Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 63 (SPG63) is an extremely rare and complex form of hereditary spastic paraplegia characterized by an onset in infancy of spastic paraplegia (presenting with delayed walking and a scissors gait) associated with short stature, and normal cognition. Periventricular deep white matter changes in the corpus callosum are noted on brain imaging. SPG63 is caused by a homozygous mutation in the AMPD2 gene (1p13.3) encoding AMP deaminase 2.

Dane

Klasyfikacja
Choroba

Synonimy
SPG63
SPG63

Kod ORPHA
401805

Kod OMIM
615686

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet