

Hiperekpleksja - padaczka

Kod Orpha: 163985 Kod OMIM: 300607

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

-

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.