CONCLUSION

Early and accurate diagnosis is essential for optimal management requiring a multidisciplinary approach including endocrinologists and surgeons.

EP_P018

ATYPICAL PRESENTATION OF SEVERE PROGNATHISM IN PATIENT WITH CONGENITAL ADRENAL HYPERPLASIA

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INTRODUCTION

Congenital adrenal hyperplasia (CAH) is a group of autosomal recessive disorders due to mutation in the genes that regulate adrenal steroidogenesis. The commonest form is due to 21-OH enzyme deficiency in which the classic form is divided into salt-losing or simple virilizing types.

CASE

A 20-year-old Malay male has been under our follow-up since his early infancy. He was diagnosed to have a salt-losing form of CAH in the neonatal period as he had adrenal crises associated with skin hyperpigmentation. The boy was treated with oral hydrocortisone 10-15 mg/m^2/day and oral fludrocortisone 150 mcg to 200 mcg per day. However, starting from the age of five, he experienced medication adherence issues due to inadequate supervision and logistical challenges.. At the age of 15 years, he was diagnosed to have testicular adrenal rest tumour. He later complained of progressive difficulty chewing his food due to the development of mandibular hyperplasia or prognathism. He was treated with high dose glucocorticoid and ketoconazole to control his hyperandrogenism and referred to the maxillofacial team for further management.

CONCLUSION

Despite being detected early and managed promptly, the outcome of treatment relies strongly on the compliance of the patient. Non-adherence to medication may lead to unforeseen detrimental complications which could worsen the long-term prognosis.

EP_P019

AN ADOLESCENT WITH UNEXPLAINED DIABETES MELLITUS AND ASSOCIATED CONGENITAL GENITOURINARY ANOMALIES: A CASE REPORT

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INTRODUCTION

Maturity-onset diabetes of the young (MODY) is a rare form of diabetes found in Malaysia and worldwide, with at least 14 recognized types linked to different genetic mutations. MODY Type 5 (MODY 5) is caused by mutations in the HNF1 gene, which encodes hepatocyte nuclear factor 1 beta. This condition is characterised by diabetes and various extra-pancreatic features, including abnormalities in the kidneys and urogenital system.

CASE

We present a young female patient who initially presented with diabetes mellitus, later diagnosed with congenital renal anomalies, including a right single kidney and Müllerian anomalies. Her strong family history of diabetes and renal issues underscores the importance of recognising this diagnosis. A diagnosis of a monogenic form of diabetes was suspected since she had an onset of diabetes at the age of 13 years, absent acanthosis nigricans, positive family history with onset less than 30 years old and renal/Mullerian duct abnormalities. A targeted gene panel, whole exome sequencing panel, was performed to test for MODY. The panel indicated a positive result for the HNF1B gene mutation c.766C>T (p. Pro256Ser), resulting in an amino acid change at codon 256 from proline to serine (p. Pro256Ser).

CONCLUSION

This case highlights the importance of recognising the potential overlap between diabetes, renal disorders and Müllerian anomalies, particularly in young patients without a clear family history. Genetic testing is the gold standard for diagnosing MODY, but access and affordability can be challenging for patients in Malaysia.