DISEASE:
Palmoplantar keratoderma-hereditary motor and sensory neuropathy syndrome

NAME:	Palmoplantar keratoderma-hereditary motor and sensory neuropathy syndrome
DESCRIPTION:	A rare, genetic, autosomal dominant hereditary axonal motor and sensory neuropathy disorder characterized by childhood-onset palmoplantar keratoderma associated with motor and sensory polyneuropathy manifestating with late-onset, predominantly distal, lower limb muscle weakness and atrophy (later associating mild proximal weakness and upper limb involvement), moderate sensory impairment (hypoesthesia with stocking-glove distribution), and normal or near?normal nerve conduction velocities. Additional variable manifestations include impaired vibratory sensation, reduced tendon reflexes, paresthesia, pain, talipes equinovarus, pes cavus, and nail dystrophy.
ORPHACODE:	538574
SYNONYMS:	Palmoplantar keratoderma-Charcot-Marie-Tooth syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	KRT1 MPZ
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Ichthyosis (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- keratin 1
- myelin protein zero

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Ichthyosis and erythroderma (98 genes) - KUL