DISEASE:
Autosomal recessive intermediate Charcot-Marie-Tooth disease type A

NAME:	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A
DESCRIPTION:	A subtype of autosomal recessive intermediate Charcot-Marie-Tooth (CMT) disease characterized by severe, early childhood-onset CMT neuropathy with prominent pes equinovarus deformity and impairment of hand muscles. Nerve conduction velocities usually range between 25-35 m/s and both axonal and demyelinating changes are observed on peripheral nerve pathology.
ORPHACODE:	217055
SYNONYMS:	RI-CMT type A
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	GDAP1
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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

- ganglioside induced differentiation associated protein 1

Related Gene Panels

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