

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