DISEASE:
ATTRV30M amyloidosis

NAME:	ATTRV30M amyloidosis
DESCRIPTION:	A rare hereditary ATTR amyloidosis (hATTR) characterized by a progressive, length-dependent sensorimotor axonal polyneuropathy and/or autonomic neuropathy in adulthood. Renal, ocular and cardiac involvement also frequently occurs. Two different phenotypes are associated with this mutation, namely early-onset V30M and late-onset V30M, that differ in terms of age on onset (<50 years or >50 years, respectively), presenting features, histopathological characteristics, rate of disease progression and response to therapy.
ORPHACODE:	85447
SYNONYMS:	ATTRV30M-related amyloidosis Familial amyloid polyneuropathy type I Familial amyloid polyneuropathy, Portuguese-Swedish-Japanese type TTR amyloid neuropathy Transthyretin amyloid neuropathy Transthyretin amyloid polyneuropathy
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	TTR

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RELATED CONTENT

Related Genetic Tests

- Amyloidosis (TTR full sanger exon sequencing)
- Amyloidosis (full sanger screening of the 4 exons for TTR)
- Amyloidosis hereditary / Dystransthyretinemic hyperthyroxinemia
- Cardiomyopathy, hereditary (gene panel)
- Cardiopathies, hereditary (gene panel)
- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Neuropathy (gene panel)
- Transthyretine amyloïdose

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- transthyretin

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Cardiomyopathy, hereditary (208 genes) - VUB
- Cardiopathies, hereditary (102 genes) - KUL
- Neuropathy (148 genes) - IPG
