Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Choroba

Synonimy
Palmoplantar keratoderma-Charcot-Marie-Tooth syndrome
Palmoplantar keratoderma-Charcot-Marie-Tooth syndrome

Kod ORPHA
538574

Kod OMIM
148360

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet