

Opis choroby *

Definicja

A rare, genetic, neurological disorder characterized by childhood-onset severe myoclonic and tonic-clonic seizures and early-onset ataxia leading to severe gait disturbances associated with normal to slightly diminished cognition. Scoliosis, diffuse muscle atrophy and subcutaneous fat loss, as well as developmental delay, may be associated. Brain MRI may reveal complete agenesis of the corpus callosum, ventriculomegaly, interhemispheric cysts, and simplified gyration (frontally).

Dane

Klasyfikacja

Choroba

Synonimy

EPM9

EPM9

PME typu 9

Postępująca padaczka miokloniczna

spowodowana niedoborem LMNB2

PME type 9

Progressive myoclonic epilepsy due to LMNB2
deficiency

Progressive myoclonus epilepsy type 9

Kod ORPHA

457265

Kod OMIM

616540

Kod ICD10

G40.3

Kod ICD11

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*Źródło

orphanet