Opis choroby *

Definicja

Hemimegalencephaly is a rare cerebral malformation characterized by overgrowth of all or part of a cerebral hemisphere, often with ipsilateral severe cortical dysplasia or dysgenesis, white matter hypertrophy and dilated lateral ventricle, presenting in early infancy with progressive hemiparesis, severe psychomotor retardation and intractable seizures. Hemimegalencephaly may be an isolated finding or associated with other syndromes such as angioosteohypertrophic syndrome, epidermal nevus syndrome and Ito hypomelanosis (see these terms). Management includes seizure control by antiepileptic medications and early hemispherectomy.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Unilateral megalencephaly
Megaencefalia jednostronna

Kod ORPHA
99802

Kod OMIM
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Kod ICD10
Q04.5

Kod ICD11
LA05.1

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*Źródło

orphanet