Apolipoprotein E (APOE) Genotyping, Cardiovascular Risk

Order Name: **APO E MUT**
Test Number: 2015053
Revision Date: 12/05/2025

TEST NAME	METHODOLOGY	LOINC CODE
Apolipoprotein E (APOE) Genotyping, Cardiovascular Risk	Illumina SNP Array	See Notes

SPECIMEN REQUIREMENTS

Specimen	Specimen Volume (min)	Specimen Type	Specimen Container	Transport Environment
Preferred	**4.5 mL**	**Whole Blood**	**EDTA (Lavender Top)**	**Refrigerated**

Instructions

Specimen: Whole blood from One 4.5mL adult tube Lavender-top (EDTA). Keep Refrigerated, Do Not Spin.
Collection: Invert tube immediately eight to 10 times once tube is filled at time of collection.
Stability Requirements: Room temperature 3 days for whole blood; Refrigerated 60 days; Frozen n/a
Cause for Rejection: Wrong specimen container; mislabeled specimen

GENERAL INFORMATION

Expected TAT

5-7 Days

Clinical Use

Apolipoprotein E (apoE) is a component of several lipoproteins. There are three main apoE isoforms reflecting three alleles (E2, E3 and E4), of which E3 is the most common. The APOE genotype E2/E2 is present in approximately 1% of the population and predisposes to type III hyperlipoproteinemia (dysbetalipoproteinemia). Type III hyperlipoproteinemia involves abnormal build-up in the blood of remnant chylomicrons and VLDL particles (collectively called B-VLDL) containing cholesterol and triglycerides. Excess B-VLDL is taken up in blood vessel walls and can lead to atherosclerosis. Therefore, individuals with type III hyperlipoproteinemia have high total serum cholesterol, LDL and triglycerides. The main clinical manifestation is premature atherosclerosis, leading to coronary artery disease or peripheral vascular disease. However, some patients may be asymptomatic. Only 1% to 5% of individuals with the E2/E2 genotype develop type III hyperlipoproteinemia. However, the E2/E2 genotype is present in 95% of individuals with type III hyperlipoproteinemia and is diagnostic of the disorder in individuals with the appropriate lipid profile. A number of secondary factors are known to provoke type III hyperlipoproteinemia in individuals with the E2/E2 genotype, including glucose intolerance, diabetes mellitus, hypothyroidism, obesity, low estrogen levels and excessive alcohol intake.

Notes

RESULT CODE	RESULT NAME	LOINC
2015053	APO E for Cardiovascular Risk	42315-2
5197137	APOE Additional Comments	77202-0
5197138	APOE Limitations	62364-5
5197139	APOE Methodology	49549-9
5197140	APOE Background Information	62364-5
5197141	APOE References	75608-0
5197142	APOE Result release:	18771-6

CPT Code(s)

81401

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