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Case Report

Ovotesticular Disorder of Sex Development with Normal Karyotype: A Rare Case Report

Reddy Purushotham[1], Vernekar Sunita[2], Choukimath S. M[3], Bhavikatti Bharati[3], Dr. Abhisha RS[4*]

[1]Professor and Head of the Department of Pathology, Karnataka Institute of Medical Sciences, Hubballi, Karnataka, India
[2]Professor of the Department of Pathology, Karnataka Institute of Medical Sciences, Hubballi, Karnataka, India
[3]Associate Professor of the Department of Pathology, Karnataka Institute of Medical Sciences, Hubballi, Karnataka, India
[4]MD Pathology, Karnataka Institute of Medical Sciences, Hubballi, Karnataka, India

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***Corresponding author:** Dr Abhisha RS
MD Pathology, Karnataka Institute of Medical Sciences, Hubballi, Karnataka, India

Abstract

Ovotesticular disorder of sex development (DSD) refers to the co-presence of testicular and ovarian tissue in one individual. Here we report a case of Ovotesticular Disorder of Sex Development in a 42 years old male, presented with abdominal mass and undescended testes who had a normal karyotype. Gonadal dysgenesis should always be kept a possibility in patient with undescended testis. Diagnosis relies on clinical findings, hormonal analysis, gonadal histology, chromosome analysis, and genetic testing.

Keywords: Gonadal dysgenesis, Karyotype, Ovotesticular disorder of sex development, undescended testes.

INTRODUCTION

Ovotesticular disorder of sex development (DSD), 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. It was formerly known as "true hermaphroditism," the rarest form among all disorders of sex development in humans [1]. The gonads in an Ovotesticular DSD are asymmetrical having both ovarian and testicular differentiation on either side separately or combined as an ovotestis. It is usually present in early childhood with ambiguous genitalia, although presentation in early adulthood is not rare. Late diagnosis of Ovotesticular DSD may have a severe psychological impact on the patients [2].

CASE HISTORY

A 42-years-old man, married since 15 years with no issues or children, presented with mass per abdomen. On examination the patient was a phenotypically developed male with incompletely developed genitalia. Physical examination revealed an apparently healthy male weighing 50 kg and 158 cm tall. The amount of facial and axillary hair was sparse. Breast development was normal. Examination of genitalia revealed an empty scrotum and well-developed penis.

Transscrotal ultrasonography showed no gonads in the scrotal sac. Abdominopelvic ultrasonography (US) depicted a large abdominopelvic solid cystic mass on right side, measured 12.4x11.7x5.4cm, with multiple septations. The solid component showed internal vascularity. Subsequently, Computerised Tomography (CT) study of abdomen and pelvis demonstrated a large mixed cystic solid heterogeneously enhancing retroperitoneal mass with multiple internal septations measured 9x17x16cm was noted displacing bowel loops anteriorly and abutting the infrarenal aorta, IVC and iliac vessels, no obvious infiltration. Prostate was normal. Patient underwent exploratory laparotomy and specimen was sent for histopathological examination.

Gross examination showed cystic right gonad, rudimentary uterus, left gonad and lymph nodes. Right gonad with spermatic cord measured 10x9.5x5cm. Externally it was cystic grey brown to grey white and congested. On cut section, multiloculated cysts, drained seromucinous fluid. Cyst wall thickness varies from 0.5cm to1.2cm. Specimen of uterus measured 8.5x2x1. 2cm.Externaly atrophic, on cut surface endometrial

thickness was 0.1 cm. Fallopian tube measured 3cm in length. Externally, unremarkable and on cut section lumen was patent. Left gonad measured 4x2.5x1cm. Externally unremarkable and string test was positive.

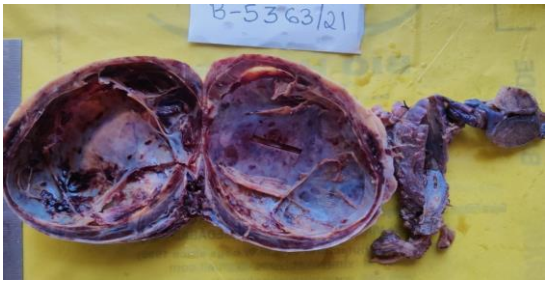
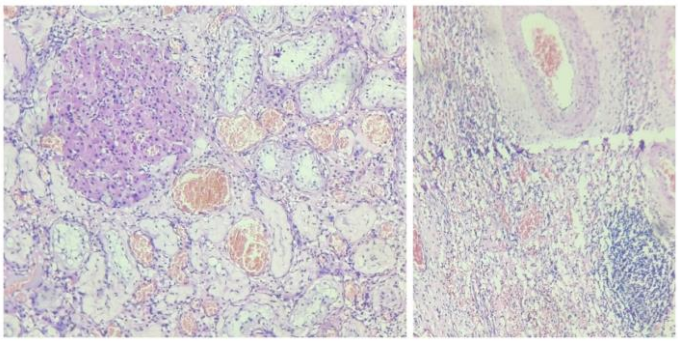


Figure 1: Gross image of Multi-loculated cystic right gonad, rudimentary uterus and left gonad

