

## Opis choroby \*

### Definicja

Periventricular nodular heterotopia (PNH) is a brain malformation, due to abnormal neuronal migration, in which a subset of neurons fails to migrate into the developing cerebral cortex and remains as nodules that line the ventricular surface. Classical PNH is a rare X-linked dominant disorder far more frequent in females who present normal intelligence to borderline intellectual deficit, epilepsy of variable severity and extra-central nervous system signs, especially cardiovascular defects or coagulopathy. The disorder is generally associated with prenatal lethality in males.

### Dane

|                     |                 |
|---------------------|-----------------|
| <b>Klasyfikacja</b> | <b>Synonimy</b> |
| Podtyp kliniczny    | PVNH<br>PVNH    |

|                  |                 |                  |
|------------------|-----------------|------------------|
| <b>Kod ORPHA</b> | <b>Kod OMIM</b> | <b>Kod ICD10</b> |
| 98892            | 615544          | Q04.8            |

**Kod ICD11**  
LA05.5Y

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### \*Źródło

orphanet