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BIOCHEMICAL DISCORDANCE IN ACROMEGALY COMPLICATED BY PITUITARY APOPLEXY AND SEVERE INSULIN RESISTANCE

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INTRODUCTION

Acromegaly is usually diagnosed by elevated age- and sex-adjusted insulin-like growth factor-1 (IGF-1) levels reflecting chronic growth hormone (GH) excess. IGF-1 is preferred as a screening biomarker due to its longer half-life and reduced pulsatility compared with GH. However, IGF-1 levels may be disproportionately low or only modestly elevated in certain clinical contexts, leading to diagnostic uncertainty. Pituitary apoplexy is one such condition in which acute tumor hemorrhage or infarction may disrupt sustained GH secretion and attenuate IGF-1 production.

CASE

A 48-year-old female with hypertension, type 2 diabetes mellitus, and dyslipidemia presented with a 2-day history of severe headache, vomiting, and visual disturbance, on a background of progressive acral enlargement over 2 years. Examination revealed coarse facial features, prognathism, enlarged hands, and cranial nerve involvement. Magnetic resonance imaging demonstrated an invasive sellar–suprasellar pituitary macroadenoma with optic chiasmal compression and cavernous sinus encasement. Intravenous dexamethasone was initiated pre-operatively due to a significant mass effect.

Biochemical evaluation showed markedly elevated random GH levels (>50 ng/mL) with only mildly elevated IGF-1 at 1.19 times the upper limit of normal, below the threshold at which confirmatory oral glucose tolerance testing may be omitted according to current guidelines. Other pituitary axes suggested evolving hypopituitarism. During admission, she developed severe hyperglycemia with marked insulin resistance, requiring high-dose insulin therapy (approximately 1.5 U/kg/day). She underwent urgent transsphenoidal surgery, with histopathology confirming a pituitary neuroendocrine tumor with extensive hemorrhage and infarction, consistent with pituitary apoplexy. Postoperatively, GH levels were suppressed to <5 ng/mL, insulin requirements decreased markedly, and hormone replacement was initiated for secondary adrenal insufficiency and central hypothyroidism.

CONCLUSION

This case highlights that IGF-1 levels below conventional diagnostic thresholds do not exclude clinically significant acromegaly, particularly in the setting of pituitary apoplexy. Integration of clinical phenotype, GH levels, and imaging findings is essential to avoid diagnostic delay and ensure timely management.

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GROWTH AGAINST THE CLOCK: HORMONAL THERAPY IN LATE-DIAGNOSED MOSAIC TURNER SYNDROME

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INTRODUCTION

Short stature and delayed puberty characterize Turner syndrome (TS). The 2024 international clinical practice guidelines recommend growth hormone (GH) for late-diagnosed patients if epiphyses remain open. For patients with remaining growth potential, initiating GH alongside low-dose estrogen effectively balances linear growth with the need for timely pubertal induction.

CASE

A 15-year-old female, born prematurely at 6 months gestation, presented with delayed puberty, primary amenorrhea, and short stature. Examination revealed a height of 122 cm (<5th percentile), weight of 26 kg, with no syndromic facies, and Tanner stage 1. Investigations confirmed hypergonadotropic hypogonadism. Metabolic screening, including thyroid, renal, and liver profile, was normal. Her baseline insulin-like growth factor 1 (IGF-1) was low at 117.5 ng/mL (127.5–541.5). Karyotyping confirmed mosaic TS (45,X/46,Xr). Her skeletal bone age was delayed at 12 years, indicating a viable window for linear growth prior to complete epiphyseal fusion.

Subcutaneous GH was initiated at 0.3 mg up titrated to 1.2 mg (0.45 µg/kg) daily over 4 weeks, then 1.35 mg (50 µg/kg) daily at month 5. Low-dose oral estradiol (0.5 mg three times weekly) was introduced for pubertal induction at month 4. After 9 months of combined GH and estrogen therapy, the patient achieved a height increment of 6 cm, reaching 128 cm without an adverse event. To achieve the clinical target of a 10–15 cm increment in the first year, her GH dose was further increased to 1.50 mg (55 µg/kg) daily. She showed an appropriate biochemical response with IGF-1 increased to 40.2 nmol/L (16.4–67.8) with a total height gain of 6 cm over the first 9 months of GH therapy.

