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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/m^2 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 ± 5.2years and mean body mass index of 19.4 ± 2.45 kg/m^2. 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).

CONCLUSION
Bodily pain, a component of physical health and hypovitaminosis D had negative impact on bone-turnover. Overall, majority of participants had SF-36 health survey scores that trended towards good physical and mental health signaling satisfactory QOL despite being largely affected by comorbidities associated with transfusion-dependent thalassemia.

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A CASE OF SEVERE PROXIMAL MYOPATHY IN A PATIENT WITH ATYPICAL PARATHYROID ADENOMA

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INTRODUCTION
Parathyroid-induced myopathy is a rare neuromuscular manifestation of primary hyperparathyroidism leading to progressive proximal muscle weakness, pain and atrophy. Severity of the weakness varies in relation to the duration and degree of hyperparathyroidism.

RESULTS
We describe a 30-year-old male who presented with progressive debilitating muscle weakness, severe muscle wasting and recurrent muscle spasms over 1 year. He also experienced bone pain, anorexia and weight loss. He had a symmetrical proximal myopathy and muscle wasting of both upper and lower limbs with MRC grade 3/5 on shoulder abduction, adduction, hip extension and flexion. Corrected calcium 4.08 mmol/L (2.10-2.55), phosphate 1.22 mmol/L (0.72-1.52), iPTH >3000.0 pg/ml (15.0-68.3) were suggestive of primary hyperparathyroidism. Parathyroid ultrasound and SESTAMIBI scan localised a hyperfunctioning left superior parathyroid adenoma. His 25 OH-Vitamin D was 39.7 nmol/L suggestive of Vitamin D insufficiency. An elevated alkaline phosphatase at 1807 U/L (40-150), skeletal survey with cortical thinning and generalise low bone density along with bilateral nephrocalcinosis and nephrolithiasis reflected skeletal and renal involvement, common complications of primary hyperparathyroidism. However, an elevated creatine kinase (CK) of 861 U/L (30-200) despite a normal nerve conduction study and electromyography was indicative of a rare myopathic involvement. He underwent successful parathyroidectomy following treatment with hyperhydration, intravenous pamidronate and denosumab. There was resolution of severe muscular spasms, improvement in muscle strength, weight gain and normalisation of his CK, calcium, PTH and vitamin D levels. The histopathological examination confirmed an atypical parathyroid adenoma.

CONCLUSION
Severe proximal myopathy is a rare complication of primary hyperparathyroidism. Cases of atypical parathyroid adenoma, a rare intermediate neoplasm of uncertain malignant potential may present with a more severe clinical and biochemical profile. Prompt diagnosis and parathyroidectomy can prevent complications and improve clinical outcomes.

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PRIMARY HYPERPARATHYROIDISM DURING PREGNANCY: A CASE REPORT

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INTRODUCTION
Primary hyperparathyroidism (PHPT) is a bone and mineral metabolism disorder caused by autonomous secretion of parathyroid hormone (PTH). PHPT is rare in pregnancy with a quoted incidence of 1%. PHPT during pregnancy is challenging to diagnose and difficult to manage. This is due to limited diagnostic and therapeutic options available during pregnancy and the lack of clinical guidelines. PHPT poses serious maternal and foetal complications such as hyperemesis gravidarum, hypercalcaemic crises in the mother, preterm delivery or miscarriage, and neonatal hypocalcaemia. The definitive treatment for PHPT in pregnancy is parathyroidectomy.

We report a case of PHPT diagnosed and managed during pregnancy.

RESULTS
A 35-year-old female who was 27 weeks pregnant, G3P2, presented with prolonged nausea and vomiting up to her second trimester of pregnancy. Blood results showed serum corrected calcium of 3.17 mmol/L (reference range 2.20-2.65), serum phosphate level of 0.56 mmol/L (reference range 0.81-1.45), alkaline phosphatase of 601U/L (reference range 30-120), intact PTH of 346 pmol/L (reference range 14.9-56.9) and normal renal function. Her calcium clearance to creatinine clearance ratio was 0.016. Ultrasound of the neck showed an enlarged left superior parathyroid gland. She was admitted to the ward for intravenous rehydration with forced diuresis. After 1 week trial of outpatient oral rehydration, repeated serum corrected calcium was 2.77 mmol/L. After multi-disciplinary discussion and family conference, a decision was reached to perform parathyroidectomy. Following left superior parathyroidectomy, her serum calcium returned to normal, and symptoms of nausea as well as vomiting has resolved.