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

485418

Kod ORPHA

Kod OMIM

Kod ICD10

G60.8

Kod ICD11

*Źródło

orphanet