p <0.01) compared to the normal group. Male participants had significantly higher BMI (23.8 ± 8.27 kg/m^2 vs 19.92 ± 12.31 kg/m^2, p <0.05) and weight (47.14 ± 25.71 kg vs 31.47 ± 16.23 kg, p <0.05 compared to female. A total of 60.5% of the participants had serum 25 (OH) D levels ≤50 nmol/L. There were no significant differences in 25 (OH) D levels between both groups. Additionally, daily exposure to sunlight for more than 2 hours was associated with higher serum vitamin D levels among children and adolescents (p <0.05). Furthermore, a high household income was associated with a high risk of weight gain (p <0.01) and waist circumference (p <0.01) among children and adolescents.

CONCLUSION

There was a high incidence of vitamin D deficiencies in children and adolescents with no association between BMI and serum 25 (OH) D levels.

EP_P022

SEVERE STUNTING: A RARE CASE OF ACROMESOMELIC DYSPLASIA WITH CONCOMITANT GHD

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INTRODUCTION/BACKGROUND

Acromesomelic dysplasia, type Maroteaux (AMDM) is a rare skeletal dysplasia, characterized by severe disproportionate short stature, primarily affecting the middle and distal segments of the limbs.

CASE

An 11-year-old female presented with extremely short stature. She was born with a birth weight of 2.6 kg and has been noticed to be short since 2 years old. Her parents were not consanguineous but had a strong family history of short stature. Father's height was 154 cm (SDS -3.16) and mother's height was 148 cm (SDS -2.36). The girl has normal intelligence and an unremarkable medical history. She was prepubertal and markedly short with a height of 108.5 cm (SDS -5.03) and a weight of 23.3 kg (SDS - 2.74). Her height was 3 SD below her mid-parental height (MPH SDS-2.3). She had a disproportionate body proportion with an upper segment to lower body segment ratio of 1.25 and shortened middle and distal segments of the limbs. Skeletal survey showed shortening of the radius, ulna, tibia, and fibula as well as short and broad metacarpals and phalanges, with cone-shaped epiphyses. She had an insulin tolerance test that showed peak growth hormone of 8.4 ng/ml and peak cortisol of 586 nmol/L (normal), which suggested isolated severe growth hormone deficiency. Bone age was indeterminate due to abnormal epiphyses. Whole exome sequencing identified compound heterozygous pathogenic variants in NPR2 which is associated with autosomal recessive AMDM. We did not proceed with the 2nd GH test since the genetic results were confirmative.

CONCLUSION

We described a child with AMDM and severe growth hormone deficiency. Average adult height is estimated to be less than 120 cm. No published data showed that ADMD is related to GHD. Literature reviews showed that 3 AMDM children who received high-dose growth hormone treatment (0.05- 0.1 mg/kg/day) had positive effects on height improvements.

EP_P023

GROWING TOO FAST: A CASE OF TESTOTOXICOSIS

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INTRODUCTION/BACKGROUND

Testotoxicosis is a rare cause of gonadotropin-independent precocious puberty in males due to an activating mutation in the luteinizing hormone (LH)) /choriogonadotropin receptor (LHCGR) gene. This disorder usually presents at 2-4 years old with virilization, advanced bone age, and increased serum testosterone levels above adult ranges, despite low LH and FSH levels.

CASE

A 3-year 2-month-old male presented with complaints of acne, pubic hair, phallic growth, height accelerations, and behavioural issues. His parents were non-consanguineous and had no family history of precocious puberty. On physical examination, his height was 106cm (+2.36 SDS) and weight was 16 kg (+0.77 SDS). He had a muscular body build, a deep voice, and acne. He had Tanner stage 2 pubic hair, stretched penile length was 6 cm (>2 SDS), and both testicular volumes were 5 mls. He did not have café au lait spots. Bone age was 7 years old. Testosterone level was high at 17 nmol/L and gonadotropin-releasing hormone (GnRH) stimulation test revealed a prepubertal response (peak LH 1.44 u/L and FSH 3.02 u/L). Thyroid function tests (FT4: 13.77 pmol/L; TSH: 1.71 m IU/L) and adrenal hormone levels (DHEAS: 0.46 umol/L; 17-OHP: 3.42

