

Opis choroby *

Definicja

A form of lissencephaly with cerebellar hypoplasia characterized by pronounced microcephaly (≤ -3 SD), intellectual disability, spastic diplegia and moderate to severe cerebellar hypoplasia involving both vermis and hemispheres.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

100014

Kod OMIM

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Kod ICD10

Q04.3

Kod ICD11

LD20.1

*Źródło

orphanet