testing was positive for NGLY-1 gene mutation, which is associated with global developmental delay, movement disorders, seizures, liver disease and alacrimia.

CASE 2

MI presented with being "easily tired" and hyperpigmentation since the age of 6 years. The endocrine team was consulted due to low cortisol. Investigations revealed ACTH >278 pmol/L, normal 17-OHP and flat response following ACTH stimulation test. Adrenal CT was normal. Genetic studies came back positive for ABCD1 mutation, a condition of adrenomyeloneuropathy, associated with progressive lower limb weakness and spasticity in the third or fourth decade of life.

With hydrocortisone replacement and fludrocortisone therapy, LA and MI improved noticeably by decreasing skin hyperpigmentation.

CONCLUSION

Non-specific presentations of PAI and the rising numbers of genetic aetiologies discovered warrant genetic testing in affected individuals. This will facilitate prompt diagnosis based on clinical features and prognostication. It provides opportunities for tailored patient management, family counselling and heightened surveillance of possible co-morbidities.

EP_P005

CO-OCCURRENCE OF OCULAR MYASTHENIA GRAVIS, TYPE 1 DIABETES MELLITUS AND GRAVES' THYROTOXICOSIS IN A YOUNG CHILD

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INTRODUCTION

Ocular myasthenia gravis (OMG), type 1 diabetes mellitus (T1DM) and Graves' thyrotoxicosis (GT) are autoimmune conditions in childhood. However, co-occurrence and sequential onset of these diagnoses is uncommon. It could signify a spectrum of polyglandular autoimmune syndrome type 2 with polygenic inheritance.

CASE

A 6-year-2-month-old female presented with progressive drooping of both eyelids for the past two months. Chest CT showed normal thymus, and the anti-acetylcholine receptor was positive (4.89 nmo/L) [reference value (RV) <0.25 nmol/L]. The diagnosis of ocular myasthenia gravis was ascertained. She responded well to pyridostigmine.

Nonetheless, she presented again at 8 years and 11 months old with polyuria, polydipsia and nocturia for three weeks, and significant weight loss. She had severe DKA requiring intensive care. Biochemical markers were consistent with T1DM: low C-peptide (57 pmol/L), low insulin (4.3 pmol/L), positive anti-ICA (45.61 IU/mL) (RV <28 IU/mL) and anti-GAD (98.18 IU/mL) (RV <17 IU/mL), while anti-IA2 was low (<2.5 IU/mL) (RV <28 IU/mL).

While her initial thyroid function was normal, thyroid auto-antibody screening was positive for anti-TPO (222 IU/mL) (RV <35 IU/mL). Following multiple daily insulin injections, her glycaemic control and weight gradually improved. Ten months later, at 9 years and 10 months old, her HbA1c worsened, and her mother reported a sudden increase in insulin needs with weight loss. She manifested symptoms of hyperthyroidism and was found to have tachycardia, tremors and diffuse goitre. She did not have Graves' ophthalmopathy; bilateral ptosis remained the same. Anti-TSH receptor antibodies were significantly positive (26.30 IU/L) (RV <1.75 IU/L).

CONCLUSION

OMG in young children is rarely associated with T1DM. Screening for diabetes auto-antibodies should be considered. In T1DM children, GT should be taken into account when there is unexplained weight loss or deterioration in glycaemic control.

EP_P006

AN UNUSUAL CASE OF MASSIVE NEONATAL GOITRE

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INTRODUCTION

Congenital hypothyroidism occurs in one out of 3000 live births in Malaysia. Over 95% of the cases have no clinical manifestations at birth. In this peculiar case, we present a patient with massive neonatal goitre with congenital hypothyroidism.

