DISEASE:
ABeta amyloidosis, Arctic type

NAME:	ABeta amyloidosis, Arctic type
DESCRIPTION:	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.
ORPHACODE:	324723
SYNONYMS:	ABetaE22G amyloidosis HCHWA, Arctic type Hereditary cerebral hemorrhage with amyloidosis, Arctic type
XREF(S):	Orphanet OMIM ICD-10 ICD-10
ANALYTE(S):	APP
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RELATED CONTENT

Related Analytes

- amyloid beta precursor protein

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