

Opis choroby *

Definicja

A rare infantile epilepsy syndrome characterized by early onset of seizures of variable type and severity, potentially associated with a spectrum of clinical signs and symptoms including delay or lack of psychomotor development, intellectual disability, poor or absent speech development, behavioral abnormalities, hypotonia, movement disorders, spasticity, microcephaly, and dysmorphic facial features, among others. Brain imaging findings are also variable and may include cerebral atrophy or white matter abnormalities.

Dane

Klasyfikacja

Choroba

Synonimy

Non-specific EOEE

Niezdeterminowana EOEE

Undetermined EOEE

Undetermined early-onset epileptic encephalopathy

Kod ORPHA

442835

Kod OMIM

617830

Kod ICD10

G40.4

Kod ICD11

-

*Źródło

orphanet