Opis choroby *

Definicja

A rare hereditary disease with peripheral neuropathy characterized by distal sensorimotor or motor neuropathy of the lower limbs with muscle weakness and atrophy. Some patients show overt connective tissue disease with signs and symptoms like increased skin elasticity and easy bruising (but no atrophic scarring), decreased clotting, aortic aneurysms, joint hypermobility, and recurrent tendon ruptures.

Dane

Klasyfikacja
Choroba

Kod ORPHA
485418

Kod OMIM
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Kod ICD10
G60.8

Kod ICD11
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*Źródło

orphanet