

Opis choroby *

Definicja

A form of hereditary cerebral hemorrhage with amyloidosis characterized by an age of onset between 50-70 years of age, recurrent lobar intracerebral hemorrhages and cognitive decline. 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, Piedmont type

Amyloidóza zależna od ABetaL34V

Beta amyloidóza, typ piemoncki

Dziedziczny krwotok mózgowy z amyloidozą, typ piemoncki

HCHWA, typ piemoncki

ABetaL34V-related amyloidosis

HCHWA, Piedmont type

Hereditary cerebral hemorrhage with amyloidosis, Piedmont type

Kod ORPHA

324703

Kod OMIM

605714

Kod ICD10

I68.0*

Kod ICD11

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*Źródło

orphanet