DISEASE:
Paramyotonia congenita of Von Eulenburg

NAME:	Paramyotonia congenita of Von Eulenburg
DESCRIPTION:	Paramyotonia congenita of Von Eulenburg is characterised by exercise- or cold-induced myotonia and muscle weakness.
ORPHACODE:	684
SYNONYMS:	Paramyotonia congenita
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	SCN4A
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- sodium voltage-gated channel alpha subunit 4

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB