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2300xxxx

REPORT

Scientific Director: George Nasioulas PhD.

SAMPLE INFORMATION

Name:
Id/Medical ID:
Date of Birth:
Material: WHOLE PERIPHERAL BLOOD
Κωδικός Δείγματος : /

Date Recieved:
Date Of Report:
Req. Physician:
Barcode: 2300xxx

Complete genetic analysis of thrombophilia & cardiovascular diseases

Results

MUTATION / POLYMORPHISM	*Result*
Factor V Leiden	***Normal***
FVHR2 mutation	***Heterozygous***
Coagulation factor II (prothrombin) G20210A mutation	***Normal***
MTHFR C677T mutation	***Normal***
MTHFR A1298 mutation	***Homozygous mutant***
Factor XIII (fibrin stabilizing factor) V34L polymorphism	***Heterozygous***
FGB (fibrinogen B) 455 G>A polymorphism	***Normal***
PAI-1 (plasminogen activator inhibitor-1) 4G polymorphism	***4G/4G***
1b polymorphism of the human platelet antigens (HPA)	***1α/1α***
Apolipoprotein B R3500Q polymorphism	***Normal***
Apolipoprotein E	***E3/E3***
eNOS (endothelial nitric oxide synthase 3) G298A	***Heterozygous***
GPIa (glycoprotein 1A) C807T polymorphism	***Normal***
LPA (lipoprotein A) A5673G polymorphism	***Normal***
AGT (angiotensinogen) T174M polymorphism	***Heterozygous***
AGT (angiotensinogen) M235T polymorphism	***Heterozygous***

Eirini Papadopoulou, PhD
Molecular Biologist

Scientific Director
George Nasioulas, PhD
Molecular Biologist

Methodology

Total genomic DNA was extracted from the sample under investigation. 15 polymorphisms in 13 genes associated with thrombophilia and cardiovascular disease were tested using a targeted replication method (Ion AmpliSeq NGS Panel - Thermo Fisher Scientific). Sequencing was performed using the Ion Gene Studio S5 Prime System (Thermo Fisher Scientific).

Table of genotype / phenotype relationship:

GENE	MUTATION/ POLYMORPHISM	PHENOTYPE
Factor V	G1691A or R508Q (Leiden)	Leads to activated protein C, is detected in 20-50% of patients with venous thrombosis and is one of the strongest genetic risk factors for thrombophilia
	H1299R (R2)	Mild risk factor for thrombosis, but increases the risk of cardiovascular disease in Leiden mutation carriers
Prothrombin	G20210A	Carriers have 30% increased levels of prothrombin in their blood and 3 times higher risk of thrombosis than normal individuals. The risk increases significantly in combination with the Leiden mutation
Factor XIII (fibrin stabilizing factor) (F13A1)	V34L	Associated with a potentially protective role against venous thrombosis
β-fibrinogen	455 G>A	*Elevated plasma fibrinogen levels* increase the risk of myocardial infarction and ischemic stroke
Plasminogen activator inhibitor-1 (PAI-1)	4G / 5G	4G allele is associated with increased plasminogen concentration in blood and is a mild risk factor for venous thrombosis and myocardial infarction
Human Platelet Antigens (HPA)	1a / 1b	1b Allele poses an increased risk of premature myocardial infarction and stroke, especially in smokers
Methylene tetrahydrofolate reductase (MTHFR)	C677T	Leads to reduced enzyme activity of the enzyme and increased levels of homocysteine in blood. Homozygosity predisposes to thrombosis, in combination with other risk factors
	A1298C	In combination with the C677T mutation it leads to reduced enzyme activity
Apolipoprotein B (ApoB)	R3500Q	Prevalent but rare genetic abnormality that causes hypercholesterolemia and increased risk of atherosclerosis
Apolipoprotein E (ApoE)	Codon 112 (Cys / Arg) Codon 158 (Cys / Arg)	Important prognostic factors for blood lipid levels. E2 isoform is related to lower serum LDL and cholesterol levels whereas E4 is related to the highest levels. E4 allele is a risk factor for atherosclerotic plaque and coronary heart disease. An increased incidence of E4

		allele has also been observed in families with a family history of early onset Alzheimer's [PMID: 7175379, PMID: 834644]
eNOS	G298A	This polymorphism is associated with an increased risk of preeclampsia and heart disease, with heterozygotes being twice as likely to develop hypertension during pregnancy. Homozygotes have a 1.5 times higher risk of ischemic heart attack [PMID 16059745] [PMID15007011]
GPIa	C807T	T allele is associated with an increased risk of myocardial infarction and ischemic stroke [PMID 10194421]
LPA	A5673G	*LPA* polymorphisms are associated with coronary heart disease (CHD), coronary artery disease (CVD), early atherosclerosis and thrombosis. This mutation, either in heterozygosity or homozygosity, is associated with elevated levels of lipoprotein A. Mutation carriers have an increased risk of heart disease, but seem to benefit from low dosage aspirin (100mg every other day) [PMID 18775538]
AGT	M235T T174M	M235T and T174M polymorphisms are associated with increased levels of angiotensinogen in the blood and increased blood pressure. Their detection is associated with the occurrence of hypertension and coronary heart disease.

***Note:** Each analysis has an internal error probability of 0,5-1%. This is due to rare events and factors involved in the production and analysis of specimens.