Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 15 is a complex form of hereditary spastic paraplegia characterized by a childhood to adulthood onset of slowly progressive lower limb spasticity (resulting in gait disturbance, extensor plantar responses and decreased vibration sense) associated with mild intellectual disability, mild cerebellar ataxia, peripheral neuropathy (with distal upper limb amyotrophy) and retinal degeneration. Thin corpus callosum is a common imaging finding.

Dane

Klasyfikacja
Choroba

Synonimy
Hereditary spastic paraparesis type 15
Dziedziczna parapareza spastyczna typu 15
Paraplegia spastyczna - zwyrodnienie rogówki
SPG15
Zespół Kjellina
Kjellin syndrome
SPG15
Spastic paraplegia-retinal degeneration syndrome

Kod ORPHA
100996

Kod OMIM
270700

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet