quality of life as a female by multispeciality approach. Prognosis is favourable if psychological support and counseling are appropriate.

KEYWORDS : Complete Androgen Insensitivity Syndrome, Testes

Introduction

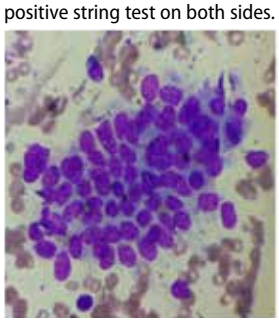
Androgen insensitivity syndrome, also known as testicular feminization syndrome is an X linked recessive disorder, a condition resulting from an end organ resistance to androgens(**particularly testosterone**). Affected individuals are genotypically male with 46XY karyotype but with a female phenotype. Incidence being 1 in 20000-60000 live births.[1]

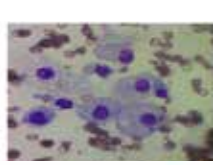
Case details:

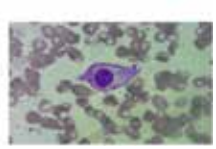
A 47yr old female presented with swelling in the left groin since 8 months with history of primary amenorrhoea, with married life of 25yrs. On physical examination, the patient had well developed breasts with infantile areola and nipple with absent axillary hair and sparse pubic hair. **She had a normal height**. External genital examination revealed presence of labia majora and labia minora. Left sided labia majora showed a swelling which showed positive cough impulse and clitoris was not made out.Per speculum examination showed blind vagina and cervix not made out.Per vaginal examination revealed blind vagina, uterus and cervix could not be made out.

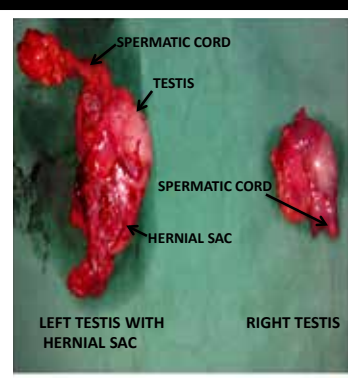
Routine investigations were within normal limits. Pelvic ultrasonography showed absence of uterus and ovaries and well defined isoechoic round to oval structures in right and left inguinal regions.Fine needle aspiration of inguinal swelling suggested the possibility of testicular tissue.Excision of bilateral inguinal swellings and left sided meshplasty was done and the specimen was sent for histopathology examination.

Grossly Specimen consisted of two round to oval structures resembling testis each measuring 4x2x1.5cms with left side hernial sac. Cut surface showed grey white to grey yellow structure which showed positive string test on both sides.



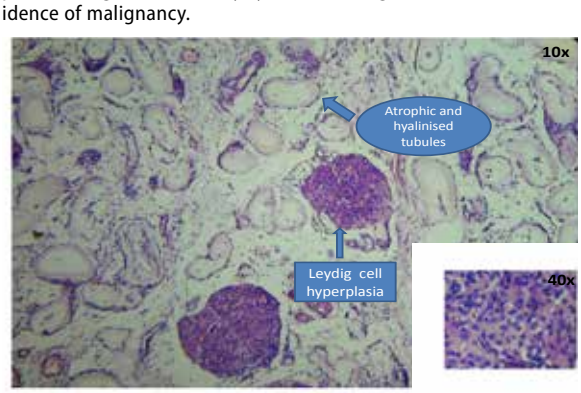




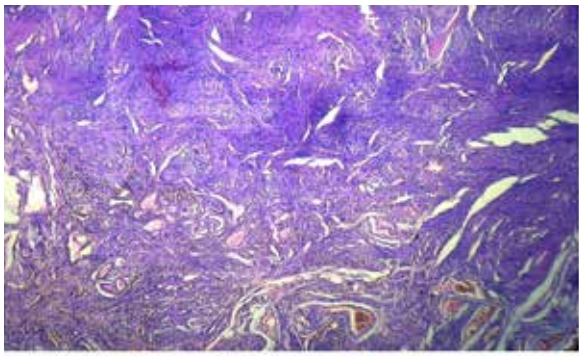


Well encapsulated grey white to grey yellow structure measuring 4×2×1.5cms, showed positive String test.

