AN INTERESTING CASE OF INTERSEX DISORDER

AN UNCOMMON PRESENTATION OF A TRUE HERMOPHRODITE!!!!

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CASE HISTORY-

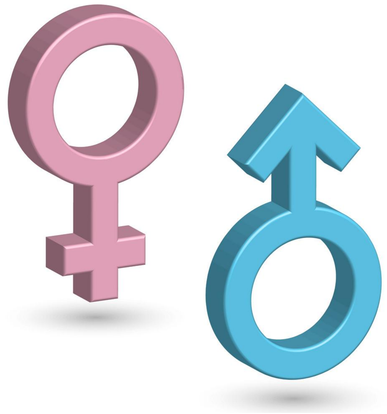
A 14 year old reared as male presented to the pediatric surgery OPD with complaints of breast tissue enlargement .No history of any drug intake. No history of hypothyroidism, obesity etc

PAST HISTORY -

History of bilateral undescended testes at birth. At 4 years underwent Left orchidopexy and Right inguinal exploration.



EXAMINATION



GENERAL PHYSICAL EXAMINATION

Patient is a young male, moderately built and. nourished , conscious
VITALS - WNL

SYSTEMIC EXAMINATION

CVS / RS- NAD

P/A- Soft, non tender, no palpable mass, bowel sounds heard

BREAST EXAMINATION

- Revealed bilateral well formed areola
- Papilla protruding and forming a secondary mound over the breast

INSPECTION OF SECONDARY SEXUAL CHARACTERS



TANNER IV



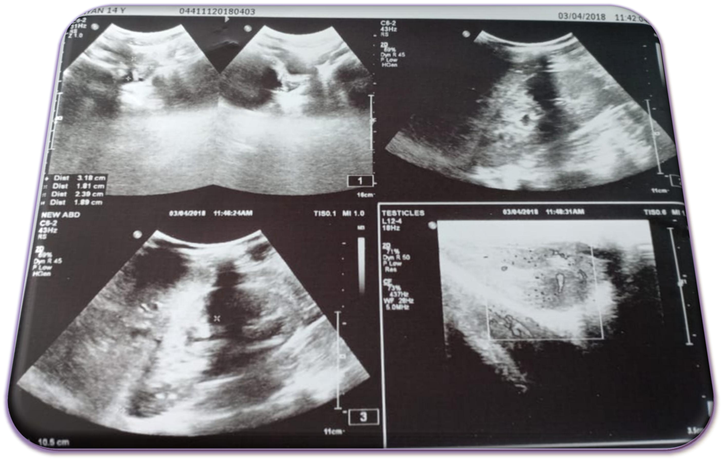
TANNER III

GENITAL EXAMINATION

- Adequately developed phallus with no signs of hypospadias
- Normal urethral opening and of adequate length
- Testis palpable in the left hemiscrotum .
- Right hemiscrotum is empty
- No other swellings noted over the inguinal region, root of the scrotum or the medial side of the thigh with

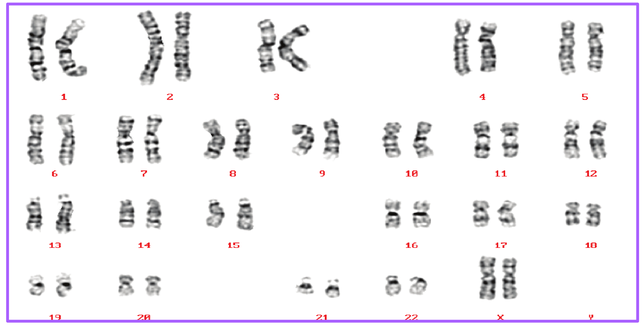
INVESTIGATIONS

ULTRASONOGRAPHY



- Normal bladder with infantile prostate noted.
- Right ovary noted measuring 32*18mm with few follicles seen within
- Scrotum revealed normal sized left testis 24*15mm with empty right hemiscrotum.

KARYOTYPE 46XX



BLOOD PARAMETERS

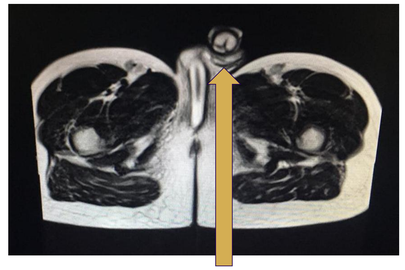
ROUTINE BLOOD INVESTIGATIONS- NORMAL

- Lipid profile- normal
- Total testosterone- 177.3ng/dl(260-1000 in males and 15-70 in females)
- Sex hormone binding globulin- 37.9nmol/dl(13-71 in males and 18-114 in females)
- Free testosterone- 3.14ng/dl(5-21 in males and 0.1 -0.85 in females)
- Bioavailable testosterone- 70.32^{ng}/dl(66-417 in males and 3.14-20.20ng/dl in females)
- Luteinizing hormone- 30.13mIU/ml(RAISED)
  Thyroid profile – NORMAL
- AFP- 0.974ng/dl(NORMAL)

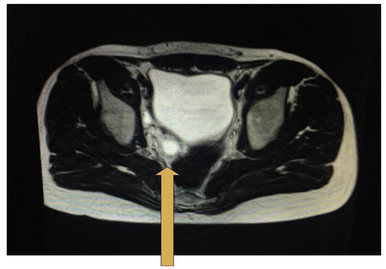
Follicle stimulating hormone- 11.9mIU/ml(NORMAL)

- Prolactin- 6.7ng/dl(NORMAL)

