DISEASE:
ATTRV122I amyloidosis

NAME:	ATTRV122I amyloidosis
DESCRIPTION:	A rare hereditary Transthyretin (TTR)-related systemic amyloidosis (ATTR) with predominant cardiac involvement resulting from myocardial infiltration of abnormal amyloid protein.
ORPHACODE:	85451
SYNONYMS:	ATTR cardiomyopathy ATTRV122I-related amyloidosis TTR-related amyloid cardiomyopathy TTR-related cardiac amyloidosis Transthyretin amyloid cardiopathy Transthyretin-related familial amyloid cardiomyopathy
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
