Opis choroby *

Definicja

A rare genetic neurometabolic disease characterized by childhood onset of global developmental delay, progressive spastic ataxia leading to loss of independent ambulation, and elevated plasma levels of glutamine. Optic atrophy, tremor, and dysarthria have also been reported. Brain imaging may show cerebellar atrophy.

Dane

Klasyfikacja
Choroba

Kod ORPHA
557056

Kod OMIM
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Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet