Opis choroby *

Definicja

A rare genetic neurodegenerative disease characterized by sudden onset of progressive motor deterioration and regression of developmental milestones. Manifestations include dystonia and muscle spasms, dysphagia, dysarthria, and eventually loss of speech and ambulation. Brain MRI shows predominantly striatal abnormalities. The disease is potentially associated with a fatal outcome.

Dane

Klasyfikacja
Choroba

Synonimy
Lenk-Ploski syndrome
Zespół Lenkego i Ploskiego

Kod ORPHA
497906

Kod OMIM
617054

Kod ICD10
G31.8

Kod ICD11
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*Źródło

orphanet