DISEASE:
Progressive myoclonic epilepsy type 6

NAME:	Progressive myoclonic epilepsy type 6
DESCRIPTION:	A rare, genetic, neurological disorder characterized by early-onset, progressive ataxia associated with myoclonic seizures (frequently associated with other seizure types such as generalized tonic-clonic, absence and drop attacks), scoliosis of variable severity, areflexia, elevated creatine kinase serum levels, and relative preservation of cognitive function until late in the disease course.
ORPHACODE:	280620
SYNONYMS:	EPM6 GOSR2-related progressive myoclonus ataxia North Sea progressive myoclonus epilepsy PME type 6 Progressive myoclonus epilepsy type 6
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	GOSR2
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