DISEASE:
Progressive myoclonic epilepsy type 1

NAME:	Progressive myoclonic epilepsy type 1
DESCRIPTION:	A rare progressive myoclonic epilepsy (PME) disorder characterized by action- and stimulus-sensitive myoclonus, and tonic-clonic seizures with ataxia, but with only a mild cognitive decline over time.
ORPHACODE:	308
SYNONYMS:	EPM1 Progressive myoclonus epilepsy type 1 ULD Unverricht-Lundborg disease
XREF(S):	Orphanet MeSH MedDRA ICD-10 OMIM OMIM OMIM
ANALYTE(S):	CSTB SCARB2 PRICKLE1

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RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)
- Myoclonic epilepsy of Unverricht and Lundborg / Progressive myoclonus epilepsy /Baltic myoclonic epilepsy

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- cystatin B
- prickle planar cell polarity protein 1
- scavenger receptor class B member 2

Related Gene Panels

- Rare epilepsy with developmental delay (> 240 genes) - UZA