

Opis choroby *

Definicja

X-linked intellectual disability, Hedera type is a rare X-linked intellectual disability syndrome characterized by an onset in infancy of delayed motor and speech milestones, generalized tonic-clonic seizures and drop attacks, and mild to moderate intellectual disability. Additional, less common manifestations include scoliosis, ataxia (resulting in progressive gait disturbance), and bilateral pes planovalgus. Physical appearance is normal with no dysmorphic features reported.

Dane

Klasyfikacja

Choroba

Synonimy

MRXSH

MRXSH

Kod ORPHA

93952

Kod OMIM

300423

Kod ICD10

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Kod ICD11

LD90

*Źródło

orphanet