

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

Klasyfikacja

Podtyp kliniczny

Synonimy

PVNH

PVNH

Kod ORPHA

98892

Kod OMIM

615544

Kod ICD10

Q04.8

Kod ICD11

LA05.5Y

*Źródło

orphanet