Opis choroby *

Definicja

A form of hereditary cerebral hemorrhage with amyloidosis characterized by age of onset between 50-66 years of age, memory impairment, myoclonic jerks, expressive dysphagia, short-stepped gait, personality changes, and lobar intracerebral hemorrhages. This subtype is due to a mutation in the APP gene (21q21.2), encoding the beta-amyloid precursor protein. This mutation causes an increased accumulation of amyloid-beta protein in the walls of the arteries and capillaries of the meninges, cerebellar cortex and cerebral cortex, leading to the weakening and eventual rupture of these vessels.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
ABetaD23N amyloidosis
Amyloidoza ABetaD23N
Dziedziczny krwotok mózgowy z amyloidozą, typ Iowa
HCHWA, typ Iowa
HCHWA, Iowa type
Hereditary cerebral hemorrhage with amyloidosis, Iowa type

Kod ORPHA
324708

Kod OMIM
605714

Kod ICD10
I68.0*

Kod ICD11
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*Źródło

orphanet