Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 18 (SPG18) is a rare, complex type of hereditary spastic paraplegia characterized by progressive spastic paraplegia (presenting in early childhood) associated with delayed motor development, severe intellectual disability and joint contractures. A thin corpus callosum is equally noted on brain magnetic resonance imaging. SPG18 is caused by a mutation in the ERLIN2 gene (8p11.2) encoding the protein, Erlin-2.

Dane

Klasyfikacja
Choroba

Synonimy
SPG18
SPG18

Kod ORPHA
209951

Kod OMIM
611225

Kod ICD10
G11.4

Kod ICD11
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*Źródło

orphanet