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

Synonimy
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