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Ovarian steroid cell tumor, not otherwise specified, associated with congenital adrenal hyperplasia: rare tumors of an endocrine disease

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Abstract Ovarian steroid cell tumors, not otherwise specified (OSCTs), are extremely rare and present a diagnostic challenge when evaluating an ovarian mass. We present a case of such a tumor in a patient with known Congenital Adrenal Hyperplasia (CAH), secondary to 21-hydroxylase deficiency, who was noncompliant with her medications. The workup, diagnosis, and treatment of this rare condition are described.

Testicular Adrenal Rest Tumors (TARTs) have been identified in adult male patients with Congenital Adrenal Hyperplasia (CAH) with a prevalence of 50%–95% depending on age and modality of diagnosis [1,2]. Ovarian steroid cell tumors, not otherwise specified (OSCTs), however pose a diagnostic challenge in the feminine gender. The current prevalence of OSCTs is ≤ 0.1% of all ovarian tumors, which is significantly less compared to the prevalence of TARTs [3]. This discrepancy in diagnosing adrenal rest tumors between the two genders necessitates further elucidation and query.

These tumors have been originally viewed to arise from the adrenal rests, which remain entrapped within the ovarian parenchyma during human ontogenesis. The urogenital ridge is formed by condensation of coelomic mesoderm and gives rise to kidneys, gonads, and adrenals. Given the close proximity of the embryological adrenal gland and gonads, ectopic adrenal gland can be seen along the pathway of gonadal gland migration or at the final destination of the gonads. This migration compounded with syndromes leading to high levels of Adrenocorticotropic Hormone (ACTH) results in excessive stimulation of the adrenal tissue, thereby leading to hypertrophy and organ enlargement of both the adrenal glands and the gonadal glands containing adrenal tissue. Syndromes associated with ACTH hypersecretion such as Addison's Disease, Nelson's Syndrome and poorly controlled CAH are common etiologic factors in the formation of adrenal rest tumors [4,5].

CAH is an autosomal recessive disease seen most commonly as a result of defects in one of several enzymes

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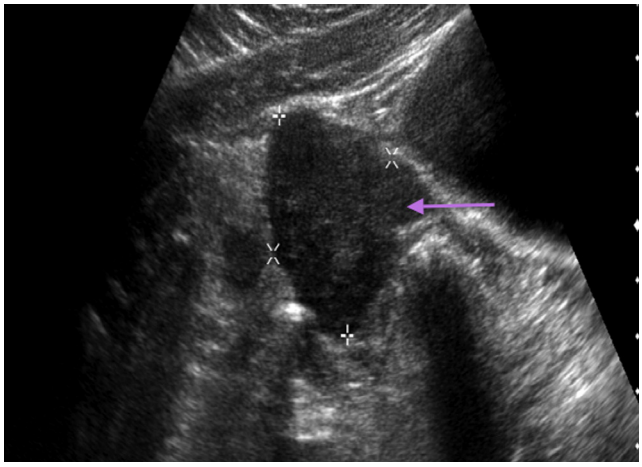

Fig. 1 Ultrasound Left ovary with 4.4 × 3.4 × 4.9 cm mass; Volume 45.2 cm^3 (arrow).

involved in the formation of cortisol from cholesterol. More than 95% of CAH is caused by 21-hydroxylase enzyme (21-OH) deficiency. The rest (5%) is commonly seen as a result of 11-beta hydroxylase deficiency [6].

In this interesting case report we present the clinical course, laboratory data, and imaging studies of a seventeen-year-old patient with CAH and resultant OSCT. The patient was poorly compliant with her medications and this was assumed to be a causative factor in the formation of her adrenal rest tumor. A systematic review of the available literature on OSCTs is also presented in this article. In the current literature, OSCTs are presented as either case reports or case series, due to the rare nature of these tumors.

