

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Choroba	SPG18 SPG18

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
209951	611225	G11.4

**Kod ICD11**  
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### \*Źródło

orphanet