Diazoxide-resistant focal hyperinsulinism due to Kir6.2 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Diazoxide-resistant focal hyperinsulinism due to Kir6.2 deficiency. ORPHA:276603

A rare, congenital, isolated hyperinsulinism disorder characterized by diazoxide unresponsive recurrent episodes of hyperinsulinemic hypoglycemia resulting from an excessive insulin secretion by the pancreatic bêta-cells due to Kir6.2 deficiency. Hypoglycemia may lead to variable clinical manifestation, ranging from asymptomatic hypoglycemia revealed by routine blood glucose monitoring to macrosomia at birth, mild to moderate hepatomegaly and life-threatening hypoglycemic coma or status epilepticus, further leading to poor neurological outcome.