

Specimen ID: 026-127-9660-0
Control ID:
Acct #: 90000999 **Phone:** (336) 436-8645 **Rte:** 00
 LabCorp Test Master
 Test Account
 5450 Millstream Road
 MCLEANSVILLE NC 27301

NORMAL, 504040


Patient Details	Specimen Details	Physician Details
DOB: 01/11/1990 Age(y/m/d): 029/00/15 Gender: F SSN: Patient ID:	Date collected: 01/26/2019 0000 Local Date received: 01/26/2019 Date entered: 01/26/2019 Date reported: 02/09/2019 0000 ET	Ordering: Referring: ID: NPI:

Ordered Items

APOE Alzheimer's Risk

TESTS	RESULT	FLAG	UNITS	REFERENCE	INTERVAL	LAB
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APOE Alzheimer's Risk

 Methodology: 01

Patient DNA is assayed for the APOE genotype by PCR amplification of a specific region in exon 4 of the APOE gene followed by digestion with restriction enzyme Hha I and separation of fragments by polyacrylamide gel electrophoresis. This approach allows the APOE E2, E3, and E4 alleles to be distinguished. Analytical sensitivity and specificity are >99.5%. Individuals are interpreted as having one of the following genotypes: E2/E2, E3/E3, E4/E4, E2/E3, E2/E4, E3/E4.

 APO E Genotyping Result: E2/E2 01

 Interpretation: 01

Negative for the APOE4 variant that is associated with increased risk for late onset Alzheimer's disease (AD). APOE2 may have some protective effect against the development of AD.

RECOMMENDATIONS

Genetic counseling is recommended. Due to the lack of measures to prevent the development of AD, the ACMG/NSGC guidelines do not recommend presymptomatic testing, but if it is performed, guidelines are provided (Goldman JS et al. 2011). The APOE Genotyping: Alzheimer's Risk test is not recommended for children.

NOTE: This is not a diagnostic test. Results should be interpreted along with clinical findings and other data. This test evaluates only for the APOE genotype and cannot detect genetic abnormalities elsewhere in the genome. It should be realized that there are possible sources of error including sample misidentification, rare technical errors, trace contamination of PCR reactions, and rare genetic variants that may interfere with analysis.

For inquiries or genetic consultation, please call Esoterix at 1-800-444-9111.

 Comment: 01

INFORMATION ABOUT THE APOE GENOTYPE AND ALZHEIMER'S DISEASE

Patient: **NORMAL, 504040**
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Alzheimer's disease (AD) is the most common form of dementia in the elderly and currently affects more than 5 million Americans. It is a progressive neurodegenerative disorder with brain findings of plaques and neurofibrillary tangles containing beta-amyloid and tau protein respectively.

The predominant form of AD is late onset (age > 60-65), which can be familial (15-20%) or sporadic. The APOE4 variant increases the risk for late onset AD and may contribute to the pathology of the disease. This risk is increased by approximately 2 to 3-fold for individuals with one copy of the APOE4 variant and by approximately 10 to 15-fold for individuals with two copies of this variant (E4/E4 genotype). The APOE2 variant has some protective effect against development of late onset AD. The lifetime risk for late onset AD is approximately 10-12% in the general population, though it is higher in women than men and doubles when there is a first degree relative with this disorder. The lifetime risk is approximately 9% for individuals negative for APOE4, and for individuals with E4/E4 may be as high as 25% for males and 45% for females. Among patients with late onset AD, the presence of APOE4 may lead to earlier development of symptoms.

However, APOE4 is neither necessary nor sufficient for the development of AD. Approximately 30-50% of patients with late onset AD do not have an APOE4 allele.

APOE4 is common, with 25% of the general population having one copy and 1% having two copies of this variant. Among patients with late onset AD, 50-70% are positive for APOE4.

The development of late onset AD is influenced by many factors other than APOE4 including age, gender, family history, level of education and history of head trauma. Midlife cardiovascular risk factors in individuals with APOE4 also increase risk for cognitive decline. A number of genetic influences in addition to APOE4 have also been reported and are under investigation.

This test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary.

REFERENCES

Altmann A et al. Sex modifies the APOE-related risk of developing Alzheimer disease. *Annal Neurol* 2014;75(4):563-573

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<p>Bird TD. Alzheimer Disease Overview. GeneReviews (internet). Pagon RA et al., editors. Seattle WA: University of Washington, Seattle, WA. Last revised 2014.</p> <p>Goldman JS et al. Genetic counseling and testing for Alzheimer disease: Joint practice guidelines of the American College of Medical Genetics and the National Society of Genetic Counselors. Genet in Med 2011;13(6)597-605.</p> <p>Schipper HM. Apolipoprotein E: Implications for AD neurobiology, epidemiology and risk assessment. Neurobiology of Aging 2011;32:778-790</p>						

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For inquiries, the physician may contact **Branch: 800-222-7566 Lab: 800-282-7300**

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APO E Genotyping Result: 01

E4/E4 (two copies of the APOE4 variant)

Interpretation: 01

Positive for two copies of the APOE4 variant that is associated with increased risk of late onset Alzheimer's disease (AD). Therefore the lifetime risk for AD is increased in this individual. However, many individuals with the E4/E4 genotype do not develop AD.

RECOMMENDATIONS

Genetic counseling is recommended. Due to the lack of measures to prevent the development of AD, the ACMG/NSGC guidelines do not recommend presymptomatic testing, but if it is performed, guidelines are provided (Goldman JS et al. 2011). The APOE Genotyping: Alzheimer's Risk test is not recommended for children.

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Comment :

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