Opis choroby *

Definicja

Childhood-onset spasticity with hyperglycinemia is a rare neurometabolic disease characterized by a childhood onset of progressive spastic ataxia associated with gait disturbances, hyperreflexia, extensor plantar responses and non-ketotic hyperglycinemia typically revealed by biochemical analysis. Additional signs of upper extremity spasticity, dysarthria, learning difficulties, poor concentration, nystagmus, optic atrophy and reduced visual acuity may also be associated.

Dane

Klasyfikacja Synonimy

Choroba Childhood-onset spasticity with variant non-

ketotic hyperglycinemia

Spasticity-ataxia-gait anomalies syndrome Childhood-onset spasticity with variant non-

ketotic hyperglycinemia

Spasticity-ataxia-gait anomalies syndrome

Kod ORPHA

401866

Kod OMIM 616859

Kod ICD10

E88.8

Kod ICD11

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*Źródło

orphanet