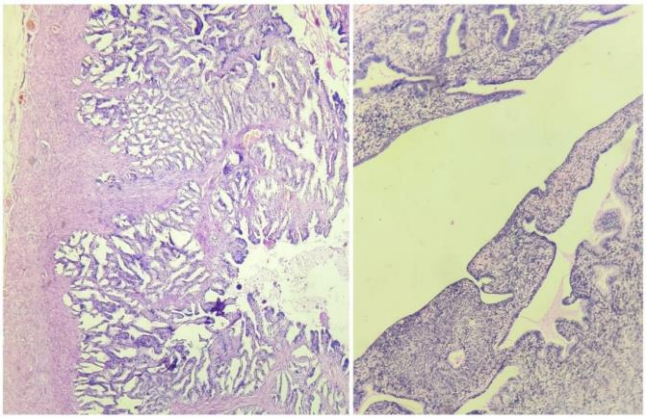
The left gonadal histology revealed testes with sclerosed seminiferous tubule, primary spermatogonial cells was showing maturation arrest. Interstitum showed Leydig cell hyperplasia. Section also showed epididymis. Section studied from uterus showed endometrial cavity lined by proliferative types of glands and myometrium was unremarkable. Section studied from cystic mass showed ovarian stroma with marked oedema with congestion. Section studied from fallopian tube like tubular structure showed reticular arrangement lined by proliferative columnar cells with round nuclei with fine chromatin. Interstitial stroma showed lymphocytes and fibroblast. Stroma showed Sertoli cell differentiation.



A **B**

Figure 2: A. Testes with sclerosed seminiferous tubule, primary spermatogonial cells was showing maturation arrest. Interstitum showed Leydig cell hyperplasia (H & E, 200X). B. ovarian stroma with marked oedema with congestion (H & E, 200X)

Ovotesticular Sex Development with atrophic testes with Leydig cell hyperplasia, Uterus with proliferative endometrium, adnexal adenomatoid tumour and lymph nodes showed reactive lymphadenitis. Based on the specific histological findings as well as highly supportive imaging findings, the patient was diagnosed as having Ovotesticular DSD with adenomatoid tumour.



A **B**

Figure 3:A. Adenomatoid tumour-reticular arrangement lined by proliferative columnar cells with round nuclei with fine chromatin. Interstitial stroma showed lymphocytes and fibroblast (Haematoxylin & Eosin, 100X).B. endometrial cavity lined by proliferative types of glands (Haematoxylin & Eosin, 200X).

On laboratory investigation, Alpha Fetoprotein (AFP) was 1.23 IU/ml and Lactate Dehydrogenase (LDH) was 1449U/L. The cytogenetic analysis of the karyotype was showed the genotype 46, XY.

DISCUSSION

Ovotesticular DSD is the rarest form of DSDs and infant is born with the gonads of both sexes, may present any combination of ovary, testes, or combined (ovotestes). The gonad may also appear as a streak gonad [1].

Gonadal differentiation is the embryonic process during which the bipotential gonad differentiates as either testes or ovaries. These developemental pathways are driven by a complex interplay of antagonistic genes. Unscheduled variations in the spatiotemporal expression or dosage of these genes can lead to simultaneous activation of both pathways and hence Ovotesticular development. The karyotype is almost always 46, XX, or rarely 46, XX/46, XY (chimerism) or 46, XX/47, XXY. Thus, the condition represents a spectrum of phenotypes and is characterized by variable penetrance [3]. The diagnosis is most often made in the first months of life, following investigations for the presence of atypical genitalia. Sex assignment may be very challenging and gender outcome largely unpredictable. If left untreated, the testicular and ovarian fractions both produce sex steroids at puberty, leading to phallic growth and breast development [3].

Ovotesticular DSD can be classified according to the position [2]:

i. **Lateral:** testis and a contralateral ovary (30%).
ii. **Bilateral:** both testicular and ovarian tissue (ovotestis) (50%).
iii. **Unilateral:** ovotestis on one side and a testis or ovary on the other side (20%).

Our case was of lateral type. 65% of the ovotestes are accompanied by a fallopian tube while a vas deferens accompanies the rest. The development of a uterus can occur in the presence of an ovotestes/ovary combination [2]. In normal male embryos, anti-Mullerian hormone (AMH) secreted by the pre-Sertoli cells causes rapid regression of the Mullerian ducts between the 8th and 10th weeks [4]. In our case, remnants of the Mullerian duct structure, rudimentary uterus, and fallopian tube were identified. In Ovotesticular disorder with 46XX chromosome carries a risk of malignancy (3%), and is higher (25%) in cases of 46XY chromosome. Many germ cell tumours have been reported till date such as gonadoblastoma, seminoma, dysgerminoma, cystadenoma, Sertoli cell tumour, and teratoma. Ovotesticular DSD with adenomatoid malformation is the first case to be reported after intrinsic literature search. Such patients of Ovotesticular disorder require a multidisciplinary approach of management, including psychological counselling and lifelong hormonal therapy, with fertility approaches and functional improvement [5]. In our case the patient was referred to psychiatric department for counselling during surgical follow up.

CONCLUSION

Ovotesticular disorder is diagnosed with a combination of tests including chromosome and genetic analysis, hormone testing, ultrasound or MRI, and gonadal biopsy. Radiological investigations are helpful in anatomical disorder, but confirmatory diagnosis for it is based on histopathological appearance, to differentiate it from mixed gonadal dysgenesis and other DSDs. The management must be carried out by a multidisciplinary team including psychological counselling and lifelong hormonal therapy, with fertility approaches and functional improvement.

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