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?

**Marisa Masera Marzukie[1], Quan Hziung Lim[1], Shireene Ratna Vethakkan[1], Jeyakantha Ratnasingam[1]**
*[1]Universiti Malaya Medical Centre*

**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

**Mohd Firdaus Mohamad Kamil[1], Masliza Hanuni Mohd Ali[1], Wan Mohd Hafez Wan Hamzah[1]**
*[1]Endocrinology Unit, Medical Department, Hospital Sultanah Nur Zahirah*

**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,