Opis choroby *

Definicja

A rare genetic neurological disorder characterized by subacute encephalopathy with confusion, seizures, and movement disorder, often following a history of febrile illness. Imaging may reveal bilateral lesions in the basal ganglia. The disease usually becomes symptomatic in childhood and is life-threatening if left untreated, but symptoms can be reversed and progression prevented by treatment with high doses of biotin and thiamine.

Dane

Klasyfikacja
Choroba

Synonimy
BBGD
BBGD
BTBGD
Choroba zwojów podstawnych wrażliwa na biotynę-tiaminę
BTBGD
Biotin-responsive basal ganglia disease

Kod ORPHA
65284

Kod OMIM
607483

Kod ICD10
G93.8

Kod ICD11
5C63.Y

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*Źródło

orphanet