

# 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

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[\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.