

DISEASE:**Autosomal dominant Charcot-Marie-Tooth disease type 2O**

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2O
DESCRIPTION:	A rare, genetic, subtype of autosomal dominant Charcot-Marie-Tooth disease type 2 characterized by early childhood-onset of slowly progressive, predominantly distal, lower limb muscle weakness and atrophy, delayed motor development, variable sensory loss, and pes cavus in the presence of normal or near-normal nerve conduction velocities. Additional variable features may include proximal muscle weakness, abnormal gait, arthrogryposis, scoliosis, cognitive impairment, and spasticity.
ORPHACODE:	284232
SYNONYMS:	CMT2O
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>DYNC1H1</u>
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RELATED CONTENT

Related Genetic Tests

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

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- dynein cytoplasmic 1 heavy chain 1

Related Gene Panels

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

Source URL: <http://gentest.healthdata.be/disease/2014>