# Porencephaly-cerebellar hypoplasia-internal malformations syndrome

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Porencephaly-cerebellar hypoplasia-internal malformations syndrome. ORPHA:2941*

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.