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.

EP_A159

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.

EP_A160

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

is limited in identifying low-level mosaicism, which may result in false-negative or misleading interpretations. Fluorescence in situ hybridization (FISH), offering higher resolution and the ability to analyze hundreds of interphase nuclei, can uncover subtle chromosomal abnormalities that significantly impact clinical decision-making.

CASE

A 34-year-old male presented with concerns about delayed pubertal development, minimal axillary and facial hair, and poor muscularity despite strength training. He reported onset of wet dreams at 16 years, with gradual deepening of the voice and scant body hair. Physical examination revealed a tall stature (187 cm), soft body habitus, mild gynecomastia, and bilaterally normal-sized testes (20 mL). Hormonal assays showed normal levels of testosterone (22.58 nmol/L), LH (3.6 mIU/mL), and FSH (4.0 mIU/mL). Initial chromosomal analysis revealed a mosaic 45,X[1]/46,XY[99] pattern, suggesting a potential diagnosis within the chromosomal disorders of sex development (DSD) spectrum. However, the discrepancy between the karyotype and the patient's unequivocally male phenotype prompted further investigation using FISH. Analysis of 300 nuclei revealed 6% 47,XXY cells and 94% 46,XY cells, confirming a diagnosis of mosaic Klinefelter syndrome. Semen analysis demonstrated severe oligoasthenoteratozoospermia with only 1% morphologically normal sperm.

CONCLUSION

This case illustrates the limitations of conventional karyotyping in detecting low-level mosaicism and underscores the diagnostic value of FISH in cases where clinical findings and cytogenetic results appear discordant. By providing higher sensitivity, FISH can uncover clinically significant mosaic patterns, facilitating accurate classification within the DSD spectrum and informing appropriate counseling and reproductive planning.

EP_A161

FUNGAL SHADOWS: DIAGNOSTIC AND MANAGEMENT CHALLENGES OF ADRENAL HISTOPLASMOSIS IN AN IMMUNOCOMPETENT ADULT

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INTRODUCTION/BACKGROUND

Histoplasmosis is a fungal infection caused by *Histoplasma capsulatum*. Disseminated histoplasmosis involving bilateral adrenal glands and resulting in adrenal insufficiency is rare, particularly in immunocompetent individuals. The non-specific symptoms often mimic other diseases, making timely diagnosis difficult, especially in resource-limited settings. We report a case of disseminated histoplasmosis with adrenal insufficiency in an immunocompetent individual presenting with bilateral adrenal masses.

CASE

A 69-year-old previously healthy male presented with generalized body weakness, intermittent fever and significant weight loss of 20 kg over three months. There were no other remarkable symptoms. Initial PET-CT scan revealed large bilateral adrenal masses with hypermetabolic rims and central metabolism (right: 8.3 x 6.6 x 8.8 cm; left: 8.6 x 6.6 x 9.7 cm) as well as hepatosplenomegaly and right lung changes suggestive of infection or malignancy. The investigations for tuberculosis and HIV were negative. Tissue biopsies of the masses revealed acute granulomatous lesions indicative of fungal infection. The patient was started on intravenous Amphotericin B, followed by maintenance therapy with oral itraconazole. An ACTH stimulation test showed inadequate adrenal response, and steroid replacement therapy was initiated. Despite nine months of antifungal therapy, he showed minimal clinical improvement. Repeat imaging demonstrated increased adrenal mass size, prompting bilateral adrenal drainage following a multidisciplinary team discussion. Histopathology confirmed ongoing fungal infection, and fungal sequencing identified Histoplasma capsulatum.