nmol/L; cortisol: 400 nmol/L) were within normal ranges. Synacthen test excluded virilising congenital adrenal hyperplasia. Low serum beta-hCG (1.2 mIU/ml) excluded beta-hCG-secreting tumour. Testicular ultrasound showed no sonographic evidence of testicular lesion. Whole exome sequencing identified a heterozygous pathogenic variant c.169A>G (p.Asp564Gly) in LHCGR gene which supports the diagnosis of testotoxicosis.

The child was started on aromatase inhibitor, Anastrozole 1mg daily, and anti-androgen, spironolactone 2 mg/kg BD. At 6 months of treatment, there was a halt in pubertal progression with reduced height velocity from 9 cm/year to 6 cm/year.

CONCLUSION

There was no consensus on the management of this rare condition. Without intervention, the patient will have rapid progressive skeletal maturation and virilization which will result in compromised adult height and psychosocial distress.

EP_P024

EARLY BISPHOSPHONATE TREATMENT IN AN INFANT WITH COL1A1 OSTEOGENESIS IMPERFECTA

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INTRODUCTION/BACKGROUND

Bisphosphonate therapy is the mainstay treatment of patients with OI. It helps to increase bone mass, decrease fracture rate, improve growth and muscle strength as well as improve mobility. Initial studies were performed among older children and adolescents; however, recently, early treatment in infants with moderate-to-severe OI has been shown to be safe. The optimal age of starting is controversial, especially less than 6 months as there is a need to balance the benefits of therapy with the safety of treatment.

CASE

We report an 11-month-old male whose prenatal scan revealed suspicion of skeletal dysplasia. Parents are non-consanguineous and with no family history of frequent fractures or genetic disorders.

He was born term via EMLSCS for intrauterine growth restriction with highly resistant Doppler. The birth history was uneventful. He has low-set ears, macrocephalic with widened anterior fontanelle, triangular facies, and grey sclera, his hips were in a flexed and abducted position with bowed bilateral lower limbs.

The child had bilateral thigh swelling with deformity at birth. Radiological evaluation showed a bilateral femur fracture. He sustained a bilateral humerus fracture at day 12 of life, a left radius fracture at 2 months old and a right humerus fracture at 3 months old. Whole Exome Sequencing test revealed a pathogenic variant of COL1A1 gene.

The child was started on pamidronate at the age of 5 months old with a dose of 0.1mg/kg then the dose was increased to 0.25 mg/kg, and was given 3 consecutive days, monthly then every 2 months. The pamidronate dose was further increased to 0.5 mg/kg for 3 days, given 3 monthly. He tolerated treatment well and no adverse effects were noted. He has had no new fractures since treatment started.

CONCLUSION

OI is a complex disorder and involves multidisciplinary management. Early and appropriate treatment could help increase bone density and prevent recurrent fractures.

EP_P025

DILATED CARDIOMYOPATHY IN A CHILD WITH GRAVES' DISEASE

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INTRODUCTION/BACKGROUND

Individuals diagnosed with Grave's disease typically exhibit symptoms of hyperthyroidism, including chest pain, palpitations, and weight loss. Uncommonly, patients may also develop cardiomyopathy, a complication that is extremely serious and potentially life-threatening. Its occurrence is higher among middle-aged and elderly patients with pre-existing heart conditions. Although rare in children, we must acknowledge this complication because of its high mortality and morbidity rates.

CASE

The report details a case of an 11-year-old female with Graves' disease, thyroid storm, and cardiomyopathy. She had palpitations for almost 2 years, followed by recurrent syncopal attacks for 6 months. Her 'unexplained' syncopal attacks were only provided reassurance when she sought medical attention. Upon her first endocrine review, she was in a hyperthyroid state with bilateral exophthalmos, diffuse goitre with signs of heart failure. The initial thyroid function test showed significantly high FT4 levels of 85.6