

Opis choroby *

Definicja

Isolated lissencephaly type 1 without known genetic defects belongs to the genetically heterogeneous group, classic lissencephaly (see this term). It is a diagnosis of exclusion, when neither associated malformations nor family history are present, and in the absence of mutations of genes known to be involved in classic lissencephaly. Clinically patients present with the common features of classic lissencephaly such as developmental delay, intellectual disability, and seizures.

Dane

Klasyfikacja

Choroba

Kod ORPHA

1084

Kod OMIM

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Kod ICD10

Q04.3

Kod ICD11

LD20.1

*Źródło

orphanet