

Opis choroby *

Definicja

A benign form of holoprosencephaly characterized by midline defects without the typical HPE defect in brain cleavage and which can variably manifest with microcephaly, hypotelorism, midline cleft lip and/or flat nose, choanal stenosis, pyriform sinus stenosis, coloboma as well as a single median maxillary incisor.

Dane

| Klasyfikacja | Synonimy |
|-----------------------|-------------------------------|
| Zespół wad wrodzonych | HPE, minor form |
| | HPE-L |
| | HPE typu mikroform |
| | HPE-L |
| | Holoprosencephaly, minor form |
| | Holoprosencephaly-like |
| | Microform HPE |

Kod ORPHA

280200

Kod OMIM

610829

Kod ICD10

Q04.2

Kod ICD11

LA05.2

*Źródło

orphanet