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PITUITARY STALK INTERRUPTION SYNDROME DIAGNOSED IN THE FOURTH DECADE: A RARE CAUSE OF PATHOLOGICAL FRACTURE IN ADULTHOOD

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INTRODUCTION
Pituitary stalk interruption syndrome (PSIS) is a rare congenital disorder characterized by the neuroradiological triad of an absent or interrupted pituitary stalk, ectopic posterior pituitary, and anterior pituitary hypoplasia. It is typically diagnosed in infancy or childhood due to growth failure or delayed puberty. Diagnosis in adulthood is uncommon and may occur after decades of untreated hypopituitarism.

CASE
A 34-year-old Malay male with underlying physical and intellectual disability presented after a mechanical fall resulting in left slipped capital femoral epiphysis, an unusual pathological fracture in adulthood. Clinical examination revealed marked infantilism with complete absence of secondary sexual characteristics (Tanner stage I). Laboratory evaluation demonstrated combined pituitary hormone deficiency, including severe central hypothyroidism, profound hypogonadotropic hypogonadism, and central adrenal insufficiency. Growth hormone and insulin-like growth factor 1 were undetectable, while prolactin was mildly elevated, consistent with pituitary stalk disruption due to loss of hypothalamic dopaminergic inhibition. Bone age assessment showed severe delay, corresponding to 15 years. Pituitary magnetic resonance imaging demonstrated the classical PSIS triad: anterior pituitary hypoplasia with partial empty sella, a high T1 signal nodule at the median eminence representing ectopic posterior pituitary, and non-visualization of the infundibulum, consistent with an absent pituitary stalk. Birth history revealed premature breech delivery, a recognized perinatal risk factor. The patient was commenced on hormone replacement therapy, including levothyroxine, hydrocortisone, testosterone undecanoate, and calcium–vitamin D supplementation.

CONCLUSION
This case illustrates that PSIS may remain undiagnosed into adulthood, leading to severe consequences of long-standing hypopituitarism such as osteoporosis and pathological fractures. Clinicians should consider hypopituitarism in adults presenting with unexplained fractures and delayed sexual maturation, particularly when supported by a suggestive perinatal history.

EP_A121

A SILENT INTERVAL WITH AGGRESSIVE RETURN: METASTATIC SDHB-MUTATED MEDIASTINAL PARAGANGLIOMA

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INTRODUCTION
Mediastinal paragangliomas (PGLs) are extremely rare extra-adrenal neuroendocrine tumors, accounting for approximately 2% of all PGLs and commonly associated with pathogenic germline variants (PGVs), particularly those involving succinate dehydrogenase (SDH) mutations.

CASE
We report a 35-year-old female presenting with chronic cough and hemoptysis, accompanied by paroxysmal symptoms and new-onset hypertension. She had a history of pheochromocytoma treated 15 years earlier with right adrenalectomy and liver lobectomy due to intraoperative adrenal adherence to the liver. Histopathology confirmed adrenal pheochromocytoma, with no evidence of PGL in the liver. She remained in biochemical remission for 3 years but subsequently defaulted. Biochemical evaluation revealed markedly elevated 24-hour urinary normetanephrine (58,950 nmol/L; ~26-fold increase). Computed tomography of the thorax demonstrated a mediastinal mass compressing the right bronchus, resulting in luminal narrowing and segmental lung collapse. Endobronchial biopsy confirmed PGL (Ki-67 index 5%). Functional imaging with DOTATATE, FDG-PET, and MIBG demonstrated metastatic disease involving the lungs and lymph nodes. Genetic testing identified a heterozygous pathogenic SDHB mutation (c.79C>T; p.Arg27), consistent with autosomal dominant hereditary PGL-pheochromocytoma syndrome; family screening confirmed the same mutation in her father and two siblings. She underwent two sessions of bronchoscopic intervention, including cryoablation, balloon dilation, argon plasma coagulation, and intratumoral alcohol injection for airway control and hemoptysis. Multidisciplinary evaluation deemed complete surgical resection high risk and not feasible; therefore, peptide receptor radionuclide therapy was initiated.

