DISEASE:
Charcot-Marie-Tooth disease type 4H

NAME:	Charcot-Marie-Tooth disease type 4H
DESCRIPTION:	Charcot-Marie-Tooth disease type 4H is a subtype of Charcot-Marie-Tooth disease type 4 characterized by onset before two years of age of severe, slowly progressive, demyelinating sensorimotor neuropathy manifesting with delayed motor development (walking), unsteady gait, distal muscle weakness and atrophy (more prominent in the lower limbs), areflexia, mild symmetrical stocking-distribution hypoesthesia, and skeletal malformations (incl. kyphoscoliosis, short neck, pes cavus and pes equinus). Severely reduced nerve conduction velocities are associated.
ORPHACODE:	99954
SYNONYMS:	CMT4H
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	FGD4
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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

- FYVE, RhoGEF and PH domain containing 4

Related Gene Panels

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