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Ovarian malignancy in an individual with 46,XY ovotesticular disorder of sexual development – A case report

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ABSTRACT

Disorders of sexual development (DSD) are diseases resulting from aberrations in sex chromosomes, gonadal, and internal/external genitalia development resulting in various phenotypes. Ovotesticular DSD represents a rarer entity in this classification of disorders characterized by simultaneous presence of testicular and ovarian tissue. Gonadal tumors in those with DSDs is a known risk, although ovarian masses discovered in adults with ovotesticular DSD is a rare entity and there is little literature pertaining to this population. We present a case of an incidental adnexal mass discovered in an elderly patient ultimately elucidated as a malignant ovarian mass.

1. Introduction

Disorders of sexual development (DSD) comprise a variety of anomalies which arise from discordance in the chromosomal sex and the development of the gonads and internal/external genitalia. Ovotesticular DSD is a rare entity seen in humans which is characterized by the simultaneous presence of ovarian and testicular tissue with potential genotypes of 46 XX, 46 XY, and mosaicism. This rare disorder has an estimated prevalence of less than 1/20,000 with roughly 500 cases reported.[1] Furthermore, individuals with DSD have an elevated risk of developing certain types of malignancies compared to the general population but varies greatly among the specific subtypes of the disorders of sexual development.[2] We report a case of a 71 year old with 46 XY ovotesticular disorder of sexual development presenting with an incidental ovarian mass.

2. Case presentation

A 71 year old patient presented to our urologic department following a CT scan revealing endometrial thickening and an adnexal cystic mass. Past medical history is significant for ambiguous genitalia and was considered a true intersex individual as he was born with a phallus and vagina. Throughout childhood he underwent hypospadias repair, bilateral orchiectomy, and testicular prosthesis placement with the intention of constructing a male external genitalia. During this time, he was receiving 200mg testosterone injections every 3 weeks.

Abdominal CT (Figs. 1 and 2) revealed endometrial thickening and a 35mm adnexal cystic mass. Subsequent MRI showed diffuse thickening of the endometrium consistent with adenomyosis plus a 24.5mm right ovarian cystic mass. The right ovarian cyst demonstrated a 16mm enhancing solid nodule along the lateral aspect of the ovarian cyst. He was later seen by a geneticist and subsequent karyotype revealed 46,XY with SRY positivity.

He was lost to follow up for many months due to social circumstances. Repeat MRI showed stable endometrial thickening and enlargement of the right ovarian mass with the ovarian mass anterior to the uterus. The ovarian lesion measured 53mm × 35mm with the largest soft tissue component being 21mm × 32mm.

Tumor markers revealed negative testicular markers but elevated CA-125 of 504.5. Through a multidisciplinary approach with gynecologic oncology and subsequent exploratory laparotomy the adnexal mass and diffuse carcinomatosis of the entire abdomen was appreciated. Pathologic evaluation was consistent with ovarian malignancy. He elected for chemotherapy but unfortunately passed 15 months following initial evaluation of the mass.

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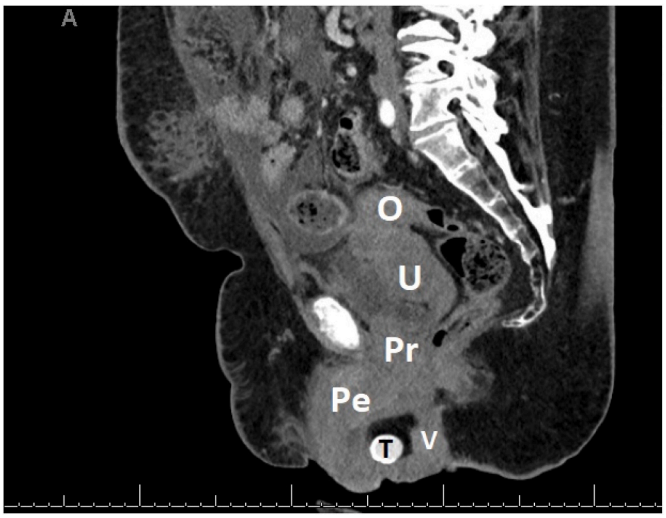

Fig. 1. Lateral CT demonstrating ovarian mass (O), uterus (U), prostate (Pr), penis (Pe), testicular implant (T) and vagina (V).

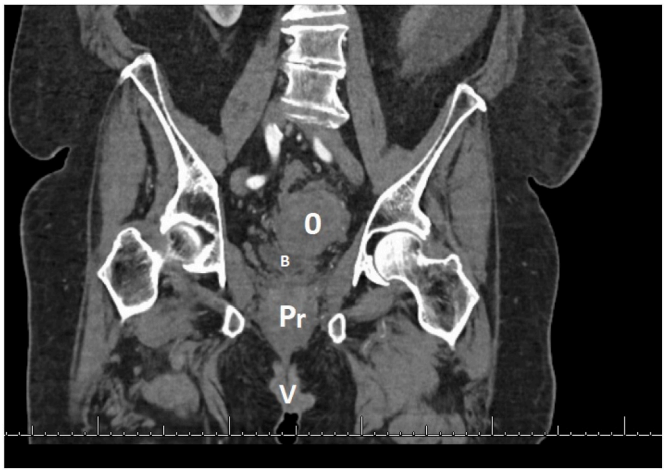

Fig. 2. Coronal CT demonstrating prostate (Pr), vagina (V), bladder (B), and ovarian mass (O).

3. Discussion

3.1. Differences of sexual development

DSDs are defined by a discordance between the chromosomal sex, external/internal genitalia, and the gonads and is to replace the term intersex.[2] The Chicago Consensus (2006) proposed a new nomenclature that previously used terms including hermaphrodite and intersex into a new categorical classification of sex chromosome DSD, 46 XX DSD, and 46 XY DSD.[2] In general, 46,XX DSD includes virilized females, 46,XY patients include those with aberrations in testicular development and/or function, and sex chromosome DSD includes Turner Syndrome, Klinefelter Syndrome, and 45,X/46,XY gonadal dysgenesis.[3] Ovotesticular DSD is the rarest DSD and comprises roughly 10% of DSDs with various karyotype possibilities and frequencies.[2] Ovotesticular DSD commonly presents as a male phenotype with hypospadias, asymmetric gonadal placement, and variable internal genitalia.[2]

3.2. Cancer risks

Development of certain types of tumors are known to occur in patients with DSDs but are less common in the general population. These include seminomatous and nonseminomatous germ cell tumors (GCT) that most commonly arise from a precursor lesion.[4] DSDs can be stratified into their risk of developing GCTs as high risk (i.e. Frasier syndrome, Denys-Drash Syndrome), intermediate risk (i.e. 17β-HSD) and low risk (i.e. ovotesticular DSD) with great variation in cancer risk (1%–60%).[4] Ovarian masses in those with ovotesticular DSD has been reported, but is rare, thus literature on this topic is minimal.[5] Therefore, it is difficult to ascertain if this ovarian mass arose from underlying factors associated with a DSD. It is of importance to further investigate any potential relationship between DSDs and ovarian masses in adults.

4. Conclusion

Ovotesticular DSD with 46,XY genotype is a rare entity in isolation, but an ovarian mass seen with this disorder is a clinical entity that is exceedingly rare. An association between ovarian masses and ovotesticular DSD has not been elucidated and there is sparse literature about adnexal masses and DSDs and whether these masses tend to be benign or malignant. Due to the potential implications of these on patient health, it would be beneficial to elucidate any potential relationship between the two entities.

CRediT authorship contribution statement

James I. Griggers: Writing – original draft, Writing – review & editing. **Robert Higgins:** Investigation, Resources. **Martha K. Terris:** Conceptualization, Data curation, Investigation, Resources, Validation, Writing – review & editing.

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