

DISEASE:
ACys amyloidosis

NAME:	ACys amyloidosis
DESCRIPTION:	A form of hereditary cerebral hemorrhage with amyloidosis characterized by an age of onset of 20-30 years, major systemic amyloidosis and recurrent lobar intracerebral hemorrhages. Unlike other forms of hereditary cerebral hemorrhage with amyloidosis, this subtype is due to a mutation in the CST3 gene (20p11.2), encoding the precursor protein cystatin C.
ORPHACODE:	100008
SYNONYMS:	CST3-related amyloidosis Cystatin amyloidosis HCHWA, Icelandic type Hereditary cerebral hemorrhage with amyloidosis, Icelandic type Hereditary cystatin C amyloid angiopathy
XREF(S):	Orphanet ICD-10 ICD-10 OMIM
ANALYTE(S):	CST3
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14
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