DISEASE:
Proximal myotonic myopathy

NAME:	Proximal myotonic myopathy
DESCRIPTION:	A rare myotonic dystrophy of juvenile or adult-onset characterized by mild and fluctuating myotonia, muscle weakness, and rarely cardiac conduction disorders.
ORPHACODE:	606
SYNONYMS:	Myotonic dystrophy type 2 Proximal myotonic dystrophy Ricker disease Ricker syndrome
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	CNBP
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Myotonic dystrophy type 2 - CCTG repeat expansion

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- CCHC-type zinc finger nucleic acid binding protein