

Adult E-Poster

hypoglycemia, particularly when occurring nocturnally. IGF-2 overproduction, typically by large mesenchymal or epithelial tumours, is the underlying reason behind these events. Although surgical resection of the tumour remains the definitive treatment, glucocorticoids and frequent glucose supplementation are used in conservative management.

EP_A083

MEN TYPE 2B SYNDROME IN A NORMOTENSIVE YOUNG FEMALE WITH INCIDENTALLY DISCOVERED PHAEOCHROMOCYTOMA

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INTRODUCTION/BACKGROUND

Phaeochromocytomas (PC) account for up to 5-25% of adrenal incidentalomas. Some PC patients, especially those with an adrenal incidentaloma, are asymptomatic and have normal blood pressure. The frequency of incidentally discovered normotensive phaeochromocytomas is increasing owing to better accessibility of imaging procedures. These tumours may be linked to certain genetic syndromes, such as Multiple Endocrine Neoplasia (MEN) type 2B, a rare condition caused by the RET proto-oncogene mutation, and includes a range of clinical manifestations such as phaeochromocytomas, medullary thyroid carcinoma and mucosal neuromas.

CASE

A 34-year-old female who had undergone total thyroidectomy for medullary thyroid carcinoma (MTC) was referred for an incidental right adrenal mass from CT staging. She had no paroxysms of headache or palpitations, no family history of MTC, pheochromocytoma and MEN-related diseases. She was normotensive. Physical examinations revealed mucosal neuromas on the tongue, buccal mucosa, lips, and eyelids. No marfanoid habitus present. Laboratory results showed normal serum calcium (2.57 mmol/L), but a 24-hour urine metanephrine was four times the upper limit of normal, along with borderline elevation of normetanephrines. The adrenal CT revealed an indeterminate right adrenal mass measuring 2 x 2 x 3 cm with peripheral calcifications suggesting pheochromocytoma.

Left lymph node and carotid sheath biopsy were reported as features consistent with ganglioneuroma. The patient is scheduled for a right retroperitoneoscopic adrenalectomy. The unifying clinical presentations are consistent with MEN 2B Syndrome. However, genetic panel testing was not done due to financial constraints.

CONCLUSION

This case underscores the importance of considering genetic syndromes, such as MEN type 2B, in patients with incidental findings of pheochromocytomas, even when the patient is normotensive. Early diagnosis and genetic testing can help guide management, including surveillance for other tumours associated with MEN type 2B and early intervention. Further research is needed to explore the clinical presentation of pheochromocytomas in normotensive patients.

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PSEUDOACROMEGALY IN A PATIENT WITH PACHYDERMOPERIOSTOSIS

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INTRODUCTION/BACKGROUND

Patients who are clinically suspected of acromegaly are usually referred to an endocrinologist. Biochemical evaluation is necessary to confirm the presence of an abnormality in the growth hormone (GH) axis. There are some conditions with physical features mimicking acromegaly in the absence of GH excess. Given the heterogeneity of conditions that can cause pseudoacromegaly, this posed a diagnostic challenge.

CASE

A 32-year-old male was referred to the Endocrine clinic for evaluation of suspected acromegaly. He had an incidental finding of pancytopenia and chronic excessive sweating when he presented to his General Practitioner. Following this, he was diagnosed with myelofibrosis by a haematologist. He reported having large hands and feet since his teens. He was also referred to a dermatologist for generalised skin thickening and itchiness and treated for photodermatitis.

Clinically, he has clubbing of the fingers of his hands and feet and furrowing of skin on his forehead but no other phenotypical features of acromegaly. Biochemically, IGF-1 and GH levels were normal.

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He had a left leg X-ray followed by a CT of the left tibia and fibula for a swelling on his left lower leg, which showed wavy periosteal thickening in the tibia and fibula suggestive of hypertrophic osteoarthropathy. With a suspicion for primary hypertrophic osteoarthropathy (PHO), it was confirmed through genetic analysis that he has homozygous pathogenic variants identified in SCLO2A1 associated with an autosomal recessive PHO.

CONCLUSION

Primary hypertrophic osteoarthropathy, or pachydermo-periostosis (PDP), is a rare genetic disorder characterised by digital clubbing, periostosis and pachydermia. Myelofibrosis is a complication of PDP where bone marrow becomes scarred and fibrotic. In patients with features of hypertrophic osteoarthropathy and acromegaly, PDP should be considered as part of the differential diagnoses.

EP_A085

ECTOPIC CUSHING'S SYNDROME: THE LONG HUNT FOR THE ELUSIVE CULPRIT

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INTRODUCTION/BACKGROUND

Ectopic Cushing's Syndrome (ECS), caused by non-pituitary ACTH-secreting tumours, is a rare but life-threatening form of hypercortisolism. Diagnosis and management can be challenging due to the small, indolent tumours of variable locations. We present two cases highlighting the complexity of diagnosing and managing ECS.

CASE

A 70-year-old male presented with hypertensive urgency, hypokalemia (K^+ 1.9 mmol/L), and new-onset diabetes mellitus (HbA1c 7.1%). He required four antihypertensives, dual oral antidiabetic therapy and potassium supplementation. Investigations revealed markedly elevated cortisol (3026 nmol/L), non-suppressible with dexamethasone (1750 nmol/L), and high ACTH (500 pg/mL) consistent with ACTH-dependent Cushing's Syndrome (CS). Initial Thorax-Abdomen-Pelvis CT, pituitary MRI and Gallium-68 PET scans were unremarkable. Treatment with ketoconazole and spironolactone led to clinical improvement, allowing discontinuation of antihypertensives, antidiabetics and potassium supplements. Serial CT TAP

later detected an enlarging 1.2 cm right middle lobe lung nodule. Surgical resection confirmed an ACTH-positive carcinoid tumour. The patient remained in remission for 6.5 years post-operatively.

A 59-year-old female with poorly controlled hypertension and diabetes was found to be cushingoid during hospitalisation for a finger abscess. Cortisol was 1164 nmol/L, ACTH 19.5 pmol/L, with non-suppression to dexamethasone. Conventional imaging (CT TAP, pituitary MRI, PET scan) showed no significant abnormality. However, IPSS confirmed an ectopic ACTH source. She exhibited cyclical CS, which was marked by fluctuations in blood pressure, glucose, potassium levels, weight and oedema. Management required a block-and-replace regimen using ketoconazole and hydrocortisone. A Ga-68-DOTATATE PET scan two years later revealed a DOTATATE-avid right lung nodule, but the biopsy was inconclusive. The patient declined further procedures.

CONCLUSION

These cases highlight the diagnostic complexity of ECS, which has required multimodal and serial imaging over the years due to elusive lesions. Biochemical control can be challenging due to cyclical CS demanding balance to avoid complications. Persistent localisation efforts remain essential as surgical resection is potentially curative.

EP_A086

LEFT ADRENAL TUBERCULOSIS MIMICKING PHAEOCHROMOCYTOMA POSSIBLY DUE TO RIFAMPICIN INTERFERENCE IN URINE METANEPHRINES

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INTRODUCTION/BACKGROUND

Phaeochromocytoma classically presents with uncontrolled hypertension and paroxysms of headache, diaphoresis and palpitations. The measurement of 24-hour urinary metanephrines is one of the standard first-line tests for detecting phaeochromocytoma. False elevation results may