

A Case of True Hermaphrodite Presenting as Cyclical Hematuria

Soumendra Mishra¹, Suchandra Ray²

¹PGT, Department of Pathology, IPGMER and SSKM Hospital, The West Bengal University of Health Sciences, Kolkata-700020, India

²Associate Professor, Department of Pathology, IPGMER and SSKM Hospital, Kolkata-700020, India

Abstract: True hermaphrodite (also known as ovotesticular disorder of sexual development or ovotesticular-DSD), is one of the rare varieties of disorder of sexual development. It is characterized by histologically confirmed both ovarian and testicular tissue in one individual. Here we report the case of a 16-year-old phenotypic male with 46, XX genotype (true hermaphrodite) presenting with cyclical hematuria and histologically diagnosed as ovotestis.

Keywords: True hermaphrodite, ovotesticular DSD

1. Introduction

Ovotesticular disorder of sex development (DSD) is a rare disease [1]. Most common presenting feature in these cases is genital ambiguity [2]. However, the phenotype may vary from normal female to normal male in appearance. Here we are describing a case of ovotesticular DSD, who presented with a complaint of cyclical hematuria.

2. Case Report

A sixteen-year-old male patient presented at the endocrinology out-patient department of our institute with complaint of cyclical hematuria for 4-5 days duration for one and a half months. Tanner staging of the patient was B5-P4-A1. External genitalia had hypospadias with chordee, with a single opening. Testis was not palpable. There was complete scrotal fusion without any pigmentation. Other physical parameters were within normal limits. He was born out of non-consanguineous marriage and the antenatal history was uneventful. His mother had a history of two previous stillbirths, with no history of virilism or intake of any offending drug.

The karyotype of the patient was 46, XX without any other genetic abnormalities. Ultra-sonographic findings revealed uterus with bilateral gonad like structures. No prostrate was found. 17-OHP and testosterone levels were estimated at 2.88 ng/ml and 4.48 mmols/ml respectively.

The patient subsequently underwent surgery. Surgical findings revealed unicornuate uterus with fallopian tubes and gonads on left side. On the right side rudimentary tube and gonad were found near the deep ring. The persistent mullerian structures were excised and sent to us for histopathology.

3. Histopathological findings

3.1 Gross Findings

Uterus measured 5cmx4cmx3cm with attached fallopian tube measuring 4cm and the left gonad measuring

3cmx2cmx1cm. The right sided gonad sent separately measured 3cmx3cmx2cm with the attached fallopian tube measuring 4cm in length. On cut section the right side gonad was partly solid and partly cystic.



Figure 1: Gross image of the right sided gonad, cut opened, showing partly solid and partly cystic areas

3.2 Microscopic Findings

The sections from the endomyometrium showed tubular endometrial glands with extensive areas of hemorrhage. The sections from the fallopian tube of the left side showed distended lumen filled with blood clots while the tube on the right side was unremarkable. The sections from the gonads on both sides show the presence of seminiferous tubules as well as ovarian stroma and follicular cysts. The overall histological features were in keeping with the diagnosis of true hermaphroditism.

