

Opis choroby *

Definicja

A form of hereditary cerebral hemorrhage with amyloidosis characterized by an age of onset of 54-61 years and progressive Alzheimer's disease-like dementia. 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

ABetaE22G amyloidosis

Amyloidoza ABetaE22G

Dziedziczny krwotok mózgowy z amyloidozą, typ arktyczny

HCHWA, typ arktyczny

HCHWA, Arctic type

Hereditary cerebral hemorrhage with amyloidosis, Arctic type

Kod ORPHA

324723

Kod OMIM

605714

Kod ICD10

I68.0*

Kod ICD11

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[*Źródło](#)

orphanet