

## **Amyloidosis, Transthyretin-Associated Familial, Blood #83674**

**EXPLANATION:** Due to technical difficulties, test #83674, Amyloidosis, Transthyretin-Associated Familial, Blood, will become obsolete effective May 21, 2009. Efforts are underway to develop a biochemical assay as a replacement. Until this assay becomes available, consider the alternative test recommended below.

**RECOMMENDED ALTERNATIVE TEST:** Test #83667, Familial Amyloidosis, DNA Sequence.

**METHODOLOGY:** Polymerase Chain Reaction (PCR) Amplification/DNA Sequencing.

**SPECIMEN REQUIREMENTS:** Draw blood in a lavender-top (EDTA) tube or a yellow-top (ACD) tube, and send 2 mL of EDTA or ACD whole blood in original VACUTAINER. Invert several times to mix blood. Forward unprocessed whole blood promptly at ambient temperature.

**NOTE:** Specimen must arrive within 96 hours of draw.

**LIST FEE:** \$550.00

**CPT CODE:** 83890/Molecular isolation or extraction  
83892/Enzymatic digestion  
83894/Separation by gel electrophoresis  
83900/Amplification, target, multiplex, first 2 nucleic acid sequences  
83901/Amplification, target, multiplex, each additional nucleic acid sequence  
beyond 2  
83909/x8 Separation and identification by high-resolution technique  
83912/Interpretation and report

**ANALYTIC TIME:** 4 days

**DAY(S) SET-UP:** Monday; 2 p.m.

**QUESTIONS:** Contact your Mayo Medical Laboratories' Regional Manager  
Sara Siewert, Mayo Medical Laboratories' Technical Support  
Telephone: 800-533-1710



**MAYO CLINIC**  
 Mayo Medical Laboratories

# TEST DEFINITION

9/18/2008

CODE NAME  
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 83667 FAMILIAL AMYLOIDOSIS, DNA SEQUENCE

ORDER CODE	EFF DATE	TC	TITLE	CHECKING NORMALS	PRINT NORMALS (# CODED)	PERFORM SITE *
83667	7/23/2004		FAMILIAL AMYLOIDOSIS, DNA SEQUENCE			MCR
			TRANSPORT TEMP : AMBIENT\REFRIG OK\FROZEN NO			
			22635 SPECIMEN			
			22636 SPECIMEN ID			
			22637 SOURCE			
			22638 ORDER DATE			
			22639 REASON FOR REFERRAL			
			22640 METHOD			
			22641 RESULT			
			22642 INTERPRETATION			
			22643 AMENDMENT			
			22644 REVIEWED BY:			
			22645 RELEASE DATE			

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 MCR MAYO CLINIC DPT OF LAB MED & PATHOLOGY  
 200 FIRST STREET SW  
 ROCHESTER, MN 55905

LAB DIRECTOR: FRANKLIN R. COCKERILL, III, M.D.

TOTAL OF 0 NORMALS CODES

\*\*\* END OF REPORT \*\*

To obtain test set-up information contact MML Client Support or log on to:  
<http://www.mayomedicallaboratories.com/customer-service/set-up/index.html>



## LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME TESTING, MICHELLE		PATIENT NUMBER		AGE 32	SEX F	ACCESSION # W1357323
ORDERING PHYSICIAN		CLIENT ORDER #				ACCOUNT # LIAISONS
COLLECTION 05/21/09 12:35 P	RECEIVED 05/21/09 12:35 P	REPORT PRINTED 05/21/09 12:52 P		SPECIMEN INFORMATION DATE OF BIRTH:		
<b>DATE</b> <b>TIME</b>	<b>DATE</b> <b>TIME</b>	<b>DATE</b> <b>TIME</b>	<b>DATE</b> <b>TIME</b>			
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
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**Familial Amyloidosis, DNA Sequence**

<b>Specimen</b>	Blood	<b>MCR</b>
<b>Specimen ID</b>	722005	<b>MCR</b>
<b>Order Date</b>	21 May 2009 12:38	<b>MCR</b>
<b>Reason for Referral</b>		<b>MCR</b>

Not Provided. Test for the presence of a mutation within the transthyretin (TTR) gene.

**Method** **MCR**

DNA sequence analysis was performed to test for the presence of a mutation in exons 1 through 4 of the TTR gene. Mutation nomenclature is based on GenBank accession number; NM 000371.

**Result** **MCR**

A mutation was NOT detected.

**Interpretation** **MCR**

If testing was ordered to rule out familial amyloidosis, these results do not rule out the diagnosis since disease-causing mutations in other genes have been described.

TTR-associated familial amyloidosis is the most common form of inherited amyloidosis. It is important to note that other hereditary forms of amyloidosis (i.e. amyloidosis due to mutations in other genes, such as: Apolipoprotein AI, Apolipoprotein AII, lysozyme, gelsolin, cystatin C, or fibrinogen A alpha-chain) can cause a similar clinical presentation. This assay does not detect mutations associated with these other forms of familial amyloidosis. If testing was ordered because of an existing family history, please contact the Molecular Genetics Laboratory for an amended report. A genetic consultation may be of benefit.

\* Perform Site Legend on last page of report

PATIENT NAME TESTING, MICHELLE	ORDER STATUS Final	COLLECTION DATE AND TIME 05/21/09 12:35 P
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DATE	TIME	DATE	TIME	DATE	TIME	
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TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
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A list of common polymorphisms identified for this patient is available upon request.

**CAUTIONS:**

Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered. Test results should be interpreted in context of clinical findings, family history, and other laboratory data.

Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Reviewed By: Keri Jane Kruckeberg  
Release Date: 21 May 2009 12:39

MCR  
MCR

## \* PERFORMING SITE

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
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PATIENT NAME TESTING, MICHELLE	ORDER STATUS Final	COLLECTION DATE AND TIME 05/21/09 12:35 P
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Specimen receipt and report times are in CST/CDT

REPRINT

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