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An ovulating follicle presenting as a testicular mass in a teenage patient with ovotesticular DSD

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1. Introduction

Disorders of sexual differentiation (DSD) occur when an incongruity exists between a child's external genitalia, gonadal histology and chromosomal sex. Ovotesticular DSD (ODSD) represents 5% of DSD.[1] In an ovotestis, the testis is always central and the ovary polar. There are three anatomic variants:

- Bilateral: Testis and ovary bilaterally, either separate or united as an ovotestis (30%)
- Unilateral: Testis or ovary on one side and a contralateral ovotestis (50%)
- Lateral/Alternating: Testis and contralateral ovary (20%)[1,2]

We report a child with histologically documented Alternating ODSD who was later reclassified as having Unilateral ODSD after an ovarian remnant produced a corpus luteum cyst.

2. Case presentation

2.1. Initial presentation

A term infant with penoscrotal hypospadias and a right undescended testicle underwent genetic workup revealing 46XX/46XY chimerism (Fig. 1). Cystogram showed a smooth, adequate capacity bladder with a blind ending prostatic utricle. Pelvic ultrasound confirmed the presence of a prostatic utricle and identified a normal appearing left testis in the dependent hemiscrotum with no gonad in the right hemiscrotum.

At 12 months, diagnostic laparoscopy revealed a vas and well-developed vascular supply exiting a closed internal inguinal ring on the left. Contralaterally, an orange appearing gonad with a central darker cystic area and a tubular structure resembling a fallopian tube traveling towards a unicornuate uterus were observed. Right gonad frozen section revealed normal ovarian tissue. A left transscrotal incision was made, revealing a 3.5 cm gonad consistent with normal testis and epididymis. It was noted to have a whitish appearance anteriorly with a darker appearance at the superior pole. Deep longitudinal biopsy frozen section revealed normal prepubertal testis in both segments (Fig. 2). Following extensive discussion with pediatric endocrinology and cytogenetics, the family elected to continue to raise the child as male.

A right salpingo-oopherectomy and hysterectomy were performed. Pathology demonstrated a normal ovary and fallopian tube with a hypoplastic uterus and normal endometrium for age. Left gonad final pathology confirmed a normal prepubertal testis.

At 18 months, he underwent an uncomplicated preputial island flap hypospadias repair. After routine post-operative evaluations, he was followed by pediatric endocrinology annually.

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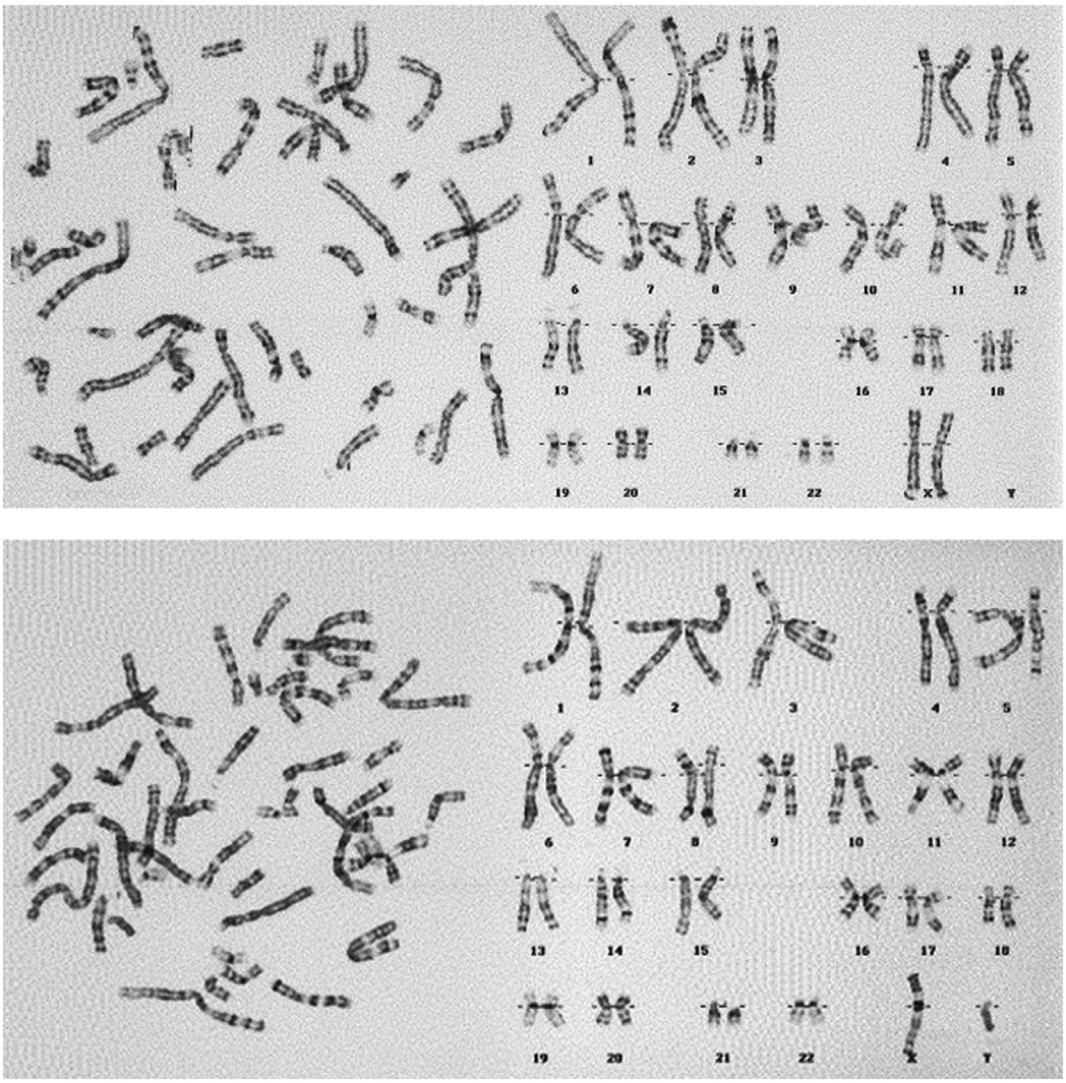

