

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

[*Źródło](#)

orphanet