



**Genova
Diagnostics**[®]

Innovative Testing for Optimal Health

63 Zillicoa Street
Asheville, NC 28801
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Patient: **Sample Report**

Order Number:

IWDL Genova Diagnostics

Completed:

Parkgate House

Received:

356 West Barnes Lane

Collected:

New Malden

Surrey, KT3 6NB




<i>Apo E</i>		<i>Apolipoprotein E : CHOLESTEROL REGULATION</i>	
<p>Location: Chromosome 19 APOE APO E2: cys / cys APO E3: cys / arg APO E4: arg / arg Your Genotype:</p>	<p>Apolipoprotein E (Apo E) plays a key role in lipid metabolism by helping to remove dietary cholesterol (chylomicrons and VLDL) from the bloodstream.</p>		
	<p>Health Implications</p> <ul style="list-style-type: none"> · This genotype is the most common (accounting for >50% of most populations) and is the genotype against which E2 and E4 are compared · APO E3 confers only a moderate tendency toward elevated total- and LDL cholesterol, and lower HDL-C · Risk is intermediate between E2 and E4 for atherosclerosis, myocardial infarction, stroke (in smokers), and osteoporosis 		
<p>3 3</p>		<p>Treatment Options</p> <ul style="list-style-type: none"> · Effects of cholesterol and dietary fat on serum cholesterol levels is least with the E2 allele and greatest with the E4 allele; thus, dietary fat restriction produces a moderate cholesterol response in E3/E3 individuals · Dietary fiber, fish oils, and exercise generally improve the lipid profile in this genotype · Alcohol appears to have a neutral effect on LDL-C · Avoid smoking, which increases risk of coronary heart disease in this genotype · E3/E3 individuals generally respond well to statins and would therefore likely respond to statin mimetics such as inositol hexaniacinate, red rice yeast, and policosanol · Hormone replacement therapy generally improves the lipid profile in all genotypes, including post-menopausal E3 carriers 	
<p>The two SNPs lead to 3 possible variants for each chromosome, known as ApoE2, E3, & E4.</p>			



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







<i>MTHFR</i>		<i>5,10-methyltetrahydrofolate reductase : METHYLATION</i>	
<p>Location: Chromosome 1 C677T Your Genotype:</p>	<p>5,10-methylenetetrahydrofolate reductase (MTHFR) is a key enzyme in folate metabolism, facilitating the formation of methyltetrahydrofolate, a required cofactor in the remethylation of homocysteine (Hcy) to methionine.</p>	<p>Health Implications</p> <ul style="list-style-type: none"> · Heterozygosity for only 1298 (-/+) is associated with baseline "normal" MTHFR enzyme activity, suggesting adequate formation of methyl-THF. (Risk of enzyme impairment and elevated homocysteine is increased only when 677CT is also positive) 	<p>Treatment Options</p> <ul style="list-style-type: none"> · Ensure adequate intake of folic acid and vitamins B2, B3, B6 (pyridoxal 5-phosphate), and B12 (or methylcobalamin)
			
<p>A1298C Your Genotype:</p>	<p>Health Implications</p>		
			

<i>COMT</i>		<i>Catechol-O-MethylTransferase : METHYLATION</i>	
<p>Location: Chromosome 22.11q V158M Your Genotype:</p>	<p>COMT is a key enzyme in the deactivation of catechol compounds such as catecholamines, estrogens, various chemicals, and toxins. COMT modulates the neurotransmitter functions of dopamine and norepinephrine.</p>	<p>Health Implications</p> <ul style="list-style-type: none"> · Moderately decreased COMT activity with increased bioavailability of catecholamines and impaired methylation of catechol estrogens · Superior mental performance (increased brain dopamine), but increased risk of nervousness, excitability, and mood disturbances · Reduced pain threshold and increased risk of fibromyalgia 	<p>Treatment Options</p> <ul style="list-style-type: none"> · Ensure adequate B6, B12, folate, magnesium, and methionine to support formation of S-adenosylmethionine and prevent elevated homocysteine · Ensure adequate anti-oxidants to prevent oxidation of dopamine and pro-carcinogenic 4-hydroxyestrogens · Exercise caution using amphetamine-based medications
			

<i>IL-1β</i>		<i>InterLeukin-1b : INFLAMMATION</i>	
Location: Chromosome 2 -31C - T Your Genotype:	IL-1 β is an inflammatory cytokine produced by macrophages in response to stimuli such as bacterial lipopolysaccharide. IL-1 β inhibits acid secretion in the stomach and stimulates bone resorption.		
	Health Implications <ul style="list-style-type: none"> · Greater gastric IL-1β production and hypochlorhydria, with increased risk of atrophic gastritis. · Increased vulnerability to <i>Helicobacter pylori</i> infection and risk of gastric cancer · Possible <i>protection</i> against breast (post-menopausal) and lung cancer 		
	Treatment Options <ul style="list-style-type: none"> · Reduced alcohol consumption to diminish risk of atrophic gastritis · Avoid smoking, to decrease persistent <i>H. pylori</i> infection · Avoid COX-1 and COX-2 inhibitors that delay gastric ulcer healing · Consider extra vitamin C during <i>H. pylori</i> infection · IL-1β levels are reduced by: Ω-3 fatty acids, stinging nettle, curcumin, silymarin, green tea, boswellia, ginkgo, resveratrol, and L-glutamine 		

<i>VDR</i>		<i>Vitamin D Receptor : HORMONAL BONE FORMATION</i>	
Location: Chromosome 12 BsmI RFLP Your Genotype:	VDR is an intracellular hormone receptor that specifically binds the active form of vitamin D and interacts with target-cell nuclei to produce effects.		
	Health Implications <ul style="list-style-type: none"> · Slight impairment of vitamin D receptor with resistance to vitamin D3 · Slightly increased risk of impaired calcium absorption, increased bone loss, lower bone mineral density, and enhanced bone lead accumulation · Moderately reduced risk of prostate cancer 		
	Treatment Options <ul style="list-style-type: none"> · Carriers of the (+) allele benefit from vitamin D supplementation · Ensure adequate calcium (Ca) intake; studies suggest minimum of 900 mg/day · Vitamin K may help to compensate for the higher risk of bone loss · Caffeine intake >300 mg/day may accelerate bone loss, especially when low calcium intake · Favorable bone response to etidronate and raloxifene and HRT 		

<i>GSTM1</i>		<i>Glutathione S-Transferase mu-1 : DETOXIFICATION</i>	
Location: Chromosome 1 Your Genotype:	GST is responsible for Phase II detoxification of xenobiotics, carcinogens, and products of oxidative stress. GSTM1 is located primarily in the liver.		
	Health Implications <ul style="list-style-type: none"> · GSTM1 enzyme activity is absent, with reduced detoxification capacity · Increased risk of toxic burden, oxidative stress, atopic asthma, lung problems, cancer, chemical sensitivity, and coronary artery disease · Decreased risk of cancer, only with high intake of cruciferous vegetables 		
ABSENT	Treatment Options <ul style="list-style-type: none"> · Eat cruciferous vegetables and allium foods to reduce cancer risk · Eat a diet rich in antioxidants (colorful foods), consider supplementation · Ensure availability of glutathione precursors and cofactors · Limit glutathione depletion with α-lipoic acid, milk thistle, or taurine · Minimize exposure to xenobiotics, including PAHs and toxic metals 		
The GSTM1 gene is either PRESENT or ABSENT (also called Null). If either copy is present, it is termed PRESENT. If both copies are absent, it is termed ABSENT.			

<i>GSTP1</i>		<i>Glutathione S-Transferase pi-1 : DETOXIFICATION</i>
Location: Chromosome 11 A114V Your Genotype:	GST is responsible for Phase II detoxification of xenobiotics, carcinogens, steroids, heavy metals, and products of oxidative stress. GSTP1 is located primarily in the brain and lungs.	
		Health Implications <ul style="list-style-type: none"> · Polymorphisms are associated with either higher or lower enzyme activity, depending on specific environmental exposures; therefore, the (-/-) genotype may still increase risk for some disorders. The I105V snp is the more significant of the two. · The I105V genotype (-/-) is associated with slightly increased risk of some cancers (especially if exposed to cigarette smoke), also atopy, xenobiotic-induced asthma, and COPD Treatment Options <ul style="list-style-type: none"> · Ensure availability of glutathione precursors and cofactors, e.g., methionine-rich foods, NAC, L-glutamine, glycine, Mg, B6 · Eat a diet rich in antioxidants (colorful foods), consider supplementation · Minimize exposure to xenobiotics, including polycyclic aromatic hydrocarbons (e.g., cigarette smoke) and toxic metals
I105V Your Genotype:		
		

This test has been developed and its performance characteristics determined by Genova Diagnostics, Inc. It has not been cleared or approved by the U.S. Food and Drug Administration.

Commentary is provided to the practitioner for educational purposes, and should not be interpreted as diagnostic or treatment recommendations. Diagnosis and treatment decisions are the responsibility of the practitioner.

The accuracy of genetic testing is not 100%. Results of genetic tests should be taken in the context of clinical representation and familial risk. The prevalence and significance of some allelic variations may be population specific.

Any positive findings in your patient's test indicate genetic predisposition that could affect physiologic function and risk of disease. We do not measure every possible genetic variation. Your patient may have additional risk that is not measured by this test. Negative findings do not imply that your patient is risk-free.

The Third Wave™ Invader DNA assay is used to detect polymorphisms in the patient's DNA sample. In this assay, a solution hybridization method is used in which two oligonucleotides hybridize in tandem with the specific DNA sequences.

Subsequent Cleavase® and hybridization reactions result in generation of fluorescent signal. The bplex format of the assay enables simultaneous detection of all variants in a single reaction tube. The sensitivity and specificity of this assay is 99.7%.

Dr. Amy Peace-Brewer

Dr. Patrick Hanaway

Laboratory Director

Chief Medical Officer

