Opis choroby *

Definicja

A rare genetic neurological disorder characterized by postnatal onset of severe global developmental delay, profound mental retardation, progressive microcephaly, progressive spasticity evolving into spastic quadriplegia with joint contractures, generalized seizures, and irritability. Severe choreoathetosis and dysmorphic features are absent. Brain imaging shows progressive cerebellar atrophy followed by cerebral atrophy affecting both white and grey matter, but no pontine involvement.

Dane

Klasyfikacja
Choroba

Synonimy
PCCA
PCCA

Kod ORPHA
247198

Kod OMIM
615851

Kod ICD10
G31.8

Kod ICD11
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*Źródło

orphanet