

Adult E-Poster

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PRIMARY AMENORRHEA AND ANOSMIA IN A YOUNG WOMAN: A LATE DIAGNOSIS OF KALLMANN SYNDROME

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INTRODUCTION/BACKGROUND

Kallmann syndrome (KS), a genetic form of congenital hypogonadotropic hypogonadism (CHH), is characterized by delayed or absent puberty in combination with anosmia or hyposmia. It is more frequently diagnosed in males and often overlooked in females due to subtler clinical features. Underrecognition of anosmia and the absence of routine olfactory assessment contribute to diagnostic delays. Early diagnosis and hormone replacement therapy are essential for initiating pubertal development and supporting reproductive planning.

CASE

A 25-year-old female was referred for evaluation of primary amenorrhea and absent secondary sexual characteristics. She reported a lifelong inability to perceive odors. She was born full-term with no perinatal complications or congenital anomalies. Physical examination showed Tanner stage I breast and pubic hair development. Hormonal evaluation revealed low estradiol, luteinizing hormone (LH), and follicle-stimulating hormone (FSH), consistent with hypogonadotropic hypogonadism. Prolactin and thyroid hormones were within normal limits. Pelvic ultrasound revealed hypoplastic uterus and ovaries. Olfactory testing confirmed anosmia. Brain MRI demonstrated bilateral atrophy of the olfactory bulbs and non-visualization of the olfactory sulci, with normal pituitary anatomy. Bone mineral density (BMD) screening revealed osteoporosis. A diagnosis of Kallmann syndrome was established. Estrogen therapy was initiated, resulting in early breast development.

CONCLUSION

This case highlights the need to consider Kallmann syndrome in women presenting with primary amenorrhea, particularly when anosmia is present. Olfactory testing and neuroimaging are essential components of the diagnostic workup. BMD screening plays a crucial role in evaluating long-term complications of untreated hypogonadism. Timely hormonal therapy supports secondary sexual development, improves bone health, and may enhance psychosocial and reproductive outcomes.

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VIRILISATION SECONDARY TO LEYDIG CELL OVARIAN TUMOR IN A POSTMENOPAUSAL WOMAN WITH PRIMARY HYPERPARATHYROIDISM

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INTRODUCTION/BACKGROUND

Ovarian sex cord-stromal tumors, which include steroid cell tumors, comprise 5–8% of all ovarian tumors. Less than half are androgen-secreting. Leydig cell tumors are a type of steroid cell tumor. They are usually androgen-secreting, unilateral, and can be either benign or malignant. We report a case diagnosed in a postmenopausal woman during follow-up for primary hyperparathyroidism.

CASE

A 66-year-old female was noted to have significant hirsutism during a follow-up consult for hyperthyroidism, which reportedly started in the past 5 years. She had primary hyperparathyroidism secondary to right inferior parathyroid adenoma for the last 10 years, not fulfilling criteria for surgery. Her other comorbidities were rheumatoid arthritis diagnosed at age 50; as well as diabetes mellitus, hypertension, dyslipidaemia and fatty liver disease, diagnosed between 54 to 62 years of age. She had 3 children and experienced early menopause at age 42. She had frontal balding and terminal hair growth on chest, back, abdomen, face and limbs. Systemic examination was unremarkable.

Testosterone level was 14.8 nmol/L (normal range 0.24–1.70) while dehydroepiandrosterone sulfate (DHEAS) level was normal. CT imaging noted an enhancing focus in the left ovary (0.8 × 1.1 × 1.4cm). She underwent exploratory abdominal hysterectomy with bilateral salpingo-oophorectomy, appendectomy and omental biopsy which revealed Leydig cell tumor of the left ovary, with intact capsule. Two months post-operatively, testosterone was undetectable (<0.087 nmol/L) and the patient reported reduced facial hair growth.

CONCLUSION

The development of true hirsutism, alopecia, and/or acne in postmenopausal women should not be disregarded, and assessment for causes of postmenopausal hyperandrogenism should be undertaken. This case illustrates a rare co-existence of an androgen-secreting ovarian tumour with concomitant long-standing primary hyperparathyroidism.