Fig. 1. Genetic workup revealing 46XX/XY chimerism.

2.2. Post-pubertal presentation

The patient returned at age 17 with complaints of a 2-week painless scrotal mass. His testosterone level was 500 ng/dL at age 14.5 years (normal 300–1000) but his FSH was elevated at 30.2, suggesting Sertoli cell impairment (normal 2–12). He was Tanner Stage V with a normal male phallus. His right hemiscrotum was empty and the left testicle was palpable in the left hemiscrotum. There was a mass on the superior aspect of the testicle, making it appear bilobed. No gynecomastia was appreciated.

Scrotal ultrasound demonstrated a 2.3 cm cystic lesion in the superior portion of the testicle with compressed testicular parenchyma along the margins of the cystic lesion. The remainder of the testicle appeared normal. AFP and βHCG were both normal.

The patient was scheduled for a partial vs. radical orchiectomy. On the day of presentation, he reported mass resolution. Intraoperative ultrasound showed the cyst had decreased in size to 1.5 cm. We performed a partial orchiectomy. Pathology demonstrated normal ovarian tissue. Tissue adjacent to the cyst was biopsied and pathology demonstrated normal testicular tissue (Fig. 3). He has done well following his partial gonadectomy.

3. Discussion

Gender assignment in patients with DSD is of critical importance. Karyotyping and histologic confirmation of testicular and ovarian tissue help guide gender assignment, medical therapy and surgical reconstructive options. Ninety percent of patients with DSD present at birth with external genitalia abnormalities, including microphallus, hypospadias, urogenital sinus, or cryptorchidism. The rest have a post-pubertal presentation with gynecomastia, hematuria, inguinal hernias, primary amenorrhea, and/or infertility.[3]

In ODSD, ovotestes are the most frequently encountered gonad (44%), followed closely by ovaries (33%).[4] Gonadal position is typically asymmetrical with ovotestes that are predominately testis descended below the inguinal ring and predominantly ovarian usually above. Not surprisingly, Wolffian structures are noted with a testicle and Mullerian structures next to an ovary the majority of the time.[5]

The most common karyotype in ODSD is 46XX (70%); genetic chimerism with 46XX/46XY in 20%. Genetic aspects leading to this disorder remain largely unknown. Gene mapping to Yp1A1, which

