Opis choroby *

Definicja

A rare genetic non-syndromic central nervous system malformation characterized by absence of the telencephalon and absent or abnormal diencephalic structures, combined with severe abnormalities of the mesencephalon and cerebellum. Further malformations, for example of the hands and feet, have been described in addition.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1126

Kod OMIM
601374

Kod ICD10
Q04.3

Kod ICD11
LA05.Y

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*Źródło

orphanet