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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.

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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.