Opis choroby *

Definicja

A rare, genetic, complex cerebral cortical malformation characterized by generalized or focal dysgyria (also named polymicrogyria-like cortical dysplasia) or alternatively by microlissencephaly with dysmorphic basal ganglia and dysgenesis of the corpus callosum. Clinical manifestations are variable and include microcephaly, seizures, hypotonia, developmental delay, severe psychomotor delay, ataxia, spastic diplegia or tetraplegia, and ocular abnormalities (strabismus, ptosis or optic atrophy).

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
300573

Kod OMIM
610031

Kod ICD10
Q04.3

Kod ICD11
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*Źródło

orphanet