# DISEASE:
# ABetaL34V amyloidosis

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| **NAME:** | ABetaL34V amyloidosis |
| **DESCRIPTION:** | 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. |
| **ORPHACODE:** | 324703 |
| **SYNONYMS:** | ABeta amyloidosis, Piedmont type<br>ABetaL34V-related amyloidosis<br>HCHWA, Piedmont type<br>Hereditary cerebral hemorrhage with amyloidosis, Piedmont type |
| **XREF(S):** | Orphanet<br>OMIM<br>ICD-10<br>ICD-10 |
| **ANALYTE(S):** | APP |
| **CREATED:** | 13 May 2019 - 01:02 |

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| **CHANGED:** | 22 Jun 2023 - 16:14 |

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### Related Analytes

- amyloid beta precursor protein

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