1. Case study

We present the case of a 17-year-old female diagnosed with congenital adrenal hyperplasia at birth with documented 21-OH deficiency. The patient suffered from delayed onset of menses, virilization, and psychiatric issues as well, which mandated psychiatric liaison as a part of her ongoing care. Her testosterone and 17-hydroxy progesterone levels were found to be elevated throughout the majority of her care secondary to her non-compliance with medical therapy. They were >50 ng/dl (normal < 40 ng/dl), and > 5000 ng/dl (normal < 169 ng/dl) respectively. ACTH levels were normal at 138 ng/dl. The patient was being managed medically by her endocrinologist with both Dexamethasone and Florinef. Shortly after she had attained puberty, it was noted that the patient had not yet started with normal menstrual cycles. For further evaluation of her introitus and secondary to her history of having perineal surgery in the past, a urological evaluation was done. As part of this, serial ultrasounds of her kidney, bladder, and pelvic organs were done. The patient also underwent a genitogram which showed a well developed vagina, with narrowing at her introitus. The patient then underwent vaginoplasty along with serial dilatations, and examinations under anesthesia (EUAs) as part of her reconstruction.

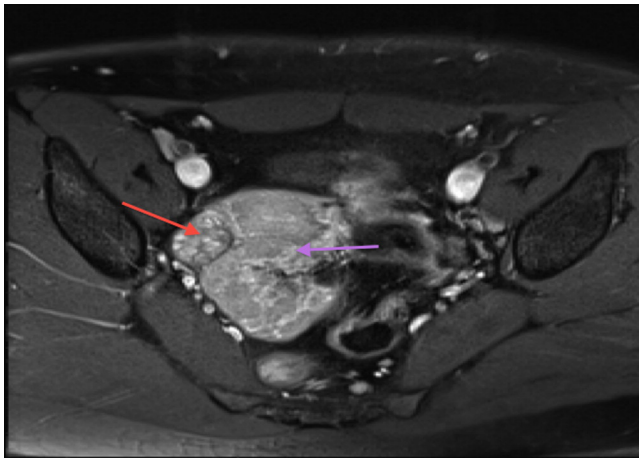

Fig. 2 MRI of pelvis, purple arrow at left ovarian mass, red arrow at right ovary.

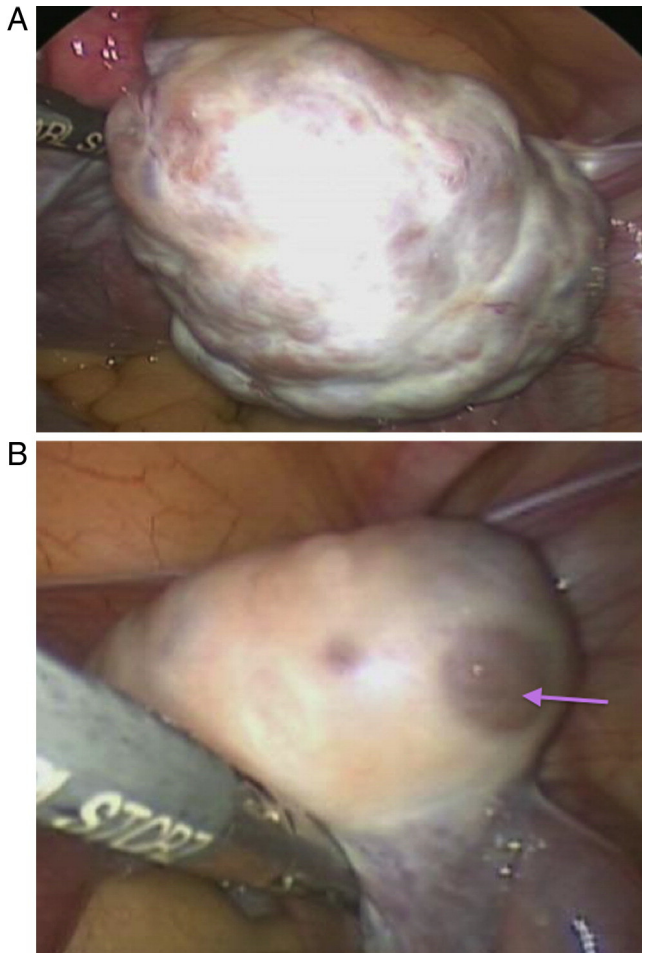

Fig. 3 A: Left ovary with heterogeneous mass; B: Right Ovary smaller mass (arrow).