Opis choroby *

Definicja

A rare, genetic, cobblestone lissencephaly disease characterized by the presence of a constellation of brain malformations, including cortical gyral and sulcus anomalies, white matter signal abnormalities, cerebellar dysplasia and brainstem hypoplasia, existing alone or in conjunction with minimal muscular and ocular abnormalities, typically manifesting with severe developmental delay, increased head circumference, hydrocephalus and seizures.

Dane

Klasyfikacja
Choroba

Synonimy
Cobblestone lissencephaly without muscular or eye involvement
Lizencefalia typu 2 bez zaangażowania mięśni lub oczu
Lizencefalia typu kostki brukowej bez zaangażowanie mięśni lub oczu
Lissencephaly type 2 without muscular or eye involvement
Lissencephaly type 2 without muscular or ocular involvement

Kod ORPHA
352682

Kod OMIM
615191

Kod ICD10
Q04.3

Kod ICD11
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*Źródło

orphanet