Opis choroby *

Definicja

Porencephaly-cerebellar hypoplasia-internal malformations syndrome is rare central nervous system malformation syndrome characterized by bilateral porencephaly, absence of the septum pellucidum and cerebellar hypoplasia with absent vermis. Additionally, dysmorphic facial features (hypertelorism, epicanthic folds, high arched palate, prominent metopic suture), macrocephaly, corneal clouding, situs inversus, tetralogy of Fallot, atrial septal defects and/or seizures have been observed.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Bonnemann-Meinecke syndrome
Zespół Bonnemanna i Meinecke'a

Kod ORPHA
2941

Kod OMIM
601322

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet