

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

-

Kod ICD10

E88.8

Kod ICD11

-

*Źródło

orphanet