

Opis choroby *

Definicja

X-linked distal spinal muscular atrophy type 3 is a rare distal hereditary motor neuropathy characterized by slowly progressive atrophy and weakness of distal muscles of hands and feet with normal deep tendon reflexes or absent ankle reflexes and minimal or no sensory loss, sometimes mild proximal weakness in the legs and feet and hand deformities in males.

Dane

Klasyfikacja

Choroba

Synonimy

ATP7A-related distal motor neuropathy

DSMAX

SMAX3

dHMN typu 3 sprzężony z chromosomem X

dSMA typu 3 sprzężony z chromosomem X

Dystalna neuropatia ruchowa zależna od ATP7A

Dystalna dziedziczna neuropatia ruchowa typu 3 sprzężona z chromosomem X

DSMAX

SMAX3

X-linked dHMN3

X-linked dSMA3

X-linked distal hereditary motor neuropathy type 3

Kod ORPHA

139557

Kod OMIM

300489

Kod ICD10

G12.2

Kod ICD11

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*Źródło

orphanet