Opis choroby *

Definicja

A rare complex hereditary spastic paraplegia characterized by neonatal to infantile onset of progressive spasticity in the lower limbs, hyperreflexia, tip-toe walking, pes equinus, and delayed motor developmental milestones. Kyphoscoliosis becomes evident in older patients, and most patients show atrophy of the lateral aspects of the tongue. Additional signs may include intellectual disability, language impairment, and moderate upper limb involvement.

Dane

Klasyfikacja Synonimy

Choroba Kyphoscoliosis-lateral tongue atrophy-HSP

syndrome

Kyphoscoliosis-lateral tongue atrophy-HSP

syndrome

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 496689
 G11.4

Kod ICD11

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*Źródło

orphanet