DISEASE:
Autosomal dominant Charcot-Marie-Tooth disease type 2DD

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2DD
DESCRIPTION:	A rare autosomal dominant hereditary axonal motor and sensory neuropathy characterized by predominantly distal weakness and muscle atrophy, decreased or absent tendon reflexes, and reduced vibratory sensation in the lower and upper extremities. Pes cavus develops in many patients. Additional symptoms like ataxia, tremor, or swallowing difficulties have been reported. Patients usually remain ambulatory even late in the disease. Age of onset ranges from childhood to adulthood, with earlier onset tending to be associated with a more severe disease phenotype.
ORPHACODE:	521414
SYNONYMS:	ATP1A1-related CMT2 ATP1A1-related autosomal dominant Charcot-Marie-Tooth disease type 2 CMT2DD
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ATP1A1
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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

- ATPase Na+/K+ transporting subunit alpha 1

Related Gene Panels

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