Autosomal recessive hyperinsulinism due to Kir6.2 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive hyperinsulinism due to Kir6.2 deficiency. ORPHA:79644

A rare, congenital, isolated hyperinsulinism disorder characterized by neonatal presentation of severe refractory hypoglycemia in the first two days of life, with limited response to medical management, sometimes requiring pancreatic resection. Newborns are often large for gestational age with mild to moderate hepatomegaly and diffuse form of hyperinsulinism due to Kir6.2 deficiency. Persistent hypoglycemia, hyperglycemia and type1 diabetes mellitus may develop later in life. Life-threatening hypoglycemic coma or status epilepticus have also been associated.