

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

*Źródło

orphanet