DISEASE:
Periventricular nodular heterotopia

NAME:	Periventricular nodular heterotopia
DESCRIPTION:	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.
ORPHACODE:	98892
SYNONYMS:	PVNH

XREF(S):	Orphanet OMIM OMIM OMIM MeSH OMIM OMIM OMIM OMIM OMIM MedDRA ICD-10
ANALYTE(S):	ARFGEF2 FLNA ERMARD NEDD4L TMTC3 ARF1 MAP1B
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