Opis choroby *

Definicja

A rare diffuse form of congenital hyperinsulinism characterized by an excessive/ uncontrolled insulin secretion (inappropriate for the level of glycemia), chronic hyperammonemia and recurrent episodes of hypoglycemia induced by fasting and protein rich meals. Epilepsy and cognitive deficit, which are unrelated to hypoglycemia but possibly related to the chronic hyperammonemia, may also occur. This disorder is usually responsive to diazoxide treatment.

Dane

Klasyfikacja
Choroba

Synonimy
HI/HA syndrome
Zespół HI/HA

Kod ORPHA
35878

Kod OMIM
606762

Kod ICD10
E72.8

Kod ICD11
5C50.AY

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*Źródło

orphanet