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Patient Report

Patient Age/Sex: 47 years Male

Specimen Collected: 08-Mar-22 10:57

Apolipoprotein E (APOE) Genotyping, AZ | **Received: 08-Mar-22 10:57** | **Report/Verified: 10-Mar-22 14:14**

Procedure	Result	Units	Reference Interval
APOE Specimen	Whole Blood		
APOE Alzheimer Disease Risk,Genotype	e2/e2 * f1 i1		

Result Footnote

f1: APOE Alzheimer Disease Risk, Genotype

Indication for testing: Determine APOE genotype for the purpose of Alzheimer disease risk assessment.

Homozygous APOE e2/e2: This genotype is associated with a decreased risk for Alzheimer disease (AD); however, the diagnosis of AD is primarily based on clinical evaluation, and APOE genotype alone is not sufficient to diagnose or exclude AD.

Homozygosity for the e2 allele is also associated with an increased cardiovascular risk. Consider further evaluation of this individual for hyperlipoproteinemia III (HPL III) or premature cardiovascular heart disease (CHD).

This result has been reviewed and approved by Rong Mao, M.D.

Test Information

i1: APOE Alzheimer Disease Risk, Genotype

BACKGROUND INFORMATION: Apolipoprotein E (APOE) Genotyping, Alzheimer Disease Risk

Characteristics: Alzheimer disease (AD), the most common cause of dementia, is characterized by progressive cognitive decline including memory, problem-solving skills, multi-step tasks, planning, and changes in personality. A clinical diagnosis of probable AD can be made based on clinical signs and neuroimaging, and the diagnosis is confirmed postmortem based on neuropathologic findings. The e4 allele of the APOE gene has been widely demonstrated to be associated with increased risk of AD. In individuals with a clinical diagnosis of AD, the presence of the e4 allele increases the likelihood that the diagnosis is correct, but is not diagnostic alone. APOE genotyping is not recommended for predicting AD risk in asymptomatic individuals.

Prevalence of APOE e4: Heterozygosity and homozygosity for the e4 allele is present in approximately 25 percent and 1-2 percent of the general population, respectively.

Inheritance of APOE e4: Semi-dominant.

Penetrance of APOE e4: Incomplete and influenced by age, gender, ethnicity, family history and environmental factors. The e4 allele is neither necessary nor sufficient for diagnosing AD; therefore, not all individuals with AD have the e4 allele and not all individuals with the e4 allele will develop AD.

Cause: Multi-factorial.

Variants Tested: Two single nucleotide polymorphisms in the APOE gene at codons 130 (rs429358) and 176 (rs7412). The e3 allele (Cysteine at 130 and Arginine at 176) is the most common in the general population. The e4 allele (Arginine at 130 and 176) is associated with increased AD risk. The e2 allele (Cysteine at codons 130 and

Test Information

i1: APOE Alzheimer Disease Risk, Genotype

176) may be associated with a lower risk for AD but homozygosity has been associated with increased risk for type III hyperlipoproteinemia.
Clinical Sensitivity: Approximately 30-60 percent of individuals diagnosed with AD carry at least one e4 allele. The e4/e4 genotype is found in approximately 13 percent of the AD population and 20 percent of the familial AD population.
Methodology: Polymerase chain reaction (PCR) and fluorescence monitoring.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Only the APOE alleles e2, e3 and e4 will be detected; rare alleles are not detected by this test. 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.