Opis choroby *

Definicja

A rare, genetic, non-syndromic cerebral malformation due to abnormal neuronal migration disease characterized by the association of cortical dysplasia and pontocerebellar hypoplasia, manifesting with global developmental delay, mild to severe intellectual disability, axial hypotonia, strabismus, nystagmus and, occasionally, optic nerve hypoplasia. Brain imaging reveals variable malformations, including frontally predominant microgyria, gyral disorganization and simplification, dysmorphic and hypertrophic basal ganglia, cerebellar vermis dysplasia, brainstem/corpus callosum hypoplasia, and/or olfactory bulbs agenesis.

Dane

Klasyfikacja
Choroba

Kod ORPHA
300570

Kod OMIM
614039

Kod ICD10
Q04.3

Kod ICD11
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*Źródło

orphanet