Opis choroby *

Definicja

A rare central nervous system malformation which includes a group of diseases that are characterized by a bumpy (or pebbled) appearance of the cerebral cortex, associated with a thickened cortex, reduction in normal sulcation, ventriculomegaly and reduced, abnormal white matter, as well as brainstem and cerebellum hypoplasia and corpus callosum agenesis. Patients generally present variable degrees of developmental delay, hypotonia and ocular abnomalities, however muscular and ocular involvement may be absent.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
Lissencephaly type 2
Lizencefalia typu 2

Kod ORPHA
51577

Kod OMIM
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Kod ICD10
Q04.3

Kod ICD11
LD20.1

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*Źródło

orphanet