DISEASE:
Autosomal recessive axonal neuropathy with neuromyotonia

NAME:	Autosomal recessive axonal neuropathy with neuromyotonia
DESCRIPTION:	A rare peripheral neuropathy characterized by slowly progressive axonal, motor greater than sensory, polyneuropathy combined with neuromytonia (including spontaneous muscular activity at rest (myokymia), impaired muscle relaxation (pseudomyotonia), and contractures of hands and feet) and neuromyotonic or myokymic discharges on needle EMG. It presents with distal lower limb weakness with gait impairment, muscle stiffness, fasciculations and cramps in hands and legs worsened by cold, decreased to absent tendon reflexes, intrinsic hand muscle atrophy and, variably, mild distal sensory impairment.
ORPHACODE:	324442
SYNONYMS:	ARAN-NM ARCMT2-NM Autosomal recessive Charcot-Marie-Tooth disease type 2 with neuromyotonia
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	HINT1
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Neuropathy (gene panel)
- Peripheral neuropathy (gene panel)

Related Laboratories

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

Related Analytes

- histidine triad nucleotide binding protein 1

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Neuropathy (148 genes) - IPG
- Neuropathy (genepanel) - UZA