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