Opis choroby *

Definicja

A rare cerebral malformation characterized by an almost or complete lack of cortex, specifically the cerebral hemispheres, with the cranium and meninges completely intact. In most cases, death occurs in utero or in the first weeks of life. Developmental delay, drug-resistant seizures, spastic diplegia, severe growth failure, deafness and blindness are typical.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2177

Kod OMIM
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Kod ICD10
Q04.3

Kod ICD11
LA05.62

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*Źródło

orphanet