A CASE OF OVOTESTICULAR SYNDROME

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ABSTRACT

Ovotestis refers to the histology of a gonad that contains both ovarian follicles and testicular tubular elements. Ovotesticular disorder of sexual development (OT-DSD), formerly known as true hermaphroditism/ Intersex. NORD- National Organization Rare Diseases – only 500 Such cases are reported – till 2012 & more rarely, the disease with males is extremely rare.

KEYWORDS: We intend to present this interesting case considering it’s rarity.

INTRODUCTION

Ovo-testes (formerly called “true hermaphroditism”) are gonads containing both ovarian and testicular tissue.[1] These are sometimes present in place of one or both ovaries or testes. A person may be born with two Ovotestis, or a person might be born with one ovary and one Ovotestis, or many more combinations.[2] Ovotestis is compartmentalized, where connective tissue separating the ovarian component from testicular components. Additionally, testicular and ovarian tissue may develop on the same side of the pelvis as a separate ovary and testis. But on rare occasions, an intermixt of these elements may occur. Ovotesticular disorder of sexual development is a rare condition.[2,3] Most cases have sporadic distribution, although there are a few documented cases of familial recurrence. Genital ambiguity occurs in 1 in 4500 births and Ovotesticular disorder of sexual development occurs in fewer than 10% of all disorders of sexual development.[3] Testicular tissue in Ovotestis involves an increased risk of gonadal cancer incidence is 2.6% of all cases of Ovotesticular disorder of sexual development. The chromosomal study and malignancy when compared 46,XY karyotype are at greater risk of developing a gonadal malignancy. These patients need removal of testicular portion of the Ovotestis.[4] We present this rare case of Ovotestis.

Case History:- A 28 year old male presented to us with episodes of severe pain in abdomen.

Feeling of fullness in lower abdomen. Such episodes last for about a week, and were interspersed by an asymptomatic period. Patient used to get bleeding Per rectum, during this period No other contributory history noted ---Rt. orchietomy 25 years back, records of which were not available. H/O neck mass 10 months back for which patient underwent investigations (FNAC) and was labelled as Metastatic testicular Teratoma, Patient was given 3 cycles of chemotheraphy BEC (Bleomycin Cisplatin Etoposide). Patient had a short stature. Secondary sexual characters were underdeveloped with micro-phallus with hypospadias. B/L Gynaecomastia was present. Scar of previous surgery in Rt. Inguinal region. Left sided Testis was small. Abdominal examination vague mass noted in lower abdomen, Systemic examination was normal. Dilemma about this case was because, Metastatic Teratoma in the Neck, was treated with Chemotherapy. So lump was Retroperitoneal lymph node mass or what?????? Routine investigations were within normal limits. Tumor markers for testicular malignancy were within Normal Limits. X ray chest PA view, No abnormality detected. Ultrasonography abdomen and pelvis evidence of a cystic lesion of 10 X 6.5 X 7 cm extending into pelvis up to retro vesical region on right side. Contrast enhanced Computed tomogram,- Abdomen & Pelvis– A well defined soft tissue attenuation of 5.5 X 4.5 X 10.4 cm in AP transverse and CC noted in the Rt. side of pelvis with hypodense areas with extensions as follows - Superior - Intestinal loops. Rt. lateral: Rt. common Iliac Artery and Psoas muscle, Inferior: Lesion abutting Rt. wall of the rectum. Anterior: Ant. Sup. and Ant. Inf. to the bladder. Posterior: Thickened cystic collection in Rt. iliac fossa. No comment on ovaries or presence of uterus in this male patient.

Figure 1:- CT Scan

Considering the history and investigatory findings provisional diagnosis of ???? Retroperitoneal lymph nodal mass with cystic degeneration after CT was made.
Decision of performing a Diagnostic laparoscopy was taken. However due to technical difficulties on the day of surgery, we had to perform a Laparotomy. Findings of operation were to our surprise was, a uterus with a functioning endometrium as bleeding is seen, so we searched for an ovary and got one ovary (Left Ovary) but Right ovary we could not be found. The possibility of Ovotestis containing Ovary belong to Right ovary. We did a hysterectomy with oophorectomy. The post operative course was uneventful.

Imaging of Pelvis by MR scan.

Uterus on Exploration of Abdomen

Functioning endometrium in Uterus

Image: H & E stained slide showing presence of seminiferous tubules with ovarian follicle.

DISCUSSION
In Ovotesticular DSD the Ovotestis is the most common gonad, and it may reside at any point along the path of testicular descent.[1,2] Verkauskas et al reported 33 patients with histologically confirmed Ovotesticular DSD. The Ovotestis was the most frequent gonadal findings (43/66, 65%) in 33 patients and 31 out of the 43 Ovotestis were located in the abdominal region.[3] Most children have the unilateral uterus, which may be fully developed as happened in our patient, Approximately 60% patients with Ovotestis have a 46,XX karyotype; 33% are mosaic with second cell line containing a chromosome (46,XX/ 46 XY;46,XX/ 46 XXY) and 7% are 46, XY.[1,2] There are reports of lately diagnosed patients with Ovotesticular DSD because of unusual presentations. As in our case patient presented in his adult age, with gynaecomastia, Hypospadias and so syndromic presentation, but the dilemma was created by the history of the patient with a cytology report of Left supraclavicular nodal metastasis of testicular tumour for which he received 3 cycles of platinum based chemotherapy. Patient had a very interesting history, of Right orchiectomy 25 years ago. It is very difficult to diagnose Ovotestis DSD. Ovotesticular DSD, can be diagnosed by histopathological methods, showing the presence of both ovarian tissue with follicles and testicular tissue with seminiferous tubules in the same individual.[3,4] Ovotesticular DSD constitutes 3% – 10% of all sexual disorders. Patients present with ambiguous genitalia in the neonatal period; May Present with complete female phenotype / male phenotype. Both Wolffian and Mullerian structures can exist depending on the maturity of the testicular tissue n patients who have a male Phenotype, micropenis, scrotal hypoplasia, cryptorchidism, hypospadias, and labioscrotal fusion may be observed.[5] Chromosomal study was performed which Revealed normal study. In our patient even CT scan could not pick up the uterus.[6] It was only the surgical exploration and thorough search revealed that it is functioning uterus and we could trace only one ovary in the abdomen. Later on after counselling with the patient, Testicular biopsy was taken and it revealed both testicular as well as ovarian tissue. Serum testosterone was found to be low. 187ng/dl- As reported in the literature, our patient did not have a complete male phenotype, although his external genitalia had an incompletely masculinized appearance. Approximately 15% patients with hypospadias and a palpable undescended gonad has DSD, while approximately 50% of patients with hypospadias and unilateral nonpalpable gonad had DSD.[7] McAleer et al do not recommend routine karyotyping in all patients with hypospadias and UDT because the incidence of sex chromosome abnormalities on karyotyping was low (2/48, 4.2%) in their patients.[8]

CONCLUSION
Ovotesticular disease is rare, one should consider the possibility of Ovotesticular DSD, when managing patients with proximal hypospadias even if both gonads are palpable in scrotum. The diagnosis is on Histopathology. Majority of patients present late in their adulthood. Report suggest that karyotyping for all patients with proximal hypospadias to avoid late diagnosis is controversial and remains unecessary.

Foot Notes
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REFERENCES