

Heterotopia neuronalna typu guzkowego

Kod Orpha: 2149 Kod OMIM: 617201

Opis choroby *

Definicja

A rare non-syndromic cerebral malformation due to abnormal neuronal migration characterized by clusters of disorganized neurons in abnormal locations such as periventricular and subcortical. The extent of the lesions ranges from isolated single to bilateral confluent nodules. Pediatric patients typically show variable degrees of developmental delay, intellectual disability, and intractable epilepsy, and concomitant cerebral and/or systemic malformations are frequent. Milder forms may present with onset of seizures in adulthood.

Dane

Klasyfikacja

Wada morfologiczna

Kod ORPHA

2149

Kod OMIM

617201

Kod ICD10

Q04.8

Kod ICD11

LA05.5Y

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.