Autosomalna recesywna paraplegia spastyczna typu 48

Kod Orpha: 306511 Kod OMIM: 613647

Opis choroby *

Definicja

A rare, pure or complex form of hereditary spastic paraplegia usually characterized by a pure phenotype of a slowly progressive spastic paraplegia associated with urinary incontinence with an onset in mid- to late-adulthood. A complex phenotype, with the additional findings of cognitive impairment, sensorimotor polyneuropathy, ataxia, parkinsonism, and dystonia as well as thin corpus callosum and white matter lesions (seen on brain and spine magnetic resonance imaging), has also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
SPG48
SPG48

Kod ORPHA
306511

Kod OMIM
613647

Kod ICD10
G11.4

Kod ICD11
-

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.