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