CASE

An 8-month-old male was diagnosed prenatally to have a neck mass on a detailed scan at 37 weeks. It was reported to be highly vascularised with possible goitre. Maternal biochemical markers showed euthyroid status, but neck ultrasound revealed multinodular goitre. There were no suggestive risk factors for iodine deficiency. Because of the possibility of airway compression, caesarean section was recommended and he was delivered via ex-utero

intrapartum treatment (EXIT procedure) at 38 weeks and 4 days. The infant was intubated and ventilated for respiratory distress. Newborn examination showed an anterior neck mass measuring 2 x 2 cm from midline to the left, with otherwise unremarkable systemic examination. Postnatal computed tomography of the neck revealed massive goitre causing airway compression and oesophageal narrowing from the oropharynx until the thoracic inlet. Laboratory studies supported the diagnosis of congenital hypothyroidism (TSH 37.41 μIU/mL) with possible thyroid dyshormogenesis. Treatment was initiated with oral levothyroxine 50 mcg daily. Serial ultrasound imaging showed a gradual reduction with resolved mass effect and airway compression.

CONCLUSION

Prompt diagnosis and meticulous thyroid replacement therapy led to significant regression of goitre to a more functional size. Rational intervals of clinical and biochemical evaluation are crucial to ensure optimum growth and neurodevelopmental outcomes.

EP_P007

INFANTILE HYPOCALCAEMIC SEIZURE AND VITAMIN D DEFICIENCY

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INTRODUCTION/BACKGROUND

Growing evidence suggests that vitamin D is essential for maternal and child health in many aspects. Nevertheless, a severe manifestation of vitamin D deficiency in the form of hypocalcaemic seizures continue to occur among Malaysian infants.

METHODOLOGY

A descriptive cross-sectional study was performed in the Paediatric Endocrinology Unit, Hospital Putrajaya. Records of all infants with hypercalcaemic seizures managed by our unit between January 2015 until April 2024 were retrieved from the electronic database system. Causes of hypercalcaemic seizure among this group of patients were identified. Further clinical, biochemical and hormonal results to assess the calcium-vitamin D-PTH axis were analysed.

CASE

A total of 24 patients were treated for hypercalcaemic seizures during the study period. Sixteen patients were male. Majority (75%) of the patients had hypercalcaemic seizures secondary to vitamin D deficiency, while 25% had hypoparathyroidism.

Among the group of hypercalcaemic seizures secondary to vitamin D deficiency, the median age of presentation was 8 weeks. Their mean corrected calcium, phosphorus, magnesium and ALP on presentation were 1.4 mmol/L, 2.35 mmol/L, 0.73 mmol/L and 690 U/L respectively. Mean iPTH and vitamin D levels of the patients were 19 pmol/L and 16.4 nmol/L, respectively. Maternal vitamin D levels were available for 7 mothers, showing a low mean value of 28.7 nmol/L.

All patients with hypoparathyroidism in this study had concomitant vitamin D deficiency or insufficiency. The median age of presentation was 3.5 weeks. Their mean corrected calcium, phosphorus, magnesium and ALP upon presentation were 1.64 mmol/L, 2.80 mmol/L, 0.64 mmol/L and 384 U/L, respectively. Mean iPTH and vitamin D levels were 2.3 pmol/L and 41.7 nmol/L, respectively.

CONCLUSION

Vitamin D deficiency or insufficiency was present in all patients in our study population. Vitamin D deficiency remains the predominant cause of hypocalcaemic seizure. Thus, vitamin D supplementation for all pregnant women should be encouraged as part of routine care. All infants during the first year of life should be encouraged to receive an oral vitamin D supplementation.

EP_P008

MALIGNANT GONADAL TUMOUR IN TRISOMY 21 WITH COMPLETE SEX REVERSAL

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INTRODUCTION/BACKGROUND

Trisomy 21 is a chromosomal disorder with a high incidence worldwide. It is associated with characteristic physical features, delay in development and some congenital organ defects. However, disorder of sex development (DSD) is not usually seen in patients with Down Syndrome.

CASE

A 14-year-old phenotypically female with Down syndrome presented with a two-month history of progressive abdominal swelling, constipation and weight loss. At birth, typical Down Syndrome facial features were present and the female gender was assigned. The karyotype result of 86 cells showed 47XY, +21 with +SRY gene via FISH study.