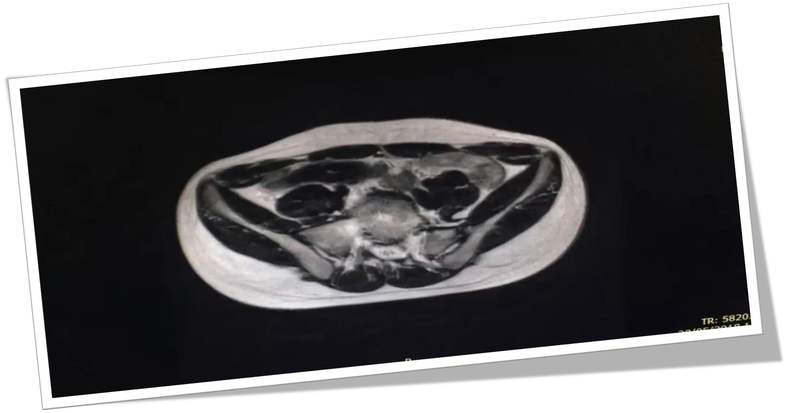
MRI ABDOMEN & PELVIS



TESTICULAR GONAD



INTRA- ABDOMINAL GONAD



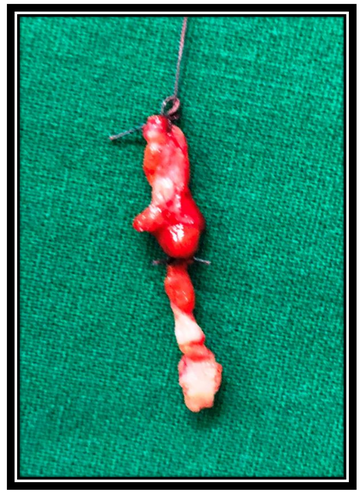
- Right ovary visualised and measures 3.1*3.0*2.6 with volume of 13cc with multiple follicles noted .
- Left ovary not visualised
- Prostate appears normal for the age
- A well defined hyperintense structure with a hypointense rim measurimg 2.0*1.8cm seen in left scrotal sac- likely rudimentary testes

INTERVENSION

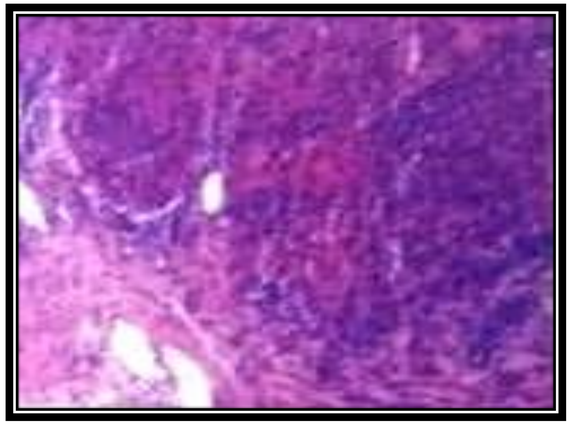


Gonadal tissue found intra-abdominally with soft follicular ovarian tissue on one pole and firm testicular tissue on the other pole





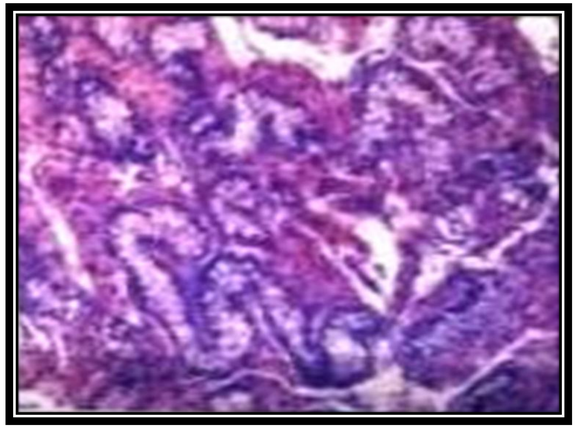
Left gonad biopsy



INTRA-OP

- Gonad excised from abdominal cavity contains both ovarian and testicular tissue
- Ovarian tissue shows corpus albicans, corpus luteum , focal aggregates of hemosiderin laden macrophages.
- Testicular tissue shows infantile seminiferous tubues containing sertoli cells. Leydig cells seen in interstitium.
- No granuloma and malignancy

- Biopsy of the testicular tissue shows Infantile seminiferous tubules containing sertoli cells.
- Leydig cells are seen in the interstitium.
- No definite ovarian tissue identified.
- No germ cell neoplasia, granuloma and malignanacy



LAST BUT NOT THE LEAST

POST OPERATIVE PICTURES





FOLLOW UP

- Endocrinology– testosterone level monitoring
- Ultrasound annually of the left gonad
- Fertlity issues

CONCLUSION

Ovotesticular disorder of sex development is the rarest form among all disorders of sex development and characterized by the simultaneous presence of both ovarian and testicular tissues in the same individual.

While it characteristically presents with ambiguous genitalia in neonates or infants, it may also present in adulthood. The management of a patient with ovotesticular DSD must be carried out by a multidisciplinary team.

DISCUSSION

- Diagnosed as 46XX Ovotesticular DSD
- Disorders of sex development (DSDs) are a group of rare and complex disorders characterized by abnormalities of chromosomal, gonadal, or phenotypic properties that determine sex development.
- The prevalence of DSD is reported to be 1.8 per 10,000 live births.
- Ovotesticular DSD rarest with prevalence of 1/20,000, and about 500 affected individuals have been reported to date.
- Characterized by presence of both ovarian and testicular
- It has been reported that only about 10% of all patients with ovotesticular DSD have a phenotypically normal or nearly normal male penis and diagnosed later during adolescence.
- Most commonly detected chromosomal makeup among patients with ovotesticular DSD is a 46, XX karyotype.
- The present case is an extremely rare one in that the external genitalia is nearly normal, and the patient (and his parents) had no doubt about any sexual abnormality until a progressive breast enlargement occurred,.

THANK YOU