Opis choroby *

Definicja

A subtype of Autosomal dominant Charcot-Marie-Tooth disease type 2 characterized by the childhood onset of distal weakness and areflexia (with earlier and more severe involvement of the lower extremities), reduced sensory modalities (primarily pain and temperature sensation), foot deformities, postural tremor, scoliosis and contractures. Optic atrophy, vocal cord palsy with dysphonia, sensorineural hearing loss, spinal cord abnormalities and hydrocephalus have also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
CMT2A2
CMT2A2

Kod ORPHA
99947

Kod OMIM
609260

Kod ICD10
G60.0

Kod ICD11
8C20.1

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*Źródło

orphanet