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Hyperinsulinaemic Hypoglycaemia

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Abstract

Pancreatic beta-cell dysfunction in the newborn period can lead either to hypoglycaemia or hyperglycemia. Hypoglycaemia occurs due to inappropriate insulin secretion which leads to Hyperinsulinaemic hypoglycaemia (HH). Hyperglycemia occurs due to too little insulin secretion and this lead to neonatal diabetes mellitus (NDM). Genetic defects in nine different genes have been described which lead to HH. On the other NDM can be due to defects in a large number of genes.

Insulin secretion from pancreatic β-cells is tightly regulated to keep fasting blood glucose concentrations within the normal range (3.5-5.5mmol/L). Hyperinsulinaemic hypoglycaemia (HH) is a heterozygous condition in which insulin secretion becomes unregulated and its production persists despite low blood glucose levels. It is the most common cause of severe and persistent hypoglycaemia in neonates and children. The most severe and permanent forms are due to congenital hyperinsulinism (CHI). Recent advances in genetics have linked CHI to mutations in 9 genes that play a key role in regulating insulin secretion (ABCC8, KCNJ11, GLUD1, GCK, HADH, SLC16A1, UCP2, HNF4A and HNF1A). Mutations in genes ABCC8 (SUR1 subunit) and KCNJ11 (Kir6.2 subunit) are the most common cause of CHI. Both the ABCC8/KCNJ11 genes are localized on chromosome 11p15.1. The most severe forms of CHI are due to recessive inactivating (loss of function) mutations in ABCC8 and KCNJ11 leading to unregulated insulin secretion despite severe hypoglycaemia. Dominant inactivating mutations in ABCC8 and KCNJ11 usually cause a milder form of CHI which is responsive to diazoxide. However, medically un-responsive forms have also been reported.

Histologically, CHI can be divided into 3 types: diffuse, focal and atypical. Given the biochemical nature of HH (non-ketotic), a delay in the diagnosis and management can result in irreversible brain damage. Therefore it is essential to diagnose and treat HH promptly. Advances in molecular genetics, imaging methods (18F-DOPA PET-CT), medical therapy and surgical approach (laparoscopic surgery) have completely changed the management and improved the outcome of these children.