

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 17 β -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,

and each was associated with a gonadal vein, suggestive of bilateral ectopic gonadal structures on imaging. These findings were consistent with atypical MRKH syndrome type II in the context of underlying renal anomalies.

CONCLUSION

This case highlights the importance of evaluating primary amenorrhea even in patients with significant chronic illness. In phenotypic females with CAKUT, associated Müllerian anomalies should be actively considered. Atypical MRKH syndrome type II should be recognized early to enable accurate diagnosis, multidisciplinary follow-up, and appropriate reproductive and psychosocial counseling.

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ACTIVE MODERATE-TO-SEVERE THYROID EYE DISEASE: A CASE SERIES

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INTRODUCTION

Thyroid eye disease (TED) is a heterogeneous condition and the most common extrathyroidal manifestation of Graves' disease. Although most cases are mild, disease progression may threaten vision and significantly impact quality of life.

METHODOLOGY

We described four patients with active moderate-to-severe TED, highlighting their clinical characteristics, therapeutic approaches, and clinical outcomes.

RESULTS

Three males and one female patient were included in this descriptive study. Most were identified by the endocrine team. Their median age was 52.5 years (range 47–56 years). All patients had long-standing Graves' disease (range 6–10 years). Two of them were active smokers. Their clinical activity score ranged from 3 to 5. Common presenting features included diplopia (60%), proptosis (60%), and ocular redness (60%).

At the onset of active TED, all patients were euthyroid while receiving either antithyroid therapy or were maintained on stable levothyroxine following radioactive iodine treatment and thyroidectomy. Thyroid receptor autoantibodies were assessed in only two patients, with one elevated result. Orbital computed tomography, performed in 3 patients, demonstrated extraocular muscle enlargement consistent with TED, particularly involving the inferior and medial recti.

All patients received standard-dose intravenous methylprednisolone (cumulative 3.5–4.5 g). They showed improvement in visual acuity following treatment, with partial improvement in ophthalmoplegia.

CONCLUSION

Clinical risk factors observed in this series were consistent with the literature. TED diagnosis relies heavily on recognition by the endocrine team, underscoring the importance of holistic Graves' disease management. High-dose corticosteroids remain the mainstay of treatment in our setting, with generally favorable outcomes when timely multidisciplinary care is provided.

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BEYOND THE PITUITARY STALK: PRIMARY HYPOTHYROIDISM AS A RARE PRESENTATION OF MULTISYSTEM LANGERHANS CELL HISTIOCYTOSIS

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INTRODUCTION

Langerhans cell histiocytosis (LCH) is a clonal proliferative disorder of langerin-positive histiocytes. Endocrine involvement most frequently manifests as central diabetes insipidus or secondary hypothyroidism, caused by infiltration of the hypothalamic-pituitary axis. Conversely, direct infiltration of the thyroid gland leading to primary hypothyroidism is an exceptionally rare clinical entity, particularly when presenting concurrently with central disease.

CASE

A 21-year-old female with known right otic and multisystem LCH presented with septic shock secondary to a right ear abscess accompanied by polyuria and polydipsia. Physical examination revealed a palpable goiter. Clinical and biochemical evaluation confirmed central diabetes insipidus with associated anterior hypopituitarism (low adrenocorticotrophic hormone, follicle-stimulating hormone, and luteinizing hormone). However, concurrent thyroid function tests demonstrated overt primary hypothyroidism, evidenced by an appropriately elevated thyroid-stimulating hormone (26.19 mIU/L) and low free T4 (5.47 pmol/L), rather than the anticipated secondary hypothyroidism. Neck ultrasound showed diffuse thyroid enlargement with heterogeneous echotexture. Crucially, anti-thyroid peroxidase and anti-thyroglobulin antibodies were both negative, rendering Hashimoto's thyroiditis highly unlikely. Although the patient declined confirmatory