DISEASE:
ABeta amyloidosis, Iowa type

NAME:	ABeta amyloidosis, Iowa type
DESCRIPTION:	A form of hereditary cerebral hemorrhage with amyloidosis characterized by age of onset between 50-66 years of age, memory impairment, myoclonic jerks, expressive dysphagia, short-stepped gait, personality changes, and lobar intracerebral hemorrhages. 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:	324708
SYNONYMS:	ABetaD23N amyloidosis HCHWA, Iowa type Hereditary cerebral hemorrhage with amyloidosis, Iowa type
XREF(S):	Orphanet OMIM ICD-10 ICD-10
ANALYTE(S):	APP
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/1685

RELATED CONTENT

Related Analytes

- amyloid beta precursor protein