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

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2B
DESCRIPTION:	A severe form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, with onset in the 2nd or 3rd decade, characterized by ulcerations and infections of feet. Symmetric and distal weakness develops mostly in the legs together with a severe symmetric distal sensory loss, tendon reflexes are only reduced at ankles and foot deformities, including pes cavus or planus and hammer toes, appear in childhood.
ORPHACODE:	99936
SYNONYMS:	CMT2B
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	RAB7A
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

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Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

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