Given the rarity of DNMT3A-mutated PPGL (<1% of cases), this case report emphasizes the necessity of comprehensive molecular profiling in advanced disease to refine risk stratification and guide the development of precision-based palliative management in rapidly progressive cases.

EP_A016

CONGENITAL ADRENAL HYPERPLASIA WITH HYPOGONADISM IN A MAN: 3β-HYDROXYSTEROID DEHYDROGENASE DEFICIENCY VS. LIPOID HYPERPLASIA?

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INTRODUCTION
3β-Hydroxysteroid dehydrogenase (3β-HSD) deficiency and lipoid congenital adrenal hyperplasia (CAH) are rare disorders of steroidogenesis that result in impaired synthesis of all adrenal and gonadal hormones. Affected males typically present with ambiguous genitalia and concurrent mineralocorticoid and glucocorticoid deficiency. Distinguishing between these two conditions is crucial, as their management strategies differ.

CASE
We report a 24-year-old male, born to non-consanguineous parents, who was clinically diagnosed with 3β-HSD deficiency during infancy. He presented at day 40 of life with ambiguous genitalia, bilateral undescended testes, and generalized hyperpigmentation. His family history was significant for an elder brother with salt-losing CAH. Initial biochemical evaluation confirmed primary adrenal insufficiency and primary hypogonadism. Notably, his steroid precursors, dehydroepiandrosterone sulfate (DHEAS) and 17-hydroxyprogesterone, were suppressed. A karyotype confirmed 46,XY. In the absence of genetic testing at that time, a clinical diagnosis of 3β-HSD deficiency was made, and he was commenced on mineralocorticoid and supraphysiological glucocorticoid replacement. He underwent bilateral orchidopexy and hypospadias repair in childhood. Pubertal induction was required, followed by maintenance testosterone therapy. Upon transitioning to adult care, a review of his biochemical profile, particularly suppressed DHEAS, which is inconsistent with 3β-HSD deficiency, raised the suspicion of a more proximal defect, such as lipoid CAH. Differentiation is imperative, as lipoid CAH requires only physiological glucocorticoid replacement, whereas 3β-HSD deficiency often needs higher doses to suppress adrenocorticotropic hormone and DHEAS. To resolve this diagnostic uncertainty and guide long-term therapy, the patient was referred for genetic studies.

CONCLUSION
Differentiating 3β-HSD deficiency from lipoid CAH is important, as both have similar clinical presentation. The absence of elevated steroid precursors, particularly DHEAS, should raise suspicion of a more proximal defect. Given the different aims of glucocorticoid therapy, establishing a precise diagnosis through genetic studies is essential to guide clinical management and minimize the morbidity associated with supraphysiological corticosteroid dosing.

EP_A017

METASTASIS OR MIMIC? NAVIGATING THE WORKUP OF A LARGE ADRENAL INCIDENTALOMA IN THE SETTING OF LUNG CANCER

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INTRODUCTION
The identification of a significant adrenal mass in a patient without a biopsy-confirmed malignancy poses a diagnostic challenge: Is it a metastatic lesion or an underlying adrenal condition? Adrenal metastases are the second most common site of spread for lung adenocarcinoma; approximately 3–7% of adrenal masses represent benign adenomas. Diagnosis is even harder if there are signs of primary aldosteronism (PA).

CASE
We present a case of a 62-year-old Chinese female with a 10-year history of hypertension, managed on dual antihypertensive therapy, who presented for evaluation of suspected PA following the discovery of hypokalemia. Biochemical screening revealed an elevated aldosterone-renin ratio (ARR, 65). The overnight dexamethasone suppression test (20 nmol/L) and testosterone (0.79 nmol/L) were both within normal limits. Saline Suppression Test (SST) showed an indeterminate post-infusion aldosterone level (202.8 pmol/L). Cross-sectional imaging via computed tomography (CT) Adrenals identified a large, 6.4 × 5.4 × 6.2 cm heterogeneous left suprarenal mass with a low mean attenuation (8.6 Hounsfield Unit [HU]). Concurrently, an incidental left upper lobe pulmonary lesion was identified, and PET-CT was performed; the SUVmax of the lung was identical to that of the adrenal lesion. An ultrasound-guided biopsy of the pulmonary lesion confirmed estimated glomerular filtration rate-mutation-positive lung adenocarcinoma. The patient started on targeted therapy with dacomitinib. Follow-up CT imaging at 9 months demonstrated disease progression within the thorax,

marked by the appearance of a new pulmonary nodule. Notably, however, the adrenal mass remained stable in size and morphology.

