Opis choroby *

Definicja

A rare neurologic disease characterized by neonatal hypotonia, global developmental delay, feeding difficulties, and often seizures or seizure-like episodes. Other frequently observed signs and symptoms include variable dysmorphic features, myopathic facies, respiratory problems, and visual abnormalities, such as strabismus or esotropia. Brain imaging may show delayed myelination and other white matter abnormalities.

Dane

Klasyfikacja
Choroba

Kod ORPHA
438213

Kod OMIM
616158

Kod ICD10
G40.4

Kod ICD11
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*Źródło

orphanet