Opis choroby *

Definicja

A rare complex hereditary spastic paraplegia characterized by juvenile to adult onset of slowly progressive spasticity mainly affecting the lower limbs, associated with spastic dysarthria and motor neuropathy. Additional manifestations include congenital bilateral cataract, gastroesophageal reflux, persistent vomiting, mild cerebellar signs, pes cavus, and occasionally short stature, among others.

Dane

Klasyfikacja
Choroba

Synonimy
AD-SPG9A
Cataracts-motor neuropathy-short stature-skeletal anomalies syndrome
Spastic paraparesis-amyopathy-cataracts-gastroesophageal reflux syndrome
AD-SPG9A
Cataracts-motor neuropathy-short stature-skeletal anomalies syndrome
Spastic paraparesis-amyopathy-cataracts-gastroesophageal reflux syndrome

Kod ORPHA
447753

Kod OMIM
601162

Kod ICD10
G11.4

Kod ICD11
8B44.00

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*Źródło

orphanet