Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 28 is a pure form of hereditary spastic paraplegia characterized by a childhood or adolescent onset of slowly progressive, pure crural muscle spastic paraparesis which manifests with mild lower limb weakness, gait difficulties, extensor plantar responses, and hyperreflexia of lower extremities. Less common manifestations include cerebellar oculomotor disturbance with saccadic eye pursuit, pes cavus and scoliosis. Some patients also present pin and vibration sensory loss in distal legs.

Dane

Klasyfikacja
Choroba

Synonimy
SPG28
SPG28

Kod ORPHA
101008

Kod OMIM
609340

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet