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

https://doi.org/10.15605/jafes.040.S1.169

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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.

## Adult E-Poster

A second course of IV liposomal Amphotericin B was administered, followed by itraconazole. The patient's fever resolved, and follow-up imaging showed reduction in adrenal mass size. He remains on drainage and long-term antifungal therapy, planned for at least 18 months.

### CONCLUSION

This case highlights the diagnostic challenges of adrenal histoplasmosis in immunocompetent individuals presenting with vague systemic symptoms and large bilateral adrenal masses. Early recognition and a multidisciplinary approach are crucial for timely diagnosis and optimal management.

## EP_A162

### BEYOND THE YELLOW: UNMASKING PHEOCHROMOCYTOMA IN A JAUNDICED PATIENT

*https://doi.org/10.15605/jafes.040.S1.170*

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### INTRODUCTION/BACKGROUND

Pheochromocytomas and paragangliomas (PPGLs) are rare neuroendocrine tumours (NET) arising from chromaffin cells. Bilateral pheochromocytomas are extremely rare, constituting 7–10% of all pheochromocytoma cases, and 60%–90% of them possess a germline mutation.

### CASE

A 17-year-old male presented initially with epigastric pain and obstructive jaundice. He is not hypertensive. ERCP revealed choledocholithiasis and a dilated common bile duct (CBD). A contrast-enhanced CT of the liver showed an enhancing CBD lesion causing biliary obstruction and incidental bilateral adrenal tumours. A CT Adrenal protocol confirmed a left adrenal lesion measuring 5.4×4.8x5.1cm with unenhanced attenuation 35.4 Hounsfield Units (HU) and a right adrenal lesion measuring 1.2 × 1.1 × 1.6 cm with unenhanced attenuation 30.7HU, both with delayed contrast washout, consistent with pheochromocytomas. Biochemical evaluation showed elevated 24-hour urinary normetanephrine at 18,558 nmol/24h (497–2489), which is seven times the upper limit of normal, confirming catecholamine excess with normal levels of Metanephrine. Other hormonal investigations were unremarkable. He underwent open cholecystectomy and choledochectomy with biliary reconstruction. Histopathology confirmed a well-differentiated Grade I neuroendocrine tumour (NET) of the CBD, positive for synaptophysin, chromogranin A, and CD56, with a Ki-67 <3%. Surgical margins and lymph nodes were negative. Thyroid ultrasound was normal. $^{68}$Ga-DOTATATE Positron Emission Tomography (PET) confirmed bilateral pheochromocytomas with no extra-adrenal paraganglioma or metastatic disease. He underwent bilateral adrenalectomy after adequate alpha-blockade and was discharged well with hydrocortisone and fludrocortisone replacement. He is awaiting genetic testing.

### CONCLUSION

This case highlights a rare pheochromocytoma with obstructive jaundice, lacking the classical triad of headache, palpitations, and sweating. Bilateral pheochromocytomas are commonly seen in Multiple Endocrine Neoplasia types 2A and 2B, von Hippel–Lindau disease, and rarely with MAX and TMEM127 mutations, though they can also occur sporadically. Genetic testing is crucial for diagnosing and managing bilateral pheochromocytoma, as it aids in treatment decisions, recurrence prediction, and family screening.

## EP_A163

### TREACHEROUS JOURNEY OF ADVANCED PAPILLARY THYROID CARCINOMA IN PREGNANCY

*https://doi.org/10.15605/jafes.040.S1.171*

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### INTRODUCTION

Papillary thyroid carcinoma (PTC) is the most common thyroid malignancy and generally exhibits a favourable prognosis, but it can manifest with metastasis in advanced stages. Pregnancy complicates the management of such cases particularly when radioactive iodine (I-131) therapy is indicated.

### CASE

A 26-year-old presented in her second trimester with acute exacerbation of bronchial asthma requiring mechanical ventilation. During intubation, a 6 × 4 cm anterior neck swelling was found. A computed tomography showed diffuse heterogeneous thyroid enlargement with tracheal narrowing, cervical lymphadenopathy, and pulmonary