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
Amyloidoza zależna od ABetaL34V
Beta amyloidoza, 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
-

---

*Źródło

orphanet