Opis choroby *

Definicja

Isolated focal cortical dysplasia is a rare, genetic, non-syndromic cerebral malformation due to abnormal neuronal migration disorder characterized by variable-sized, focalized malformations located in any part(s) of the cerebral cortex, which manifests with drug-resistant epilepsy (usually leading to intellectual disability) and behavioral disturbances. Abnormal MRI findings (e.g. abnormal white and/or grey matter signal, blurred gray-white matter junction, localized volume loss, cortical thickening, abnormal gyral pattern, abnormal hippocampus) and variable histopathologic patterns are associated.

Dane

Klasyfikacja Synonimy

Choroba Epilepsy due to FCD

> Dysplazja korowa, typu Taylora Padaczka spowodowana FCD

Kod ORPHA Kod OMIM

Kod ICD10 65683 607341 Q04.8

Kod ICD11 LA05.51

*Źródło

orphanet