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
-

---

*Źródło

orphanet