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DIAGNOSTIC DILEMMA IN INTERPRETING INFERIOR PETROSAL SINUS SAMPLING IN SUSPECTED CYCLICAL CUSHING'S DISEASE IN AN ADOLESCENT

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INTRODUCTION

Cushing's disease in adolescents is rare and frequently difficult to diagnose due to overlapping features with common pubertal and metabolic conditions. We present an adolescent who underwent inferior petrosal sinus sampling (IPSS) with possible cyclical Cushing's disease, highlighting the diagnostic challenges and the interpretive value of IPSS.

CASE

A 16-year-old female presented with progressive weight gain, acne, hirsutism, secondary amenorrhea, proximal myopathy, violaceous striae, and facial plethora over 1 year. Her 24-hour urinary cortisol was >3,310 nmol/L with failure of suppression following a 1-mg overnight dexamethasone suppression test (701 nmol/L). Her adrenocorticotrophic hormone (ACTH) was raised (77.9 pg/mL), consistent with ACTH-dependent Cushing syndrome. Her hemoglobin A1c was 5.9%, and she has low bone mass for age with a Z score of -2.9 at the Lumbar spine.

Pituitary magnetic resonance imaging identified a right-sided microadenoma measuring 0.6 × 0.5 cm, while computed tomography of the thorax, abdomen, and pelvis showed no ectopic source. IPSS with desmopressin stimulation was performed to confirm the ACTH source. No steroidogenesis inhibitors were administered prior to testing. Notably, cortisol levels during IPSS were relatively low, with midnight cortisol of 91 nmol/L and morning cortisol of 200 nmol/L, suggesting testing during a trough phase of possible cyclical hypercortisolism. Despite this, IPSS demonstrated a significant central-to-peripheral ACTH gradient supporting a pituitary source.

CONCLUSION

This case highlights the diagnostic dilemma of interpreting IPSS in suspected cyclical Cushing's disease. Fluctuating cortisol secretion may result in discordant biochemical findings and complicate localization studies. IPSS findings

should be interpreted cautiously and integrated with clinical features, imaging, and serial biochemical monitoring. Recognition of cyclical disease patterns is essential to ensure appropriate management in adolescent patients.

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WHEN IGF-1 MISLEADS: DISCORDANT BIOCHEMICAL FINDINGS IN ACROMEGALY

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INTRODUCTION

Acromegaly is an endocrine disorder caused by excess growth hormone (GH), causing somatic overgrowth, multiple comorbidities, and premature mortality. It is confirmed biochemically by an elevated GH level, which is not suppressed post oral glucose tolerance test (OGTT). The serum level of insulin-like growth factor-1 (IGF-1) is recommended for diagnosis, monitoring, and screening, and a normal level effectively excludes the disease. We report a patient with acromegaly who presented with normal IGF-1.

CASE

A 21-year-old female, with no known medical illness, presented with a persistent, progressive headache and amenorrhea for the past 6 months. She also noticed a change in facial appearance and an increase in the size of her hands and feet. Blood parameters revealed raised GH level of >50 ng/mL, normal IGF-1 30.3 nmol/L (12.17–44.80), mildly raised prolactin 692 mIU/L, low am cortisol 62 nmol/L (166–507), thyroid-stimulating hormone of 0.63 mIU/L (0.27–4.20), free thyroxine 4 11 pmol/L (12–22), low follicle-stimulating hormone 0.90 IU/L, luteinizing hormone <0.30 IU/L, estradiol <18.4 pmol/L, and fasting blood sugar of 17.7 mmol/L with hemoglobin A1c 9%. In view of normal IGF-1 with a high index of suspicion for acromegaly, she underwent OGTT which showed unsuppressed GH. Magnetic resonance imaging pituitary showed sellar mass 2.4 × 2.9 × 2.1 cm with suprasellar extension as well as extension into the right cavernous sinus, suggestive of pituitary macroadenoma. She was diagnosed with acromegaly with secondary adrenal insufficiency and central hypothyroidism with hypogonadotropic hypogonadism, complicated with poorly controlled diabetes. She was started on thyroxine and hydrocortisone replacement and required basal bolus insulin of 1.3 u/kg/day. Repeated IGF-1 showed a raised