CONCLUSION

The absence of mixed androgen/glucocorticoid hypersecretion, combined with the radiographically static nature of the mass, suggests a lower probability of adrenocortical carcinoma. This case highlights that while size is a major risk factor for adrenocortical carcinoma, it must be interpreted in conjunction with hormonal activity, HU, and growth patterns. Identifying these "mimics" helps avoid over-staging lung cancer and ensures patients receive targeted therapy instead of unnecessary adrenalectomies.

EP_A018

A HIDDEN DIAGNOSIS: DISSEMINATED HISTOPLASMOSIS MIMICKING TUBERCULOSIS IN THE ELDERLY

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INTRODUCTION

Histoplasmosis is a rare opportunistic, inhalation-acquired systemic mycosis caused by *Histoplasma capsulatum*, endemic to Southeast Asia, including Malaysia. Although classically seen in immunocompromised hosts, it is increasingly reported in immunocompetent individuals. Infection is associated with environmental exposures such as bat or bird droppings and soil disruption. Histoplasmosis is a progressive granulomatous disease that can closely mimic tuberculosis and malignancy, making diagnosis challenging.

CASE

An 87-year-old Malay male with a history of treated pulmonary tuberculosis presented with a 6-month history of intermittent fever, anorexia, weight loss, and hypotension. Initial computed tomography imaging demonstrated bilateral heterogeneous adrenal masses (right: 2.2 × 3.7 × 5.0 cm; left: 2.0 × 3.7 × 5.2 cm) with indeterminate washout characteristics. Biochemical adrenal evaluation, including a short Synacthen test, confirmed primary adrenal insufficiency. Extensive microbiological and malignancy workup, including bronchoscopy, cultures, and tumor markers, was non-diagnostic. In view of clinical deterioration and epidemiological risk, empirical antituberculous therapy was initiated; however, no clinical improvement was observed after 2 months. PET-FDG revealed intensely hypermetabolic bilateral adrenal masses (SUVmax right 16.7, left 13.2) with no other abnormal foci. Non-invasive fungal investigations were negative. Definitive diagnosis was established via computed tomography-guided adrenal biopsy, which demonstrated necrotizing granulomatous inflammation with intracellular yeasts, subsequently identified as *H. capsulatum*. The patient was treated with oral itraconazole and corticosteroid replacement, resulting in significant clinical improvement and planned interval radiological reassessment.

CONCLUSION

Disseminated histoplasmosis is a diagnostic challenge, particularly in frail elderly patients, where invasive procedures may be delayed. It can mimic tuberculosis and malignancy and may lack an identifiable exposure history. Non-invasive tests may be inconclusive, making tissue biopsy essential. Adrenal involvement may result in primary adrenal insufficiency, further complicating the clinical picture. Early consideration and timely confirmation are crucial for appropriate management.

EP_A019

OVERWHELMING OPPORTUNISTIC INFECTIONS AS THE INITIAL PRESENTATION OF SEVERE CUSHING SYNDROME

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INTRODUCTION

Severe hypercortisolism is associated with profound impairment of both innate and adaptive immune responses, predisposing affected individuals to opportunistic infections. Excess glucocorticoids alter leukocyte trafficking, suppress pro-inflammatory cytokine production, and impair cellular immunity, increasing susceptibility to bacterial, viral, and fungal pathogens. In some cases, severe infections may precede the diagnosis of Cushing syndrome and represent the initial clinical manifestation. Early recognition is important as untreated hypercortisolism can lead to substantial morbidity and mortality.

CASE

A young adolescent male presented with progressive facial fullness and facial hyperpigmentation for 4 months, followed by 1 month of intermittent fever, cough, lower limb weakness, and hallucinations. On examination, he was tachypneic with cushingoid features including moon facies, pigmented acne over the face and chest, and nail bed hyperpigmentation. He was hypertensive and had severe hypokalemia with lymphopenia.