Adult E-Poster

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 pachydermoperiostosis (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

be brought about by various factors such as urine volume, medication interference and certain foods.

CASE

We report a case of a 67-year-old male with Hepatitis B and smear-positive pulmonary tuberculosis on maintenance treatment with rifampicin and isoniazid, who was referred for left adrenal incidentaloma from CT of the hepatobiliary system. He denied any paroxysmal symptomatology of phaeochromocytoma and was normotensive. 24-hour urinary metanephrines revealed significantly elevated normetanephrine (14 times the upper limit of normal [30.15 µmol/day]), with normal metanephrine and 3-methoxytyramine levels. The adrenal CT demonstrated a left adrenal mass measuring 2.7 x 1.4 x 2.6 cm, 32 Hounsfield units (HU), with absolute and relative washout of 62.8% and 19.6%, respectively, indicating an indeterminate adrenal mass. The patient was diagnosed with left phaeochromocytoma and underwent laparoscopic left adrenalectomy with Phenoxybenzamine cover. However, the histopathological findings revealed multiple granuloma formation, with special stains negative for acid-fast bacilli, suggestive of chronic changes of right adrenal tuberculosis (non-active) and no features of phaeochromocytoma. Thoracic and abdominopelvic CT scans showed no evidence to suggest paraganglioma, which might contribute to elevated normetanephrine levels. A post-operative repeat 24-hour urine metanephrine came back normal. This repeated sample was taken after the patient completed tuberculosis treatment (including rifampicin). Some reports recognised rifampicin interference with urinary metanephrine measurement as it is eluted with normetanephrine, causing significantly elevated levels. These findings correlate with this patient as urine normetanephrine returned to normal once he was off rifampicin.

CONCLUSION

Histopathological findings of the left adrenal mass were suggestive of post-adrenal tuberculosis rather than phaeochromocytoma. Rifampicin was found to be an interferent in urine metanephrines measurement, which led to falsely elevated normetanephrine levels with no catecholaminergic signs or symptoms.

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A RARE CASE OF TURNER MIMICKER

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INTRODUCTION/BACKGROUND

Primary amenorrhea and delayed puberty are frequently encountered in primary care, prompting suspicion of Turner syndrome, especially in cases with short stature. This case underscores the importance of considering Swyer syndrome even when significant growth impairment is present.

CASE

A 28-year-old phenotypic female, born of a non-consanguineous union, presented with primary amenorrhea and a short stature of 1.31 meters. Physical examination revealed absent secondary sexual characteristics (Tanner stage 1). External genitalia were unambiguously female. Bone age assessment identified significant delay, corresponding to a 15-year-old. The hormonal evaluation showed hypergonadotropic hypogonadism. Thyroid function and insulin growth factor-1 levels were normal. Pelvic MRI demonstrated an atrophic uterus, absent fallopian tubes and ovaries. Karyotype analysis confirmed a 46, XY genotype, consistent with Swyer syndrome. Following pubertal induction for 3 months, she developed regular menstruation and progression to Tanner stage 3.

Swyer syndrome is a rare disorder of sex development featuring female phenotype, hypergonadotropic hypogonadism and streak gonads. While 15-20% of cases result from SRY gene mutations impairing testis-determining factor function, other genes have also been implicated. Swyer syndrome classically causes tall stature from estrogen-deficient delayed epiphyseal fusion. However, our case exhibited profound short stature and severely delayed bone age, explained by the complete prepubertal estrogen deprivation abolishing both growth spurt and fusion. Additional factors, like SHOX gene variations, may have contributed to her growth impairment. Diagnostic complexity arose from initial Turner syndrome overlap; however, the absence of other Turner stigmata and 46, XY karyotype confirmed Swyer syndrome. This emphasises karyotyping's diagnostic importance in primary amenorrhea with hypergonadotropic hypogonadism, regardless of phenotype. Hormone replacement therapy remains crucial for puberty induction, bone health and cardiovascular protection.