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