## 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 <i>ERLIN2</i> gene (8p11.2) encoding the protein, Erlin-2.

Dane

Klasyfikacja Synonimy Choroba SPG18

SPG18

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 209951
 611225
 G11.4

**Kod ICD11** 

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## \*Źródło

orphanet