MAYO CLINIC LABORATORIES

1-800-533-1710

TTRX

Amyloidosis, Transthyretin-Associated Familial, Reflex, Blood

Patient ID	Patient Name		Birth Date	Sex	Age
SA00175543	TESTINGRNV, SAMPLEREPORT		1982-06-29	F	42
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00175543	SA00175543	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		06 May 2025 05:00			

Familial Amyloidosis Reflex

Wild Type Mass MCR

13764.0 Da

Wild Type Width at Half Height MCR

7.5 Da

Second Mass MCR

13793.5 Da

Mass Difference MCR

29.5 Da

Abnormal result MCR

Yes

Interpretation [1] MCR

The presence of a structural change in TTR is suggestive of a gene mutation that requires confirmation by DNA sequence analysis. DNA sequencing of the TTR gene is currently underway in the genomics laboratory; interpretive report to follow.

ADDITIONAL INFORMATION

Affinity Chromatography-Mass Spectrometry (MS)

Reviewed By MCR

Amy Breitlow

Received: 07 May 2025 05:52 **Reported:** 07 May 2025 06:34

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292

MAYO CLINIC

1-800-533-1710

TTR Gene, Full Gene Analysis

TTRZ

PATIENT NAME				ORDER NUMBER
TESTINGRNV, SAMPLEREPORT				**P407000079**
PATIENT ID SA00175543	DATE OF BIRTH 06/29/1982	AGE 42 Y	SEX Female	REQUESTED BY CLIENT CLIENT
COLLECTED 5/6/2025, 5:00 AM	RECEIVED 5/8/2025, 10:01 AM	REPORTED 5/8/2025, 10:18 AM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL RochesterCampus Rochester MN 55901				CLIENT ORDER NUMBER SA00175543 CLIENT MRN SA00175543

TEST DESCRIPTION

Evaluation of the TTR gene associated with amyloidosis

SPECIMEN

WB Whole Blood

RESULT SUMMARY

Pathogenic Variant Detected

RESULT

The following heterozygous PATHOGENIC variant was detected:
TTR (NM_000371.3), chr18(GRCh37):g.29172937G>A, c.148G>A, p.Val50Met (p.V50M)

INTERPRETATION

TTR c.148G>A (p.Val50Met), PATHOGENIC
The heterozygous c.148G>A (p.Val50Met) missense variant in the TTR gene (MIM:176300) is an established pathogenic variant. Pathogenic variants in the TTR gene have been associated with autosomal dominant hereditary amyloidosis (1). This result is supportive of a diagnosis of TTR-related amyloidosis for this individual. Clinical correlation is recommended.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

Consultation with a genetics professional is recommended for interpretation of this result and to determine whether reproductive risk assessment and familial testing may be of benefit to this family. Genetic testing for family members is available by ordering FMTT / Familial Mutation, Targeted Testing for the specific variant(s) detected. Please contact the laboratory at 1-800-533-1710 or the online test catalog at www.mayocliniclabs.com for information about FMTT.

REFERENCES:
1) GeneReviews: Hereditary Transthyretin Amyloidosis (https://www.ncbi.nlm.nih.gov/books/NBK1194/) (PMID: 20301373)

METHOD

Next generation sequencing (NGS) and/or Sanger sequencing was performed to test for the presence of variants in coding regions and intron/exon boundaries of the gene analyzed. The human genome reference GRCh37/hg19 build was

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used for sequence read alignment. At least 99% of the bases are covered at a read depth >30X. Sensitivity is estimated at >99% for single nucleotide variants, >94% for indels up to 39 base pairs, >95% for deletions up to 75 base pairs and insertions up to 47 base pairs. NGS and/or a PCR-based quantitative method was performed to test for the presence of deletions and duplications in the gene analyzed. See the Genes Analyzed field for a list of gene(s) tested.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria. See www.mayocliniclabs.com (TEST ID TTRZ) for details regarding genes with regions not routinely covered.

GENES ANALYZED

TTR

DISCLAIMER

Clinical Correlations
An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

If testing was performed because of a clinically significant family history it is often useful to first test an affected family member. Detection of a reportable variant(s) in an affected family member would allow for more informative testing of at risk individuals.

To discuss the availability of further testing options or for assistance in the interpretation of these results, Mayo Clinic Laboratory genetic counselors can be contacted at 1-800-533-1710.

Technical Limitations
Next generation sequencing may not detect all types of genomic variants. In rare cases, false negative or false positive results may occur. The depth of coverage may be variable for some target regions, but assay performance below the minimum acceptable criteria or for failed regions will be noted. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria.

Additionally, low level mosaic variants may not be detected.

This test is not designed to differentiate between somatic and germline variants. If there is a possibility that any detected

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variant is somatic, additional testing may be necessary to clarify the significance of results.

Reclassification of Variants Policy
See www.mayocliniclabs.com (TEST ID TTRZ) for information regarding the laboratory's policy for reclassification of variants.

RELEASED BY

CRYSTAL ELLIS

Code : MCR Laboratory : Mayo Clinic Laboratories - Rochester Main Campus Address : 200 FIRST STREET SW
Lab Director : NIKOLA A BAUMANN PHD CLIA Certificate : 24D0404292 ROCHESTER MN 55905