## 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