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.

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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