ovarian tumor and fallopian tube favored a diagnosis of extra-adrenal paraganglioma as the immunohistochemical staining was positive for S-100, synaptophysin, and chromogranin. Retrospectively, the tumor was likely a non-functioning paraganglioma, as the patient underwent surgery without complications.

CONCLUSION

Paraovarian paraganglioma is an exceptionally rare entity that presents significant diagnostic challenges due to its atypical location and non-specific clinical features. This case highlights the importance of considering paraganglioma in the differential diagnosis of adnexal masses as the perioperative management may differ.

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46,XY DSD WITH RETAINED MÜLLERIAN STRUCTURES AND GENDER TRANSITION IN ADULTHOOD: A STEPWISE DIAGNOSTIC APPROACH

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INTRODUCTION/BACKGROUND

Disorders of Sex Development (DSD) are congenital conditions marked by atypical chromosomal, gonadal, or anatomical sex development. A structured diagnostic approach—starting from phenotype assessment through to chromosomal and molecular studies—is essential, particularly in 46,XY DSD where clinical presentations may vary widely. This report discusses a young adult with delayed-diagnosed 46,XY DSD who transitioned to male gender, analyzed through a stepwise framework.

CASE

A 20-year-old individual, assigned female at birth, presented with progressive virilization since early adolescence. The patient had no breast development or menstrual history. Instead, a deepened voice, facial and body hair, spontaneous erections, and wet dreams were reported. The patient urinated from an orifice beneath the clitoral area in a squatting position.

Physical examination revealed masculine features, gynecomastia, and clitoromegaly measuring approximately 3 cm in length. A bifid scrotum resembling labia majora was observed, with no palpable testes. Tanner staging was M2P4.

Hormonal analysis showed hypergonadotropic hypogonadism: LH 29.7 mIU/mL, FSH 47.8 mIU/mL, testosterone 21.1 nmol/L, estradiol 27.5 pmol/L. Karyotyping confirmed a 46,XY complement. MRI revealed bilateral gonads in the inguinal canals suspected as testes, and a uterine-like structure between the bladder and rectum. No ovaries or prostate were identified. FISH and SRY gene sequencing were performed; SRY was positive, and no pathogenic variants were found.

The presence of virilized phenotype, retained Müllerian structures, and undescended testes in a 46,XY individual suggests a disorder in androgen action or synthesis. While partial androgen insensitivity syndrome (PAIS) or 5α-reductase deficiency are possible, definitive diagnosis awaits further molecular studies such as SRD5A2 or AR gene sequencing.

CONCLUSION

This case illustrates the complexity of evaluating 46,XY DSD and emphasizes the utility of a stepwise diagnostic algorithm. Clinicians should remain vigilant to consider rare etiologies in late-presenting cases and provide multidisciplinary, gender-affirming care tailored to the patient's identity and needs.

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REFINING THE DIAGNOSIS: A CASE REPORT ON THE ROLE OF FISH IN DETECTING SUBTLE MOSAIC KLINEFELTER SYNDROME

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INTRODUCTION/BACKGROUND

Mosaic forms of Klinefelter syndrome (KS) can pose a diagnostic challenge, particularly in patients with a normal male phenotype and unremarkable hormonal profiles. While conventional karyotyping is a widely used first-line tool for detecting chromosomal abnormalities, its sensitivity