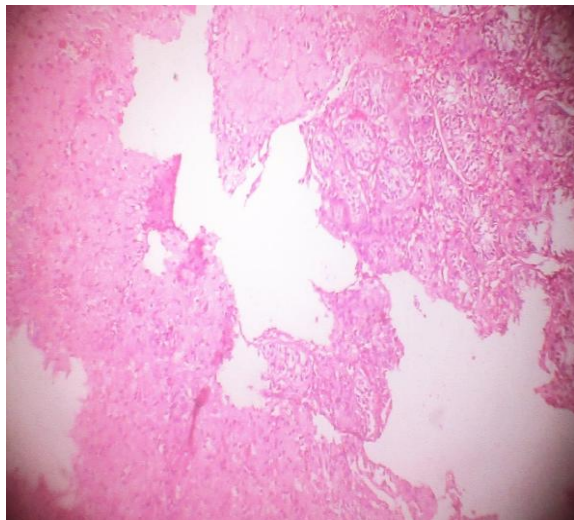


Figure 2: Figure showing seminiferous tubules in the section of right gonad

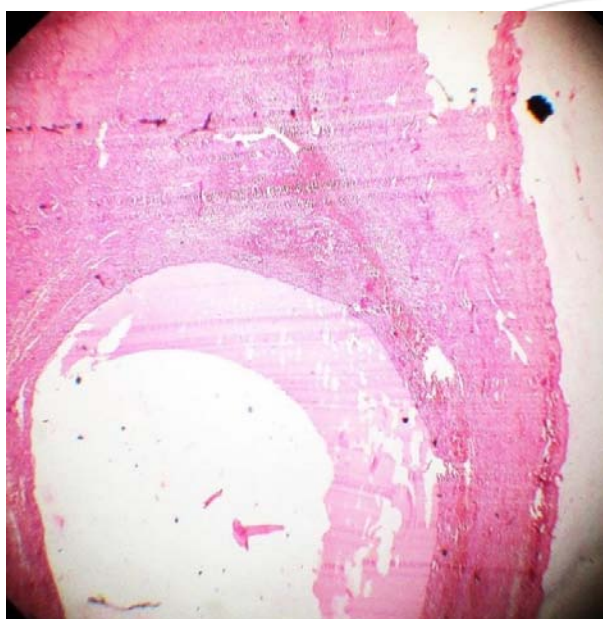


Figure 3: Figure showing ovarian stroma and luteal cyst in the same section of the right gonad

4. Discussion

Ovotesticular diseases of sexual differentiation (Ovo-DSD) was previously known as true hermaphroditism [3]. The term 'hermaphrodite' is derived from the term Hermaphroditus, the Greek God with bisexual attributes, who was the only child of Hermes, the God of athletics and Aphrodite the Goddess of love [4].

Ovotesticular disorder is a rare condition characterized by mixed ovarian and testicular tissue. The exact cause of true hermaphroditism still eludes human knowledge. Few mechanisms that might explain the testicular development include the translocation of the testis determining genes from the Y-chromosome to the X-chromosome or an autosome along with autosomal dominant mutations that promote testicular development in the absence of the Y-chromosome [5].

Ovotesticular DSD may present as bilateral ovotestis or an ovotestis on one side and contralateral ovary or testis. Majority of the patients of ovotesticular DSD have a 46,XX karyotype. However, 7% of patients of ovotesticular DSD are seen to have 46 XY karyotype and 10 to 40% exhibit chromosomal mosaicism. [6]

It has also been observed that gonads containing testicular tissue are located most frequently on the right, normal ovaries are located most frequently on the left [6], though in our case both the gonads were ovotestis. Usually both müllerian and wolffian genital structures are present and one seem to correspond with the adjacent gonads. Though in most cases there is presence of vagina, the uterus can be present and functional, hypoplastic, vestigial or altogether absent [6]. In our case described here, there is absence of cervix, uterus present was unicornuate and hypoplastic. The phenotype and external genitalia development is the reflection of the level of androgen production and reflection and can vary widely from ambiguous genitalia to isolated hypospadias. [7]

The following factors come to play while assigning a sex of rearing in all patients affected with DSD. [8].

- Karyotype
- Appearance of the external genitalia
- Types of the gonads present
- Need for surgical intervention to provide consistency with the sex of rearing of the preferred phenotypic sex
- Use of hormonal therapy
- Potential for fertility
- Parental wishes and parental cultural beliefs

Management of DSD is multidisciplinary with recent emphasis being placed on the use of the prescribed guidelines and consideration of the cultural background and parental input in decision making. [9]

- Surgical reconstruction of external genitalia depends on final sex assignment with an aim to achieve cosmetic, functional and sensitivity improvements. [10]
- Patients of DSD seem to have a slightly higher potential for gonadal malignancies like seminoma and dysgerminoma. The risk is more in the 46 XY genotype and undermasculinization. [11]

5. Footnotes

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Conflict of interest: None declared.

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Author Profile



Dr. Souendra Mishra received his MBBS degree from the West Bengal University of Health Sciences in 2012. Thereafter he joined as Pathology PGT at Institute of Post Graduate Medical Education and Research and SSKM Hospital, Kolkata under The West Bengal University of Health Sciences, Kolkata, India in 2015.



Dr. Suchandra Ray received her MBBS degree from the Burdwan University, India in the year 1984. She was awarded with the degree of Diploma in Gynaecology and Obstetrics in 1991 and MD in Pathology in 2004, both from the Calcutta University, India. She joined the West Bengal Medical Education Services, and has been posted as Associate Professor in the Department of Pathology, Institute of Post Graduate Medical Education and Research and SSKM Hospital, Kolkata for the last 6 years.