Opis choroby *

Definicja

A rare disorder characterized by early-onset progressive encephalopathy with migrant, continuous myoclonus. Three cases have been reported. The focal continuous myoclonus appeared during the first months of life. Prolonged bilateral myoclonic seizures and generalized tonic-clonic seizures occurred later. Subsequently, a progressive encephalopathy with hypotonia and ataxia appeared. Cortical atrophy was revealed by computed tomography (CT) scan and magnetic resonance imaging (MRI). The aetiology is unknown.

Dane

Klasyfikacja
Choroba

Kod ORPHA
1943

Kod OMIM
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Kod ICD10
G40.4

Kod ICD11
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*Źródło

orphanet