DISEASE:
Hereditary sensorimotor neuropathy with hyperelastic skin

NAME:	Hereditary sensorimotor neuropathy with hyperelastic skin
DESCRIPTION:	A rare, genetic, demyelinating hereditary motor and sensory neuropathy disorder characterized by slowly progressive, mild to moderate, distal muscle weakness and atrophy of the upper and lower limbs and variable distal sensory impairment, associated with variable hyperextensible skin and age-related macular degeneration. Hypermobility of distal joints, high palate, and minor skeletal abnormalities (e.g. pectus excavatum, dolichocephaly) may also be associated.
ORPHACODE:	280598
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	FBLN5
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)
- Neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- fibulin 5

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Neuromuscular disorders (166 genes) - VUB
- Neuropathy (148 genes) - IPG