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Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Sex: 51 years Male

Specimen Collected: 08-Mar-22 11:06

A1A Genotype with Reflex to Phenotype | **Received: 08-Mar-22 11:06** **Report/Verified: 10-Mar-22 14:36**

Procedure	Result	Units	Reference Interval
Alpha-1-Antitrypsin	182 [i1]	mg/dL	90-200
Alpha-1-Antitrypsin Genotype Specimen	Whole Blood		
Alpha-1-Antitrypsin S Allele	Negative		
Alpha-1-Antitrypsin Z Allele	Negative		
Alpha-1-Antitrypsin Interpretation	See Note [f1 i2]		
Alpha-1-Antitrypsin Phenotype	Not Applicable		

Result Footnote

f1: Alpha-1-Antitrypsin Interpretation

Indication for testing: Carrier screening or diagnostic testing for alpha-1-antitrypsin (AAT) deficiency.

Negative: This sample has a serum AAT protein concentration in the normal range and is negative for the S and Z deficiency alleles by genotyping. This individual is not predicted to be affected with AAT deficiency; however, rare deficiency alleles are not detected by this genotyping assay.
This result has been reviewed and approved by Rong Mao, M.D.

Test Information

i1: Alpha-1-Antitrypsin
To convert to umol/L, multiply mg/dL by 0.185

i2: Alpha-1-Antitrypsin Interpretation
BACKGROUND INFORMATION: A1A (SERPINA1) Enzyme Concentration and 2 Mutations with Reflex to A1A Phenotype

CHARACTERISTICS of Alpha-1-Antitrypsin (AAT) Deficiency: Coughing, wheezing, bronchiectasis, chronic obstructive pulmonary disease, emphysema, and cirrhosis.
INCIDENCE: 1 in 3000 to 5000 North American individuals.
INHERITANCE: Autosomal recessive.
CAUSE: Two pathogenic mutations in the SERPINA1 gene on opposite chromosomes.
CLINICAL SENSITIVITY: 95 percent.
MUTATIONS TESTED: S allele (c.791A>T) and Z allele (c.1024G>A).
METHODS: Genotyping performed by polymerase chain reaction (PCR) and fluorescence monitoring; AAT protein concentration measured using immunoturbidmetric assay; phenotyping performed by isoelectric focusing electrophoresis. Genotyping and AAT serum protein concentration determination are performed on all specimens. Protein phenotyping is only performed on specimens that have AAT protein concentrations of

Test Information

i2: Alpha-1-Antitrypsin Interpretation

less than 90 mg/dL and are not homozygous or compound heterozygous for the S or Z deficiency alleles by genotyping.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: SERPINA1 mutations, other than the S (c.791A>T) and Z (c.1024G>A) alleles, will not be detected. Diagnostic errors can occur due to rare sequence variations.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.