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

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*Źródło

orphanet