Opis choroby *

Definicja

A form of hereditary cerebral hemorrhage with amyloidosis characterized by an age of onset of 50 years of age, dementia and lobar intracerebral hemorrhage. 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
ABetaE22K amyloidosis
Amyloidoza ABetaE22K
Dziedziczny krwotok mózgowy z amyloidozą, typ włoski
HCHWA, typ włoski
HCHWA, Italian type
Hereditary cerebral hemorrhage with amyloidosis, Italian type

Kod ORPHA
324713

Kod OMIM
605714

Kod ICD10
E85.4+

Kod ICD11
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*Źródło

orphanet