

Opis choroby *

Definicja

Fatty acid hydroxylase-associated neurodegeneration (FAHN) is a very rare, autosomal recessive form of neurodegeneration with brain iron accumulation (NBIA) characterized by childhood-onset focal dystonia, progressive spastic paraplegia that progresses to tetra paresis, ataxia, dysarthria, intellectual decline, and oculomotor disturbances (optic atrophy), accompanied by iron deposition in the globus pallidus.

Dane

Klasyfikacja

Choroba

Synonimy

FAHN

FAHN

Kod ORPHA

329308

Kod OMIM

612319

Kod ICD10

G23.0

Kod ICD11

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*Źródło

orphanet