Opis choroby *

Definicja

A rare genetic skeletal muscle disease characterized by neonatal to childhood onset of slowly progressive muscle weakness and atrophy primarily affecting the lower limbs, joint contractures, kyphosis or lordosis of the spine, lateral tongue atrophy, and pes equinus. Progression to upper limb involvement, facial weakness, language impairment, intellectual disability, and behavioral abnormalities have been reported in addition. Muscle biopsy shows myopathic changes with increased fiber size variation, internalized nuclei, fiber atrophy, as well as rod structures and core targetoid defects.

Dane

Klasyfikacja
Choroba

Kod ORPHA
496686

Kod OMIM
617114

Kod ICD10
G71.2

Kod ICD11
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*Źródło

orphanet