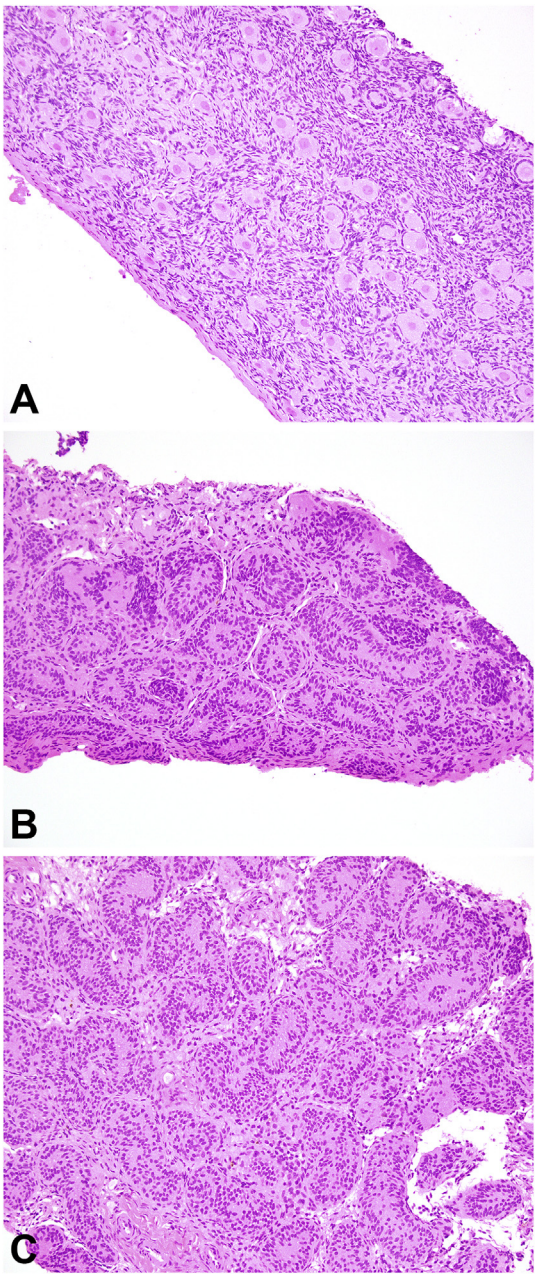

Fig. 2. Pathology slides from the initial operation. 2A: Right gonad demonstrating ovarian tissue at 20X. 2B: Left lower gonad demonstrating testicular tissue at 20X. 2C: Left upper gonad demonstrating testicular tissue at 20X.

determines testicular development, has been found in males with 46XX/46XY karyotype. This gene may be translocated in 46XX individuals with ODSD.[3]

A 22-year old phenotypic male presenting with an intratesticular mass comprised of ovarian tissue has been reported.[5] Our case is truly unique as our patient had histologically documented Alternating ODSD as a newborn. Despite the apparent absence of ovarian tissue in the descended gonad, he was later found to have an ovarian remnant when the ovarian tissue formed a corpus luteum cyst, thus changing his classification to Unilateral ODSD. This emphasizes the important point that gonadal biopsies in patients with DSD may be prone to sampling errors.

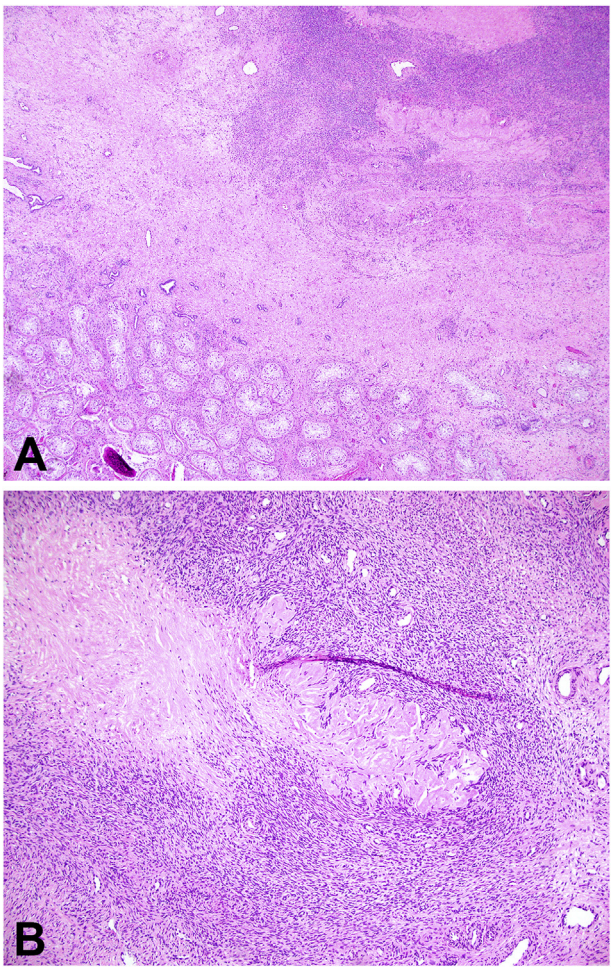

Fig. 3. Pathology slides from post-pubertal operation. 3A: Left gonad demonstrating both testicular and ovarian tissue at 4X. 3B: Left gonad demonstrating corpus albicans, the regressed form of the corpus luteum.

4. Conclusion

ODSD has a spectrum of phenotypic, histologic and karyotypic findings. Careful documentation of gonadal histology with longitudinal biopsies is critical to informed gender assignment and management of retained gonadal tissue. Our case underscores the need for continued vigilance of gonadal tissue as patients mature through puberty because despite appropriate gonadal biopsy, small areas of ovarian or testicular tissue can be missed.

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