

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 hypoplastic anterior corpus callosum, and unpaired anterior cerebral artery.

Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	Septopreoptic HPE HPE przegrodowo-przedwzrokowa HPE

Kod ORPHA	Kod OMIM	Kod ICD10
280195	609637	Q04.2

Kod ICD11
LA05.2

*Źródło

orphanet