Opis choroby *

Definicja

A rare, X-linked, syndromic intellectual disability disease characterized by neonatal hypertonia which evolves to hypotonia and an exaggerated startle response (to sudden visual, auditory or tactile stimuli), followed by the development of early-onset, frequently refractory, tonic or myoclonic seizures. Progressive epileptic encephalopathy, intellectual disability, and psychomotor development arrest, with subsequent decline, may be additionally associated.

Dane

Klasyfikacja
Choroba

Kod ORPHA
163985

Kod OMIM
300607

Kod ICD10
G25.8

Kod ICD11
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*Źródło

orphanet