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Challenges in Managing a Rare Case of Female Kallman Syndrome

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INTRODUCTION

Kallmann syndrome (KS), characterised by anosmic hypogonadotrophic hypogonadism, is a very rare genetic disorder in females. Delayed diagnosis presents additional management challenges. We report the case of a 58-year-old female diagnosed at 37 years old.

CASE

She had asymptomatic primary amenorrhoea, anosmia, absence of secondary sexual characteristics and low body mass. There were no eunuchoidal features but her weight was subnormal, with hypogonadotrophic hypogonadism. Breakthrough menstruation with progestin challenge test suggested anovulation. Her gender was confirmed with cytogenetic analysis. Hormone replacement therapy (HRT) was initiated with a priming dose of conjugated estrogen. Subsequently, estrogen was increased to 0.625 mg BD (day 1 to 21) and progesterone 5 mg (day 14 to 12) was added. In the first 6 months after HRT initiation, there were notable physical changes including increase in axillary hair growth and breast development of stage 2 to 3. Comorbidities of multinodular goitre, osteoporosis, dyslipidemia and paroxysmal supraventricular tachycardia evolved during the follow up, and were intensively investigated and managed accordingly. Despite HRT and subsequent anti-osteoporotic treatment with alendronate and optimal nutrition for 2 years, there was no improvement in bone mineral density.

CONCLUSION

To the best of our knowledge, we reported the first female Kallmann syndrome in the postmenopausal age group with multiple comorbidities. While dyslipidemia may reflect the weaning of HRT, the evidence base on how to optimise the benefits of HRT in a female patient with Kallmann syndrome including type, combination, dosage and duration of treatment, are lacking. Following delayed presentation beyond the period of accrual of peak bone mass, instituting HRT may not optimise bone mineral composition. Furthermore, the use of bisphosphonate in this patient is based on extrapolation of findings in subjects with postmenopausal osteoporosis. Prompt diagnosis and treatment in early childhood and prepuberty as well as continual active surveillance are important in managing the female patient with KS.

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Discordant Thyroid Function Tests Due to Dysalbuminemic Hyperthyroxinemia Confounds Management of Thyroid Autoimmunity

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INTRODUCTION

Familial dysalbuminemic hyperthyroxinemia (FDH) is a cause of discordant thyroid function tests (TFT) due to interference in free thyroxine (FT4) assays caused by the mutant albumin. The coexistence of thyroid disease and FDH can further complicate diagnosis and potentially result in inappropriate management.

We describe a case of Hashimoto's thyroiditis and Graves' disease occurring on a background of FDH.

CASE

A 42-year-old lady with longstanding autoimmune hypothyroidism was treated with varying dosages of thyroxine because of discordant TFTs, showing high (FT4) and normal thyroid stimulating hormone (TSH). Discontinuation of thyroxine led to marked TSH rise but with normal FT4 levels. She then developed Graves' disease and thyroid ophthalmopathy, with markedly elevated FT4 (62.7 pmol/L), suppressed TSH (0.03 mIU/L) and positive anti-TSH receptor antibody levels. However, propylthiouracil treatment even in low dosage (100 mg daily) resulted in profound hypothyroidism (TSH 138 mIU/L, FT4 4.8 pmol/L), prompting its discontinuation and recommencement of thyroxine. Discordant thyroid hormone measurements using two different methods suggested analytical interference. Elevated circulating total T4 (TT4) [227 nmol/L, normal range (NR) 69 to 141] but normal thyroxine binding globulin (TBG) levels (19.2 μg/mL, NR 14.0 to 31.0) together with increased binding of patient's serum to radiolabeled T4 suggested FDH. ALB sequencing confirmed a causal albumin variant (R218H).

CONCLUSION

This case highlights the difficulty in ascertaining true thyroid status in patients with autoimmune thyroid disease and coexisting FDH. Early recognition of FDH as a cause for discordant TFTs, with use of either TSH or FT4 measured by equilibrium dialysis as markers of true thyroid status, may improve patient management.