

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,