

Opis choroby *

Definicja

A form of diazoxide-sensitive diffuse congenital hyperinsulinism due to HNF4A deficiency and, characterized by macrosomia, transient or persistent hyperinsulinemic hypoglycemia (HH), responsiveness to diazoxide and a propensity to develop maturity-onset diabetes of the young subtype 1 (MODY).

Dane

Klasyfikacja

Choroba

Synonimy

Hyperinsulinemic hypoglycemia due to HNF4A deficiency

Hipoglikemia hiperinsulinemiczna z powodu niedoboru HNF4A

Wrodzony hiperinsulinizm z powodu niedoboru HNF4A

Kod ORPHA

263455

Kod OMIM

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Kod ICD10

E16.1

Kod ICD11

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*Źródło

orphanet