Opis choroby *

Definicja

A rare genetic neurological disorder characterized by postnatal microcephaly, hypotonia during infancy followed in most cases by progressive spasticity mainly affecting the lower limbs, and spastic diplegia or paraplegia, intellectual disability, delayed or absent speech, and dysarthria. Seizures and mildly dysmorphic features have been described in some patients.

Dane

Klasyfikacja
Choroba

Kod ORPHA
477673

Kod OMIM
616281

Kod ICD10
G11.4

Kod ICD11
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*Źródło

orphanet