

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Bonnemann-Meinecke syndrome Zespół Bonnemann i Meinecke'a

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
2941	601322	Q87.8

**Kod ICD11**

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### \*Źródło

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