sample type: **BUCCAL**

GENOVATIONS®

# CardioGenomicPlus® Profile

**CardioGenomicPlus® Profile** evaluates genetic variations, called single nucleotide polymorphisms (SNPs), in genes that modulate blood pressure regulation, lipid balance, nutrient metabolism, inflammation, and oxidative stress.

## The test uncovers potential genetic susceptibility to:

- Hypercholesteremia
- Atherosclerosis
- Obesity
- Hypertension
- Coronary Artery Disease
- Myocardial Infarction
- Thrombosis
- Endothelial Dysfunction
- Stroke

## Cholesterol Regulation and Atherosclerosis

**ApoE** (apoliprotein E)
**CETP** (cholesteryl ester transfer protein)
**SELE** (selectin E)
These genes affect how the body breaks down and clears fats and how cholesterol is processed. They also affect lipid balance, plaque formation, and blood vessel integrity and function.

## Methylation

**MTHFR** (methylenetetrahydrofolate reductase)
Polymorphisms of this enzyme can disrupt the metabolism of homocysteine, resulting in its accumulation as well as impaired methylation. The presence of these SNPs can increase risk of cardiovascular disease, blood vessel damage, thrombosis (blood clots), stroke, and degenerative aging.

## Hypertension

**GNB3** (guanine nucleotide-binding protein)
**AGTR1** (angiotensin II receptor–1)
Polymorphisms of these genes are associated with blood vessel constriction, sodium and water retention, obesity, and increased susceptibility to hypertension.

## Coagulation

**Factor 2** (prothrombin)
**Factor 5** (Leiden)
**PAI-1** (Plasminogen activator inhibitor-1)
**GP3a** (Glycoprotein 3)
These genetic variants can over-activate blood clotting processes, increasing the risk of sudden cardiac events such as thromboses, heart attacks, and strokes.

- ***Specimen Requirements:***
  - **Buccal**–Swab
- ***Before Patient Takes this Test:***
  - See instructions inside test kit for more details

GENOVA DIAGNOSTICS®

# *CardioGenomicPlus Profile*

GENOVA DIAGNOSTICS

63 Zillicoa Street
Asheville, NC 28801


Patient: **SAMPLE PATIENT**
Age: 54
Sex: M
MRN:

**Order Number:**
Completed: January 31, 2008
Received: January 26, 2008
Collected: January 15, 2008

## *Apo E* — *Apolipoprotein E : CHOLESTEROL REGULATION*

**Location:**
Chromosome 19
**APOE**
APO E2: cys / cys
APO E3: cys / arg
APO E4: arg / arg
**Your Genotype:**

| 2 | 3 |
|---|---|

The two SNPs lead to 3 possible variants for each chromosome, known as ApoE2, E3, & E4.

Apolipoprotein E (Apo E) plays a key role in lipid metabolism by helping to remove dietary cholesterol (chylomicrons and VLDL) from the bloodstream.

**Health Implications**

- The APO E2/3 genotype is common, accounting for 10-15% of most populations
- APO E2 is associated with lower LDL cholesterol and higher HDL-C, but higher triglycerides (as found in Metabolic Syndrome) compared to the other genotypes
- APO E2 also confers a lower risk of atherosclerosis, myocardial infarction, stroke, and osteoporosis, and higher antioxidant activity

**Treatment Options**

- The cholesterol-lowering effect of a low saturated fat and low cholesterol diet is least effective with E2 individuals
- Minimize high-glycemic index foods, which produce the largest triglyceride (TG) response in E2 carriers
- Dietary fiber, fish oils, and exercise generally improve the lipid profile in this genotype; fish oils reduce TGs most effectively in E2 individuals
- Alcohol may reduce LDL-C in men (neutral in women)
- E2 individuals generally respond the most favorably to statins and would therefore likely respond to statin mimetics such as inositol hexaniacinate, red rice yeast, and policosanol
- Gemfibrozil may be particularly effective at lowering TGs and total cholesterol
- HRT improves the lipid profile in this genotype, although oral estrogen may significantly increase TGs

**Key**

- - Neither chromosome carries the genetic variation.
+ - One chromosome (of two) carries the genetic variation.
+ + Both chromosomes carry the genetic variation.

*(You inherit one chromosome from each parent)*

+ ▲ Gene activity increased
+ ▼ Gene activity decreased

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GALA RMS 3079

## Customized commentary includes:

***Health Implications***
*Outlines disease risks associated with specific SNP results*

***Minimizing Risk***
*Suggests possible dietary, environmental, lifestyle, supplement, and pharmaceutical interventions to optimize genomic potential of patient*

***Further Evaluation***
*Alerts to the potential for related physiological imbalances and the clinical need for follow-up assessment*

## Related Phenotype Assessments

***Baseline:***
*To assess baseline expression of genetic tendency*

***Follow-up:***
*To regularly monitor therapeutic interventions that modify genetic expression*

- ***Comprehensive Cardiovascular Profile 2.0***
- ***Amino Acids Analysis*** *(plasma or urine)*
- ***Elemental Analysis*** *(hair, urine, or packed erythrocytes)*

For test kits, clinical support, or more information contact:

Client Services
Genova Diagnostics
63 Zillicoa St.
Asheville, NC 28801-1074
800-522-4762 • Fax: 828-252-9303

More detailed publications with references are also available: www.GDX.net