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 Choroba	ketotic hyperglycir Spasticity-ataxia-ga Childhood-onset s ketotic hyperglycir	ait anomalies syndrome pasticity with variant non-
Kod ORPHA	Kod OMIM	Kod ICD10

616859

401866

Kod ICD10 E88.8

Kod ICD11

<u>*Źródło</u>

orphanet