Opis choroby *

Definicja

A rare genetic neurological disorder characterized by neonatal onset of rigidity and intractable seizures, with episodic jerking already beginning in utero. Affected infants have small heads, remain visually inattentive, do not feed independently, and make no developmental progress. Frequent spontaneous apnea and bradycardia usually culminate in cardiopulmonary arrest and death in infancy, although some cases were described with a milder clinical course and survival into childhood.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Lethal neonatal rigidity-multifocal seizure syndrome
Zespół śmiertelnej spastyczności noworodków i wieloogniskowych ataków padaczkowych

Kod ORPHA
435845

Kod OMIM
614498

Kod ICD10
G40.4

Kod ICD11
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*Źródło

orphanet