CONCLUSION

This case demonstrates the aggressive and metastatic nature of SDHB-mutated PGLs. This group of patients require long term surveillance given the risk of developing new tumors even years or decades after primary tumor resection. It highlights the importance of genetic testing following pheochromocytoma surgery to guide targeted therapy, lifelong surveillance, and family screening within a multidisciplinary care approach.

EP_A122

PRUDENT MANAGEMENT OF MICROPROLACTINOMA IN A TRANSGENDER WOMAN ON FEMINIZING HORMONAL THERAPY

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INTRODUCTION

Gender-affirming hormone therapy (GAHT) for transgender women utilizes estrogen and an anti-androgen like cyproterone acetate (CPA) which can both lead to hyperprolactinemia. Although mild prolactin elevations are common, the development of prolactinomas in transgender women on GAHT is rare.

CASE

A 23-year-old trans-woman on self-purchased estradiol hemihydrate (17β-estradiol) 4 mg and CPA 12.5 mg daily for GAHT presented with galactorrhea. Investigations revealed markedly elevated prolactin at 2,369 mIU/L and elevated estradiol at 848 pmol/L. A pituitary magnetic resonance imaging (MRI) identified a 0.4 × 0.5 cm microprolactinoma.

She declined clinical advice to reduce her medication dose and continued on the same treatment. In the subsequent year, her peak prolactin was 1,383 mIU/L, and a repeat MRI showed the microprolactinoma remained stable. She continues to be on close clinical monitoring.

Transgender females experience approximately a fourfold higher rate of developing prolactinomas. The patient's choice to persist with GAHT reflects the challenges in managing gender dysphoria alongside medical complications. A recent paper by BJ Nolan et al. suggests using prolactin levels exceeding 2,000–3,000 mIU/L to guide further investigations including a pituitary MRI to rule out a prolactinoma. In most cases, the serum prolactin levels will return to the normal range with a reduction or discontinuation of the GAHT. Normalization of prolactin levels have been reported after gonadectomy and CPA cessation in transgender females on GAHT, which suggests that CPA usage may be associated with higher risk of hyperprolactinemia compared to estrogen therapy.

CONCLUSION

This case highlights that GAHT can lead to development of prolactinomas. Prolactin monitoring and MRI investigations should be reserved for symptomatic patients, and for those with significantly elevated or increasing prolactin levels. While treatment using dopamine agonists have been reported, this case demonstrates that conservative management and close monitoring can be a viable approach for structurally stable microprolactinomas.

EP_A123

DIAGNOSTIC AND THERAPEUTIC ROLE OF ENDOSCOPIC ULTRASOUND (EUS) IN A CT-NEGATIVE OCCULT INSULINOMA

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INTRODUCTION

A negative computed tomography (CT) scan does not preclude an insulinoma, as small lesions frequently remain undetected on conventional imaging. This case highlights the indispensable role of endoscopic ultrasound (EUS)—not just for localizing occult tumors, but as a definitive, minimally invasive therapeutic alternative to high-risk surgical resection.

CASE

A 39-year-old female with underlying hypertension presented with a 5-month history of predominantly fasting hypoglycemia (glucose <3.0 mmol/L) and neuroglycopenic symptoms, fulfilling Whipple's triad. A supervised 72-hour fast confirmed endogenous hyperinsulinemic hypoglycemia at 31 hours, with a nadir glucose of 1.4 mmol/L, insulin 116 pmol/L, and C-peptide 821 pmol/L. Notably, contrast-enhanced CT of the pancreas was reported as normal. To overcome this, EUS was performed, successfully identifying a hidden 19 × 18 mm lesion in the head of the pancreas, intimately abutting the main pancreatic duct.

Despite medical therapy with diazoxide and strict dietary modifications, her hypoglycemia remained refractory. Given the tumor's proximity to the main pancreatic duct, surgical enucleation carried a prohibitively high risk of