CONCLUSION
Concomitant GH and estrogen therapy in late-diagnosed TS successfully induced clinically significant height gain. This dual approach maximized the limited window for linear growth, without delaying pubertal induction and compromising patient's psychosocial well-being.

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FULVESTRANT INTERFERENCE WITH ESTRADIOL IMMUNOASSAYS: A CASE REPORT AND REVIEW OF LABORATORY IMPLICATIONS

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INTRODUCTION
Fulvestrant, a selective estrogen receptor degrader widely used in hormone receptor-positive breast cancer, can cause significant analytical interference with estradiol immunoassays due to structural similarity with 17β-estradiol. This interference may result in falsely elevated estradiol measurements, potentially leading to diagnostic confusion and inappropriate clinical interventions. We report a case demonstrating this clinically significant analytical interference and emphasize the importance of liquid chromatography-tandem mass spectrometry (LC-MS/MS) in hormone monitoring for patients receiving fulvestrant therapy.

CASE
A 31-year-old female with metastatic hormone receptor-positive breast cancer receiving combination therapy (fulvestrant, letrozole, ribociclib, and goserelin) presented with inappropriately elevated serum estradiol levels of 566 pmol/L (early follicular phase 200–500 pmol/L), measured by chemiluminescence immunoassay. Despite effective gonadotrophin suppression evidenced by low luteinizing hormone (0.2 IU /L) and follicle-stimulating hormone (4.1 IU/L) levels and clinical amenorrhea, the discordant estradiol elevation raised suspicion of assay interference. Confirmatory testing with LC-MS/MS revealed an estradiol concentration of <36 pmol/L, consistent with expected hormonal suppression and confirming immunoassay cross-reactivity with fulvestrant.

The case demonstrates significant fulvestrant interference with estradiol immunoassays, resulting in a greater than 15-fold overestimation compared to LC-MS/MS measurement. The clinical presentation was consistent with effective endocrine therapy, supported by suppressed gonadotropin levels, clinical amenorrhea, and stable disease on imaging. LC-MS/MS provided accurate estradiol measurement, avoiding potential diagnostic confusion and unnecessary clinical interventions.

CONCLUSION
Clinicians should be aware of potential fulvestrant interference with estradiol immunoassays when interpreting hormone levels in breast cancer patients. Discordant laboratory findings, particularly elevated estradiol despite clinical evidence of effective hormonal suppression, should prompt consideration of LC-MS/MS confirmation. This case underscores the clinical importance of analytical method selection in hormone monitoring and highlights the need for improved laboratory-clinical communication to optimize patient care in oncology practice.

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PRIMARY AMENORRHEA IN A PATIENT WITH CONGENITAL ANOMALIES OF THE KIDNEY AND URINARY TRACT (CAKUT): UNMASKING ATYPICAL MRKH SYNDROME TYPE II

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INTRODUCTION
Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is characterized by congenital agenesis or hypoplasia of the uterus and upper vagina in phenotypic females with normal secondary sexual characteristics and a 46,XX karyotype. Atypical MRKH syndrome, or MRKH type II, is associated with extragenital anomalies, particularly renal abnormalities. Its diagnosis may be delayed when overshadowed by complex medical comorbidities.

CASE
A 19-year-old phenotypic female with CAKUT, complicated by right duplex kidney and end-stage renal failure on continuous ambulatory peritoneal dialysis, was admitted in September 2025 for peritonitis. Further history revealed primary amenorrhea. Physical examination demonstrated preserved pubertal development with Tanner stage III pubic hair and at least Tanner stage IV breast development. Hormonal profile was unremarkable, and chromosomal analysis showed a normal female karyotype (46,XX). Pelvic ultrasonography and computed tomography of the thorax, abdomen, and pelvis demonstrated uterine agenesis. In addition, bilateral oval-shaped heterogeneous soft tissue masses were identified within the paracolic gutters; ultrasonography showed multiple cysts within both masses,