EP_A014

BILATERAL ADRENAL MASSES WITH RAPID DETERIORATION: A RARE CASE OF PRIMARY ADRENAL LYMPHOMA

Melody Tsen Shu Ling[1], Cheong Yee Weai[2], Hwee Ching Tee[2], Jin Hui Ho[2], Shireen Siow Leng Lui[2]
[1]Department of Medicine, Hospital Queen Elizabeth
[2]Diabetes & Endocrine Unit, Department of Medicine, Hospital Queen Elizabeth II

INTRODUCTION
Primary adrenal lymphoma (PAL) is a rare extranodal lymphoma, accounting for less than 1% of cases. It often presents with non-specific constitutional symptoms and features of adrenal insufficiency. The absence of pathognomonic findings frequently leads to delayed diagnosis, by which time the disease is often advanced, contributing to poor prognosis.

CASE
A 59-year-old male with type 2 diabetes mellitus and previously treated pulmonary tuberculosis presented with 1 month of lethargy, anorexia, weight loss, and abdominal pain. He was cachectic but hemodynamically stable (BP 120/81 mmHg; glucose 5.1 mmol/L), with otherwise unremarkable systemic examination. Laboratory evaluation revealed hyponatremia (120 mmol/L), hyperkalemia (6.0 mmol/L), and severe hypercalcemia (4.4 mmol/L). A short Synacthen test confirmed adrenal insufficiency.

Computed tomography demonstrated marked bilateral adrenal enlargement with bulky masses (8.9 × 8.7 × 8.7 cm right; 9.9 × 7.6 × 10.2 cm left), suggestive of malignant infiltration. Adrenal protocol imaging revealed indeterminate lesions with attenuation >40 Hounsfield Unit and relative washout <40%. Ultrasound-guided biopsy of the right adrenal gland was performed.

The patient was discharged on hydrocortisone replacement with plans for early follow-up. One week later, he presented again in adrenal crisis with severe hypoglycemia (1.1 mmol/L) and cardiovascular collapse. Despite resuscitative efforts, he succumbed. Histopathology subsequently confirmed diffuse large B-cell lymphoma, activated B-cell subtype, consistent with PAL.

CONCLUSION
This case highlights the diagnostic pitfalls of PAL, particularly when constitutional symptoms mimic chronic infections such as tuberculosis. It underscores the need for high clinical suspicion in patients with bilateral adrenal masses and adrenal insufficiency. Early recognition, adequate steroid replacement including mineralocorticoid therapy, and prompt tissue diagnosis are critical to prevent rapid deterioration and improve outcomes.

EP_A015

METASTATIC MALIGNANT PHEOCHROMOCYTOMA DRIVEN BY DNMT3A SOMATIC MUTATION

Vijayrama Rao Sambamoorthy[1], Zanariah Hussein[1], Anthony Louis Kindu[2]
[1]Endocrine Unit, Medical Department, Hospital Putrajaya
[2]Nuclear Medicine Department, Institut Kanser Negara

INTRODUCTION
Pheochromocytomas and paragangliomas (PPGL) are rare neuroendocrine tumors with high heritability. While most are benign, approximately 25% are malignant, defined by distant metastases. Molecular classification has identified three clusters, with Cluster 3 (Wnt-signaling) being exclusively somatic and associated with aggressive behavior. We report a rare case of metastatic malignant pheochromocytoma driven by a somatic DNMT3A mutation, highlighting its unique imaging characteristics and rapid clinical progression.

CASE
A 55-year-old female presented with paroxysmal hypertension, headache, and a 10-kg weight loss. Biochemical workup revealed markedly elevated 24-hour urine metanephrines (163 × ULN) and normetanephrines (47 × ULN). Imaging confirmed a 15-cm left adrenal mass with liver and widespread skeletal metastases. Functional imaging demonstrated a striking mixed avidity: liver metastases were predominantly fluorodeoxyglucose-avid (SUVmax 7.0), skeletal lesions showed high Ga-68 DOTATATE avidity (SUVmax 6.9), and the primary tumor exhibited the strongest avidity on 131 I-MIBG scan. Whole Exome Sequencing identified a rare pathogenic somatic variant in the DNMT3A gene (c.2645G>A) with no other germline or somatic mutations in known susceptibility genes. Despite adequate alpha-blockade and supportive care, the patient developed acute liver failure and coagulopathy, rendering her unfit for any form of invasive intervention and finally succumbing to the disease within 3 months of presentation.

CONCLUSION
This case underscores the aggressive nature of Cluster 3 PPGLs associated with DNMT3A mutations, which likely promote tumorigenesis via Wnt-pathway activation and epigenetic dysregulation. The discordant functional imaging reflects significant tumor heterogeneity, which may complicate diagnostic and therapeutic strategies.

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,