DISEASE:
X-linked Charcot-Marie-Tooth disease type 6

NAME:	X-linked Charcot-Marie-Tooth disease type 6
DESCRIPTION:	X-linked Charcot-Marie-Tooth disease type 6 is a rare, genetic, principally axonal, peripheral sensorimotor neuropathy characterized by an X-linked dominant inheritance pattern and the childhood-onset of slowly progressive, moderate to severe, distal muscle weakness and atrophy of the lower extremities, as well as distal, panmodal sensory abnormalities, bilateral foot deformities (pes cavus, clawed toes), absent ankle reflexes and gait abnormalities (steppage gait). Females are usually asymptomatic or only present mild manifestations (mild postural hand tremor, mild wasting of hand intrinsic muscles).
ORPHACODE:	352675
SYNONYMS:	CMT6X CMTX6
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PDK3
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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

- pyruvate dehydrogenase kinase 3

Related Gene Panels

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