Warde Medical Laboratory

LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 08/17/2023 14:13 Received: 08/17/2023 14:13

Test Name	Result	Flag	Ref-Ranges	Units	Site

Alpha-1-Antitrypsin Genotype

Alpha-1-Antitrypsin Genotype	See Below				QCRL

RESULT: NO MUTATION DETECTED

Interpretation: DNA testing indicates that this individual is negative for the PI*Z and PI*S alleles in the alpha-1-antitrypsin (PI) gene (genotype PI*M/PI*M). This negative result does not rule out the presence of other mutations within the PI gene or other causes of alpha-1-antitrypsin deficiency. Therefore, these results should be interpreted in the context of the individual's clinical presentation, and other laboratory tests such as measurement of serum alpha-1-antitrypsin levels.

Laboratory results reviewed and released by qualified personnel.

Alpha-1-antitrypsin deficiency is a relatively common autosomal recessive condition. The two most common deficiency alleles in the alpha-1-antitrypsin gene (protease inhibitor locus, PI) are designated PI*Z and PI*S, and the normal allele is designated PI*M. The PI*Z/PI*Z, PI*S/PI*Z, and PI*S/PI*S genotypes associated with decreased serum PI levels that are equivalent to approximately 10-20%, 35-40%, and 50-60% of normal, respectively. The PI*Z/PI*Z and PI*S/PI*Z genotypes are reported to be associated with an increased risk of liver disease in childhood, and chronic obstructive pulmonary disease (COPD) and emphysema in adult life. The PI*M/PI*Z, and PI*M/PI*S genotypes are also associated with decreased serum PI levels but these levels, and the PI levels associated with the PI*S/PI*S genotype, are apparently adequate to protect the lungs in the vast majority of individuals. Individuals with the PI*M/PI*Z genotype may have decreased pulmonary function, and may be at increased risk for COPD, especially if they smoke.

It should be noted that serum alpha-1-antitrypsin levels can be induced by a wide variety of conditions that include pregnancy, infection, numerous inflammatory conditions, cancer, and liver disease. Levels of alpha-1-antitrypsin may be reduced by other conditions. Therefore, immunological and functional determinations of serum alpha-1-antitrypsin levels may not correlate with the individual's PI genotype.

The PI*Z, PI*S, and PI*M alleles are detected by multiplex polymerase chain reaction (PCR) amplification of specific regions of the PI gene, followed by restriction enzyme digestion and capillary electrophoresis. This assay does not test for the presence of other mutations within the alpha-1-antitrypsin gene or non-genetic causes

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of alpha-1-antitrypsin deficiency. Although rare, false positive or false negative results may occur. All results should be interpreted in the context of clinical findings, relevant history, and other laboratory data. Health care providers may also contact your local Quest Diagnostics' genetic counselor or call 1-866-GENEINFO (866-436-3463) for assistance with the interpretation of these results.

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols Institute San Juan Capistrano. It has not been cleared or approved by FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.
Test Performed at:
Quest Diagnostics Nichols Institute
33608 Ortega Highway
San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

Clinical Indication	N/A	QCRL
Referring Physician	N/A	QCRL
Physician Phone	N/A	QCRL

Performing Site:
QCRL: QUEST DIAGNOSTICS REFERENCE LAB CAPISTRANO 33608 Ortega Highway San Juan Capistrano CA 92675

Reported Date: 2023.08.17 14:13 A1AGQ

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED