Opis choroby *

Definicja

A rare subtype of holoprosencephaly characterized by midline fusion limited to the septal and/or preoptic regions of the telencephalon without a significant frontal neocortical fusion. Midline craniofacial malformations are generally mild and include solitary median maxillary incisor and pyriform sinus stenosis. Other reported manifestations include language delay, learning difficulties, and behavioral disorders. Imaging reveals abnormal fornix, absent or hypoplasic anterior corpus callosum, and unpaired anterior cerebral artery.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Septopreoptic HPE
HPE przegrodowo-przedwzrokowa HPE

Kod ORPHA
280195

Kod OMIM
609637

Kod ICD10
Q04.2

Kod ICD11
LA05.2

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*Źródło

orphanet