Opis choroby *

Definicja

A form of hereditary cerebral hemorrhage with amyloidosis characterized by an age of onset of 45 years of age, progressive Alzheimer's disease-like dementia, and lobar intracerebral hemorrhage in some patients. 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
ABeta amyloidosis, Flemish type
Amyloidoza zależna od ABetaA21G
Beta amyloidoza, typ flamandzki
Dziedziczny krwotok mózgowy z amyloidozą, typ flamandzki
HCHWA, typ flamandzki
ABetaA21G-related amyloidosis
HCHWA, Flemish type
Hereditary cerebral hemorrhage with amyloidosis, Flemish type

Kod ORPHA
324718

Kod OMIM
605714

Kod ICD10
I68.0*

Kod ICD11
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*Źródło

orphanet