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

https://doi.org/10.15605/jafes.039.S1.239

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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

https://doi.org/10.15605/jafes.039.S1.240

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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

pmol/L and suppressed TSH. Her TSH Receptor antibody showed elevated levels, and the initial CXR revealed cardiomegaly. She was started with oral carbimazole 30 mg daily, oral propranolol 20 mg 4 times a day, Lugol's iodine 4 drops 4 times a day, and intravenous hydrocortisone 50 mg q 6 hourly. Two anti-failure medications were used to treat her heart failure. Her symptoms improved, and she was discharged with oral carbimazole and oral propranolol.

CONCLUSION

It is important to plan an early definitive therapy in this case to prevent future cardiac decompensation during relapse. Medical practitioners need to be aware of the rare presentations of Graves' disease to avoid delayed diagnosis and treatment.

EP_P026

MATERNAL PREGNANCY LUTEOMA: A RARE CAUSE OF VIRILISATION IN A FEMALE NEWBORN AND MOTHER

https://doi.org/10.15605/jafes.039.S1.241

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INTRODUCTION/BACKGROUND

Virilisation of a female newborn is commonly attributed to congenital adrenal hyperplasia but there are rarer causes that can be maternal in origin. Luteomas, a rare, benign androgen-producing ovarian tumour arising during pregnancy can result in both maternal and fetal virilization.

CASE

We describe a case of a newborn with ambiguous genitalia. This baby was born at 36 weeks via caesarean section for poor progress, weighing 2.8 kilograms at birth. Examination at birth revealed a prominent clitorophallic structure, fused labioscrotal folds but no palpable gonads. Otherwise, on general examination, there were no dysmorphic features or hyperpigmentation and serum electrolytes were normal with no hypoglycaemic episodes. On further assessment, 17 Hydroxyprogesterone (17-OHP) level was not elevated; karyotyping and radiological findings were consistent with a female gender. In hindsight, the mother recollected having signs of virilization, i.e., acneiform eruption on her upper chest and back, hirsutism, and deepening voice since the second trimester. Bilateral unhealthy, friable ovarian tumours were revealed intra-operatively which ruptured on handling. As the nature of the tumours was suspicious of malignancy, bilateral oophorectomy was done. Maternal beta human chorionic gonadotrophin (b-HCG) and alpha-fetoprotein (AFP) levels were elevated. The histopathological examination of the ovarian mass confirmed the diagnosis of pregnancy luteoma.

CONCLUSION

This case attests to the fact that rare causes of virilisation in a female baby cannot be overlooked. We thus need to be vigilant and have a high index of suspicion of maternal pregnancy luteomas as a possible cause of virilisation in a female baby.

EP_P027

PAEDIATRIC GRAVES' DISEASE AND DEFINITIVE TREATMENT

https://doi.org/10.15605/jafes.039.S1.242

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INTRODUCTION

Paediatric Graves' disease (GD) is managed by antithyroid drugs (ATD), radioactive iodine (RAI) or thyroid surgery. This study aimed to describe the characteristics and outcomes of paediatric patients who received definitive therapy.

METHODOLOGY

Children and adolescents diagnosed with GD from 2012 to 2024 at the University Malaya Medical Centre were included in this retrospective review.

RESULT

A total of 37 patients were referred and diagnosed with GD; majority (78.4%) were female. Definitive therapy was given to 48%: 5 (35%) had total thyroidectomy and 9 (64%) received RAI. They had an average of four relapses during the disease. On average, the patients received ATD for 4.37 ± 2.28 years prior to the definitive treatment. The main factor in determining the choice of treatment was the size of the goitre. The mean goitre size for the RAI group was 21.68 ± 7.9 g, compared to 76.7 ± 22.88 g for the thyroidectomy group. Mean age in the RAI group was 15.53 ± 1.23 years. The youngest patient was 8 years old. Mean RAI dose was 9.3 ± 0.66 mCi. Six patients achieved hypothyroidism within 2.17 ± 2.44 months, while 1 patient achieved hypothyroidism 8 months post-RAI. Three had relapses post-RAI. Two patients required a second RAI one year later and achieved hypothyroidism within 2 to 4 weeks. Those who required a second RAI were given lower RAI doses initially (mean 5.6 ± 2.2 mCi). The mean age of patients who underwent total