DISEASE:
Dejerine-Sottas syndrome

NAME:	Dejerine-Sottas syndrome
DESCRIPTION:	A clinical entity that represents a severe phenotype of Charcot-Marie-Tooth disease characterized by onset occurring in infancy, severe motor weakness, delayed motor development, extremely slow nerve conduction (< 10-12 m/s), areflexia and foot deformity. Mutations in the genes PMP22 (17p12), MPZ (1q22), EGR2 (10q21.1) and PRX (19q13.2) have been implicated.
ORPHACODE:	64748
SYNONYMS:	Charcot-Marie-Tooth disease type 3 HMSN 3 HMSN III Hereditary motor and sensory neuropathy type 3 Hereditary motor and sensory neuropathy type III
XREF(S):	Orphanet OMIM OMIM MeSH ICD-10

ANALYTE(S):	PMP22 PRX EGR2 MPZ
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Charcot-Marie-Tooth type 1A (CMT1A) / Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)
- 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

- early growth response 2
- myelin protein zero
- peripheral myelin protein 22
- periaxin

Related Gene Panels

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

- Neuropathy (genepanel) - UZA
