Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Choroba

Synonimy
Hyperinsulinemic hypoglycemia due to Kir6.2 deficiency, diazoxide-resistant focal form
Hipoglikemia hiperinsulinemiczna z powodu niedoboru Kir6.2, postać ogniskowa oporna na diazoksyd

Kod ORPHA
276603

Kod OMIM
601820

Kod ICD10
E16.1

Kod ICD11
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*Źródło

orphanet