

## 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 manifesting 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  
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#### Kod ORPHA

538574

#### Kod OMIM

148360

#### Kod ICD10

G60.0

#### Kod ICD11

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