DISEASE:
Progressive myoclonic epilepsy type 7

NAME:	Progressive myoclonic epilepsy type 7
DESCRIPTION:	A rare, genetic, neurological disorder characterized by childhood to adolescent onset of progressive myoclonus (which becomes very severe and results in major motor impediment) associated with infrequent tonic-clonic seizures, and, occasionally, ataxia. Learning disability prior to seizure onset and mild cognitive decline may be associated.
ORPHACODE:	435438
SYNONYMS:	EPM7 MEAK Myoclonus epilepsy and ataxia due to potassium channel mutation PME type 7 Progressive myoclonic epilepsy due to KV3.1 deficiency Progressive myoclonus epilepsy type 7
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	KCNC1
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