

Opis choroby *

Definicja

A rare glycogen storage disease characterized by slowly progressive myopathy with storage of polyglucosan in muscle fibers. Age of onset ranges from childhood to late adulthood. Patients present proximal or proximodistal weakness predominantly of limb-girdle muscles. Variable features include exercise intolerance or myalgia. Serum creatine kinase is normal or mildly elevated. There is usually no overt cardiac involvement.

Dane

Klasyfikacja

Choroba

Kod ORPHA

456369

Kod OMIM

616199

Kod ICD10

E74.0

Kod ICD11

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*Źródło

orphanet