

Opis choroby *

Definicja

COASY protein-associated neurodegeneration (CoPAN) is a very rare, slowly progressive form of neurodegeneration with brain iron accumulation (NBIA) characterized by classic NBIA features. The clinical manifestations include early-onset spastic-dystonic paraparesis, oromandibular dystonia, dysarthria, parkinsonism, axonal neuropathy, progressive cognitive impairment, complex motor tics, and obsessive-compulsive disorder.

Dane

Klasyfikacja

Choroba

Synonimy

CoPAN

CoPAN

NBIA6

Neurodegeneracja z magazynowaniem żelaza w mózgu z powodu mutacji COASY

NBIA6

Neurodegeneration with brain iron accumulation due to COASY mutation

Kod ORPHA

397725

Kod OMIM

615643

Kod ICD10

G23.0

Kod ICD11

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*Źródło

orphanet