PE-13

INS GENE MUTATION IN NEONATAL DIABETES MELLITUS

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INTRODUCTION

Neonatal diabetes mellitus (NDM) is a rare disorder with estimated prevalence of 1:90000 to 1:160000 live births. They usually present with persistent hyperglycemia within the first 6 months of life and some may persist up to 12 months of life. Approximately 80% of NDM patients have genetic mutation and more than 20 genes have been identified. Amongst these, KCNJ11 and ABCC8 genotype have been the most common cause of NDM accounting for 38.2%. INS gene mutation is also a known cause of NDM manifesting as damage to the pancreatic beta cells. Studies have shown that heterozygous autosomal dominant mutation is the 2nd most common cause of NDM. The average age for diagnosis in INS gene mutation NDM patients are 10 weeks old and 30% of them presented with DKA. INS gene mutation causes misfolding of the insulin protein which leads to the damage of the beta cells therefore the treatment of choice is insulin therapy. They usually present with low birth weight resulting from IUGR. INS gene mutation is known to cause both Transient Neonatal Diabetes Mellitus (TNDM) and Permanent Neonatal Diabetes Mellitus (PNDM) of which PNDM is more common and accounts for 20% of NDM. Here we present 2 different patients with homozygous mutations of INS gene; for which we want to emphasize the importance of genetic testing in diagnosing different types of NDM and its role in management.

PE-14

CONGENITAL HYPOPITUITARISM PRESENTING DURING THE NEONATAL PERIOD: A CASE REPORT

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INTRODUCTION

Congenital hypopituitarism may present as isolated or combined pituitary hormone deficiencies. The incidence reported is in between 1 in 4000 to 1 in 10,000. It may occur due to developmental defects of the pituitary gland or genetic mutations. Prompt recognition during infancy and appropriate replacements of hormone deficiencies are essential. However, the diagnosis can be challenging due to its variable presentation and non-specific symptoms.

RESULTS

The index case is a 2-year-old girl who presented with recurrent hypoglycemia and salt-losing crisis during the neonatal period. On examination, she has soft dysmorphism (widened sagittal sutures, down-slating palpebral fissure, high arch palate, flat nasal bridge). Critical sampling taken during the hypoglycemia episode (blood glucose 1.5 mmol/L) revealed extremely low serum cortisol (3.8 nmol/L) and growth hormone (0.02 mcg/L) levels. She also had central hypothyroidism characterized by a low free-thyroxine but inappropriately normal TSH (TSH 5.68 miU/L, FT4 7.12 pmol/L). She was started on hydrocortisone, L-thyroxine and daily subcutaneous recombinant growth hormone (GH) therapy. GH was started early for the hypoglycaemia. There was no clinical or biochemical picture of diabetes insipidus. Magnetic resonance imaging (MRI) of pituitary done at 10 months of age shows a hypoplastic pituitary gland with a normal infundibulum and absence of high signal changes in the posterior lobe of the gland.

CONCLUSION

Congenital hypopituitarism can be a life-threatening condition manifesting during the neonatal period with hypoglycaemia and salt-wasting crisis. Early diagnosis and treatment are essential.