Opis choroby *

Definicja

A rare demyelinating hereditary motor and sensory neuropathy characterized by prominent gait ataxia, pes cavus, tendon areflexia, distal limb weakness, tremor in the upper limbs, distal sensory loss, kyphoscoliosis, and progressive muscle atrophy. The disease becomes symptomatic in infancy or childhood, mode of inheritance is autosomal dominant.

Dane

Klasyfikacja
Choroba

Synonimy
Hereditary areflexic dystasia, Roussy-Lévy type
Dziedziczna dystazja z brakiem odruchów, typ Roussy'ego i Lévy'ego

Kod ORPHA
3115

Kod OMIM
180800

Kod ICD10
G60.0

Kod ICD11
8C20.Y

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*Źródło

orphanet