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A RARE CASE OF HYPOGONADOTROPHIC HYPOGONADISM IN AN ADOLESCENT FEMALE

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INTRODUCTION

Hypogonadotrophic hypogonadism refers to hypogonadism due to deficiency in gonadotrophins. Reduced levels of gonadotrophins [luteinizing hormone (LH) and follicle-stimulating hormone (FSH)] results in lack of stimulation for estradiol production leading to amenorrhea, absent breast and uterine development in females. The underlying gonadotrophin deficiency is commonly due to lesions in the pituitary or hypothalamus but can be due to rare genetic causes.

RESULTS

We present a case of a 16-year-old girl who presented with primary amenorrhea and lack of secondary sexual characteristics. She had no underlying medical conditions and denied any other symptoms. There were no excessive stress or weight changes noted. Both her sister and mother attained menarche at 14 years old. On examination, her BMI was 25 kg/m² and height was 151 cm (mid-parental height was 155 cm). There were no syndromic features, no hirsutism, and no features suggestive of virilization. External genitalia examination revealed an infantile labia with intact introitus. Tanner staging for breast was 2/5 and for pubic hair 1/5. Her hormonal profile showed hypogonadotrophic hypogonadism. Her estradiol levels were undetectable at <36.7 pmol/L. LH was 1.14 IU/L (NR 2.4–12.6) and FSH was 2.61 IU/L (NR 3.5–12.5). Testosterone level was also low at 0.1 nmol/L (NR 0.3–2.4). Other anterior pituitary hormones were normal. Her bone age was delayed at 14 years compared to her chronological age of 16 years. Karyotyping showed female genotype of 46XX. Pelvic MRI showed hypoplastic uterus with normal vagina and ovaries. Pituitary MRI revealed normal pituitary gland. There were no obvious causes for her condition.

CONCLUSION

The diagnosis for this case is Idiopathic Hypogonadotrophic Hypogonadism (IHH). This is a diagnosis of exclusion. There may be rare genetic defects affecting neurons in the hypothalamus and/or pituitary responsible for this presentation and genetic testing can be helpful.

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QUALITY OF LIFE AND ITS ASSOCIATION WITH BONE TURNOVER MARKERS IN PATIENTS WITH THALASSEMIA

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INTRODUCTION

As a result of improved treatment and longer life expectancy, thalassemia is now extending into adulthood. Although morbidity and mortality of patients have been reduced significantly, some aspects of the disease impact patients' lives. This study investigated the quality of life of patients with transfusion dependent thalassemia and its association with bone turnover markers (BTM).

METHODOLOGY

This cross-sectional study recruited patients with transfusion-dependent thalassemia (n=40) from an adult haematology clinic. Patients younger than 18 years, with liver disease, on anti-resorptive therapy or corticosteroids were excluded. Participants underwent anthropometric measurements, pubertal assessment, biochemical profiles-ferritin, calcium, phosphate, 25-hydroxyvitamin D, bone turnover markers (s-CTX and s-P1NP), anterior pituitary hormone levels and glucagon stimulation testing. A self-administered 36-item Short Form (SF-36) health survey questionnaire was used to measure the patients' quality of life (QOL) in the form of scores ranging from 0 (worst health) to 100 (best possible health).

RESULTS

A total of 40 patients were included. 47.5% were female and 52.5% were male, with mean age of 27.5 ± 5.2 years and mean body mass index of 19.4 ± 2.45 kg/m². Hypovitaminosis D (<50 nmol/l), elevated serum ferritin (>500 ug/l) and endocrinopathies were found in 90% of patients while 27.5% had abnormal BTM, with significant negative correlation between vitamin D and bone formation marker, P1NP (r=-0.364, p=0.024). Majority of the patients had a physical and mental component summary score >50 (87.5% and 90% of patients respectively). Among the eight SF-36 domains, vitality showed the highest percentage of patients (40%) with score below 50, followed by general health and role physical (37.5% each). Bodily pain domain had significant correlation with P1NP (r=-0.311, p=0.05), whereas other components of patients' physical or mental health were not affected by the abnormal bone turnover markers or hypovitaminosis D (p>0.05).