Opis choroby *

Definicja

A form of diazoxide-sensitive diffuse hyperinsulinism (DHI) characterized by hypoglycemic episodes that are usually mild, escaping detection during infancy, and usually a good clinical response to diazoxide, (but some are diazoxide resistant). Autosomal dominant hyperinsulinism due to Kir6.2 deficiency usually has a milder phenotype when compared to that resulting from recessive K+ (K-ATP) channel mutations (Recessive forms of diazoxide-resistant hyperinsulinism).

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant hyperinsulinemic hypoglycemia due to Kir6.2 deficiency
Autosomalna dominująca hipoglikemia hiperinsulinemiczna z powodu niedoboru Kir6.2
Dominant KATP hyperinsulinism due to Kir6.2 deficiency

Kod ORPHA
276580

Kod OMIM
601820

Kod ICD10
E16.1

Kod ICD11
-

---

*Źródło

orphanet