

Opis choroby *

Definicja

A rare complex hereditary spastic paraplegia characterized by early onset of slowly progressive spastic para- or tetraparesis, increased tendon reflexes, positive Babinski sign, global developmental delay, cognitive impairment, and pseudobulbar palsy. Additional manifestations include dysmorphic facial features, tremor, short stature, and urinary incontinence.

Dane

Klasyfikacja

Choroba

Synonimy

AR-SPG9B

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Kod ORPHA

447760

Kod OMIM

616586

Kod ICD10

G11.4

Kod ICD11

8B44.00

*Źródło

orphanet