Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive hyperinsulinemic hypoglycemia due to Kir6.2 deficiency
Autosomalna recesywna hipoglikemia hiperinsulinemiczna z powodu niedoboru Kir6.2

Kod ORPHA
79644

Kod OMIM
601820

Kod ICD10
E16.1

Kod ICD11
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*Źródło

orphanet