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