

Opis choroby *

Definicja

Distal anoctaminopathy is a rare, autosomal recessive distal myopathy characterized by early adult-onset, slowly progressive, often asymmetrical, lower limb muscle weakness initially affecting the calves (with relative anterior muscle sparing) and later proximal muscle involvement, as well as highly elevated creatine kinase (CK) serum levels.

Dane

Klasyfikacja

Choroba

Synonimy

MMD3

Dystrofia mięśniowa Miyoshi typu 3

MMD3

Miyoshi muscular dystrophy type 3

Kod ORPHA

399096

Kod OMIM

613319

Kod ICD10

G71.0

Kod ICD11

-

*Źródło

orphanet