DISEASE:
Progressive myoclonic epilepsy type 3

NAME:	Progressive myoclonic epilepsy type 3
DESCRIPTION:	A rare, genetic, neuronal ceroid lipofuscinosis disorder characterized by infantile- to early childhood-onset of progressive myoclonic seizures (occasionally accompanied by generalized tonic-clonic seizures) and severe, progressive neurological regression, leading to psychomotor and cognitive decline, cerebellar ataxia, dementia and, frequently, early death. Vision loss may be associated. EEG typically reveals epileptiform activity with predominance in the posterior region and photosensitivity.
ORPHACODE:	263516
SYNONYMS:	CLN14 disease EPM3 PME type 3 Progressive myoclonic epilepsy due to KCTD7 deficiency Progressive myoclonus epilepsy type 3
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	KCTD7
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/1094

RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- potassium channel tetramerization domain containing 7

Related Gene Panels

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