

Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 14 is a rare, complex hereditary spastic paraplegia characterized by adulthood-onset of slowly progressive spastic paraplegia of lower limbs presenting with spastic gait, hyperreflexia, and mild lower limb hypertonicity associated with mild intellectual disability, visual agnosia, short and long-term memory deficiency and mild distal motor neuropathy. Bilateral pes cavus and extensor plantar responses are also associated.

Dane

Klasyfikacja

Choroba

Synonimy

SPG14

SPG14

Kod ORPHA

100995

Kod OMIM

605229

Kod ICD10

G11.4

Kod ICD11

8B44.01

*Źródło

orphanet