

DISEASE:
Progressive myoclonic epilepsy type 5

NAME:	Progressive myoclonic epilepsy type 5
DESCRIPTION:	A rare, genetic neurological disorder characterized by early-onset progressive ataxia associated with myoclonic seizures, generalized tonic-clonic seizures (which are often sleep-related), and normal to mild intellectual disability. Dysarthria, upward gaze palsy, sensory neuropathy, developmental delay and autistic disorder have also been associated.
ORPHACODE:	402082
SYNONYMS:	EPM5 PME type 5 Progressive myoclonus epilepsy type 5
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PRICKLE2 POLG
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