

# Laboratory Service Report

# 1-800-533-1710

MCR

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Patient Name SAMPLEREPORT,A1ATR	Patient ID SA00042654	<b>Age</b> 45	Gender F	Order # SA00042654
Ordering Phys				<b>DOB</b> 06/10/1966
Client Order # SA00042654	Account Information	1		Report Notes
<b>Collected</b> 01/30/2012 15:57	3050 SUPERIOR DRI	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE		
<b>Printed</b> 02/02/2012 08:48	ROCHESTER,MN 559	901		

Reference Perform Test Flag Results Unit Value Site\*

A1AT Phenotype, S REPORTED 02/02/2012 08:43 Alpha-1-Antitrypsin Phenotype MZbands

Heterozygous for M and Z isoforms. This phenotype may be associated with modestly reduced alpha-1-antitrypsin

concentrations.

AlaT Deficiency Profile REPORTED 02/02/2012 07:39

Alpha-1-Antitrypsin Genotyping, B

Alpha-1-Antitrypin, S

68 mg/dL (100-190 mg/dL)

Alpha-1-Antitrypsin Phenotype MZ

A1AT Genotype

S allele: Not detected.

Z allele: Heterozygous. One copy of the Z allele

identified.

Interpretation

The alpha 1 antitrypsin (A1A) serum level is lower than expected for an individual carrying one copy of the Z

allele. AlA phenotyping analysis was performed to rule out the presence of other gene variants. Phenotyping

demonstrated the MZ phenotype.

Taken together these findings are most consistent with carrier status for AlA deficiency. Carriers are usually not at risk for developing symptoms related to A1A deficiency. However, if A1A serum concentrations fall below protective amounts (approximately 60 mg/dL) they may be at risk for developing symptoms related to this disorder. Therefore, this result should be interpreted in the context of clinical presentation and other laboratory tests.

Since a mutation has been identified, genetic testing of other at risk family members is possible.

A genetic consultation may be of benefit.

#### 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

### \*\*\*Performing Site Legend on Last Page of Report\*\*\*

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,A1ATR	01/30/2012 15:57	Final
Page 1 of 2		>> Continued on Next Page >>

<sup>\*</sup> Report times for Mayo performed tests are CST/CDT



Specimen

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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.

Laboratory developed test.

Specimen ID

Order Date

Reason For Referral
Determine the A1A genotype.

Method

A PCR based analysis was used to specifically detect the S and Z alleles associated with A1A deficiency in the SERPINA1 gene. Other variants are not detected by this assay. The serum A1A level was measured by nephelometry, and A1A phenotyping was performed by isoelectric focusing.

Extraction Performed?

Yes

Reviewed By

Melody Elizabeth Kimball

Release Date 02 Feb 2012 07:38 MCR

Blood

# \* Performing Site:

MCR Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905 Lab Director: Franklin R. Cockerill,	, III, M.D.
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Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT.A1ATR	01/30/2012 15:57	Final
Page 2 of 2		

<sup>\*</sup> Report times for Mayo performed tests are CST/CDT