Cardiovascular Disease (CVD) Genetic Marker

Protein	Gene	Mutation/Polymorphism (Trivial name)	Ref SNP*)	Biological consequences	Clinical effects / Disease
Blood Coagulation Factor V	F5	1691 G>A (Leiden)	rs6025	altered cleavage site prevents efficient inactivation of FV, confers APC resistance	increased risk of clot formation in veins, risk factor for venous thromboembolism (VTE)
		His1299Arg (HR2 haplotype)	rs1800595	impaired cofactor activity in APC-mediated FVIIIa inactivation	increased risk for venous thrombosis in carriers of FV Leiden
Prothrombin (Blood Coagulation Factor II)	F2	20210 G>A	rs1799963	associated with elevated prothrombin levels	increased risk for cerebral and deep vein thrombosis
5,10-Methylene-tetrahydrofolate Reductase	MTHFR	677 C>T	rs1801133	reduced enzyme activity	hyperhomocysteinemia, risk factor for VTE
		1298 A>C	rs1801131	reduced enzyme activity	hyperhomocysteinemia, risk factor for VTE
Blood Coagulation Factor XIII	F13A1	Val34Leu	rs5985	altered structure of cross-linked fibrin, results in less stable clots	protective effect against VTE and myocardial infarction (MI), elevated risk for recurrent pregnancy loss
Plasminogen Activator Inhibitor 1 (PAI-1)	Serpine1	4G/5G	rs1799762	4G allele is associated with increased PAI-1 expression	risk factor for venous thromboembolism and pregnancy complications
Endothelial Protein C Receptor (EPCR)	EPCR	4600 A>G	rs867186	define haplotypes A1, A2 and A3; A3 haplotype increases soluble EPCR plasma levels	haplotype A1 exerts a slight protective effect on VTE and recurrent pregnancy loss in carriers of other genetic risk factors
		4678 G>C	rs9574		
Endothelial Nitric Oxide Synthase (eNOS)	eNOS	-786 T>C	rs2070744	reduced transcription rate of eNOS	increased risk for coronary artery disease
		894 G>T (Glu298Asp)	rs1799983	unknown	increased risk for premature MI
Lymphotoxin Alpha (LTA)	LTA	804 C>A (Thr26Asn)	rs1041981	increased VCAM1 and E-Selectin transcription	proinflammatory effect, increase risk of coronary artery disease
Angiotensin-Converting Enzyme (ACE)	ACE	I/D (Insertion/Deletion)	rs1799752	D allele is associated with higher ACE activity	DD genotype is a risk factor for fetal loss and MI in young smokers
Human Platelet Antigen 1 (HPA1, GP IIIa))	ITGB3	1a/b (Leu33Pro)	rs5918	conformational change of the ß$_3$-subunit impacts platelet aggregation	risk factor for early-onset MI
ß-Fibrinogen	FGB	-445 G>A	rs1800790	increased ß-fibrinogen plasma levels	associated with premature MI
Apolipoprotein B (Apo B)	ApoB	Arg3500Gln	rs5742904	altered conformation of Apo B receptor binding domain	associated with defective LDL and hypercholesterolemia
Apolipoprotein E (Apo E)	ApoE	Cys112Arg	rs429358	define Apo E isoforms E2, E3 and E4; they interact differently with specific lipoprotein receptors	E4 isoform is associated with increased susceptibility to coronary atheriosclerosis and Alzheimer Disease
		Arg158Cys	rs7412		

*) www.ncbi.nlm.nih.gov