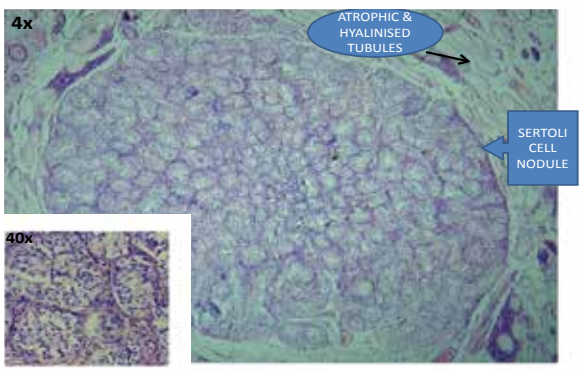
Microscopyof both the specimens showed structures of testes and vas deferens. Both testes showed atrophic and hyalinised seminiferous tubules lined by sertoli cells. Interstitium was edematous with dense leydig cell hyperplasia. Few areas showed abundant reticular fibres resembling ovarian stroma. A bundle of smooth muscle was present at one pole of both the testes. Left testis also showed a sertoli cell nodule. Final impression of complete androgen insensitivity syndrome was given.There was no evidence of malignancy.



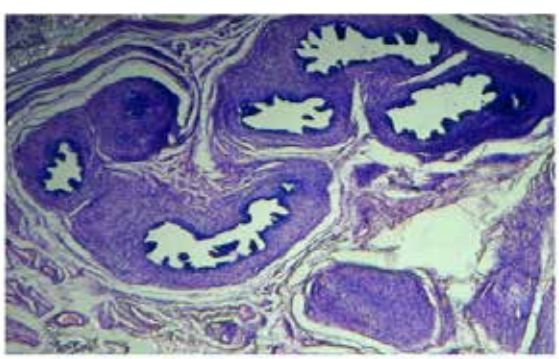
ATROPHIC AND HYALINISED SEMINIFEROUS TUBULES WITH AREAS OF LEYDIG CELL HYPERPLASIA



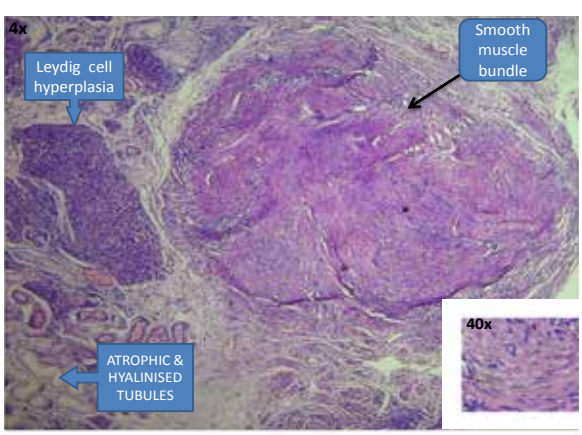
ABUNDANT RETICULAR FIBRES RESEMBLING OVARIAN STROMA



SERTOLI CELL NODULE IN THE LEFT TESTIS



VAS DEFERENS SHOWING FOLDED MUCOSA LINED BY PSUEDOSTRATIFIED COLUMNAR EPITHELIUM AND THICK MUSCLE COAT



BUNDLE OF SMOOTH MUSCLE WITH AREAS OF LEYDIG CELL HYPERPLASIA AND FEW ATROPHIC AND HYALINISED TUBULES

Discussion:

Androgen Insensitivity syndrome is most common cause of male psuedohermophroditism and 3rd most common cause of primary amenorrhoea.[3]Typical mode of presentation is an adolescent female, who has breast developments with pubertal growth but has not attained menarche with absent or sparse pubic and axillary hair.[1] As a rule both cervix and body of uterus are absent with blind vagina. The testes are cryptorchid and may be located in inguinal canal, pelvis and rarely labia. The epididymis ,vas deferens ,seminal vesicle and prostate are absent.[2]In our case vas deferens was present which was consistent with the study done by Hannema SE et al who found epididymis and or vas deferens in 36% of the cases of CAIS they studied.[6]

The incidence of tumors in CAIS cases is rare before puberty and significantly higher after the age of 35yrs.[3]Hamartomatous nodules have been present, usually bilaterally in virtually every case. The typical size varies from 1mm to 1cm but occasionally upto 4cm. The bulk of nodule is usually composed of seminiferous tubules lacking lumina. **Spermatogonia may be present.** [2]HirashiTsubamoto et al. and FarukZorlu et al. reported that 2 cases of CAIS developed malignancy at the age of 36yrs and 31yrs respectively.[4] Unexpectedly, in our case there was absence of malignancy even though the patient was 47yrs old.

In our case the diagnosis of complete androgen insensitivity syndrome was made based on gynecological examination, ultrasonography and histopathology.Management includes removal of the testes either after puberty when feminization is complete or before puberty followed by estrogen replacement therapy at the age of puberty and surgical correction.[5] Prognosis is favourable if psychological support and counselling are appopriate.[5]

Conclusion:

Androgen insensitivity syndrome although very rare, is extremely distressing to the concerned individual and requires expert and sympathetic handling. Patients can be helped to achieve an excellent quality of life as a female by a multispeciality approach including gonadectomy, surgical correction, psychological counselling, hormonal replacement along with bone mineral density scan every 5yrs.

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