Paediatrics E-Poster

EP_P001

A CHILD WITH AN AGGRESSIVE FUNCTIONAL ADRENOCORTICAL CARCINOMA

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INTRODUCTION
Adrenocortical carcinomas, among the rarest and most aggressive paediatric endocrine tumours, manifest with diverse symptoms like virilization, Cushing's syndrome, or both.

CASE
We present a case of functional adrenocortical carcinoma in a female aged 6 years and 7 months, who initially presented with hypertensive encephalopathy and hypokalaemic hypochloremic metabolic alkalosis, which resolved with symptomatic treatments. Ten months later, she presented with frank Cushing's syndrome, refractory hypertension, generalised virilization, extensive skin fungal infection and severe backache. Breast cancer was diagnosed in her maternal aunt. Hormonal tests showed non-ACTH dependent hypercortisolism and marked elevation of androgens. Computed tomography revealed a large left suprarenal mass, with multi-focal liver lesions and lung nodules suggestive of distant metastasis, left renal vein thrombosis and multiple osteoporotic vertebral fractures.
A clinical diagnosis of stage 4 functional adrenocortical carcinoma was made. While complete surgical removal of the tumour is the gold standard, it was not feasible immediately due to the substantial size of the tumour and presence of distant metastases. Neo-adjuvant chemotherapy was started. Mitotane and ketoconazole were introduced concomitantly to control hypercortisolism, with initial success. Hydrocortisone replacement was needed for a short period when there was a rapid decline in cortisol levels following chemotherapy. Unfortunately, with poor commitment from the family, the disease advanced rapidly with worsening lung and liver metastases. Following a family conference, the parents opted for palliative treatment with mitotane monotherapy, and the child was transferred to the district hospital for comfort care.

CONCLUSION
Medical treatment is useful in controlling the symptoms of severe hypercortisolism. Steroid replacement may be needed with the use of adrenolytic agent. Adrenocortical carcinoma is aggressive and a high index of suspicion is needed for early diagnosis.

EP_P002

CONGENITAL HYPERINSULINISM SECONDARY TO ABCC8 MUTATION: A CASE STUDY

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INTRODUCTION
Congenital hyperinsulinism (CHI) results in persistent hypoglycaemia beyond infancy. Mutations in the ABCC8 and KCNJ11 genes are the most common aetiologies of congenital hyperinsulinism that leads to inappropriate insulin secretion irrespective of hypoglycaemia.

CASE
A 3-month-old male was referred to the clinic for persistent hypoglycaemia. He was born with a birth weight of 4480 g by elective caesarean section for macrosomia. His mother had an uneventful antenatal period and had a normal OGTT during the pregnancy.

Post-delivery, he was initially well until when he developed hypoglycaemia at 28 hours of life. He was transferred to the NICU from the postnatal ward. Hypoglycaemia was persistent requiring high glucose delivery rate up to 10 mg/kg/min. Glucagon infusion was started and was difficult to wean. Hypoglycaemic hyperinsulinaemia was confirmed at day 21 of life, with concomitant results of insulin 3.4 m IU/L and random blood glucose 1.8 mmol/L. Diazoxide was started. Glucagon infusion was stopped at day 26 of life, and the baby was discharged.

He was born from a non-consanguineous marriage. He was the second child in the family. His parents and older sister were well and healthy. There was no known history of neonatal hypoglycaemia nor early-onset diabetes in his family.

At 2 years, the patient still required diazoxide with episodes of hypoglycaemia when feeding was delayed. He was referred to the genetic team. Further investigation revealed compound heterozygous mutations at the ABCC8 gene (likely autosomal recessive type) which were c.2992C>T (path); similar to his mother, and another mutation c.4607C>T (VUS). The father did not have any ABCC8 mutations.

CONCLUSION

Clinical suspicion of CHI should be highly considered in macrosomic babies with persistent hypoglycaemia in the absence of maternal diabetes. Expedited genetic study should be considered to assist clinical management.

EP_P003

EXTREME SPECTRUM OF DYSGLYCAEMIA IN TWO SISTERS WITH CDKN1C MUTATION

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INTRODUCTION/BACKGROUND

CDKN1C mutation is mainly associated with Beckwith-Wiedemann syndrome (BWS), an overgrowth disorder, and IMAGe syndrome, an undergrowth disorder. In both conditions, hypoglycaemia can be part of the presenting features. Defect in this gene has not been directly linked with diabetes; however, evidence supports the hypothesis that loss of CDKN1C function leads to increased beta cell proliferation and causes hypoglycaemia. Some hypotheses also suggest that overactivity of CDKN1C gene results in the opposite phenotype: decreased proliferation of beta cells leading to reduced insulin production and onset of diabetes.

CASE

We report two cases of Malay siblings who presented with dysglycaemia of opposite ends of the spectrum. Both siblings were not dysmorphic with normal BMI. The elder sister, now 21 years, presented at the age of 5 years with hyperglycaemic symptoms and was treated as type 1 diabetes. Her diabetes autoantibodies were negative. She has been on insulin with an average HbA1c of 8%.

The younger sister presented at 16 years with frequent postprandial hypoglycaemia episodes associated with recurrent cramps and muscle weakness. Investigations showed hyperinsulinaemic hypoglycaemia with concurrent hypokalaemia. PET scan and MRI were negative for insulinoma. Oesophagogastroduodenoscopy did not find any suspicious gastrointestinal lesions. Munchausen by proxy was excluded. Her symptoms improved with oral diazoxide but not fully resolved. She is dependent on potassium supplements. Genetic testing on both sisters revealed same mutation at the CDKN1C gene, reported as variant of uncertain significance (VUS).

CONCLUSION

In our patients, CDKN1C mutation manifested with polar opposites of dysglycaemia. The molecular function of the gene in glucose homeostasis is yet to be defined.

EP_P004

UNVEILING THE MYSTERIES: GENETIC PERSPECTIVE ON PRIMARY ADRENAL INSUFFICIENCY

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INTRODUCTION

Primary adrenal insufficiency (PAI) in childhood is rare and potentially life-threatening. The most common cause is congenital adrenal hyperplasia (CAH) resulting from 21-hydroxylase deficiency. Advancements in molecular genetics have revealed more genetic mutations causing PAI, which helps in explaining associated clinical features and prognosis. Clinical data and genetic tests were reviewed for two patients who presented with PAI.

CASE 1

LA presented with hypotonia and global developmental delay at the age of 1 year, with normal brain MRI/MRA and inborn error of metabolism (IEM) workup. She defaulted follow-ups until she presented again with generalized hyperpigmentation to dermatology at 3 years. Workup showed ACTH >278 pmol/L, normal 17-hydroxyprogesterone (17-OHP), and no rise in cortisol (<1.8 nmol/L) on ACTH stimulation test. Adrenal ultrasound was normal. Genetic