sample type: **BUCCAL**

GENOVATIONS®

EstroGenomic™ Profile

EstroGenomic™ Profile evaluates genetic variations, called single nucleotide polymorphisms (SNPs), in genes that modulate estrogen metabolism, coagulation, cardiovascular function, bone health and inflammation.

The test uncovers potential genetic susceptibility to:

- Breast Cancer
- Osteoporosis
- Thromboses
- Stroke
- Atherosclerosis
- Heart Disease

Estrogen Metabolism

- CYP1A1
- GST (M1 and P1)
- CYP1B1
- COMT (catechol-O-methyl transferase)

Estrogen metabolism SNPs focus on the Phase 1 enzymes involved in the formation of anti- or procarcinogenic metabolites such as 2-hydroxyestrone and 4-hydroxyestrone, respectively, as well as the Phase 2 enzymes responsible for the subsequent metabolism of these compounds. Levels of the various estrogen metabolites modulate risk of both breast cancer and osteoporosis.

HyperCoagulation

- GP3a (Glycoprotein 3)
- Factor 2 (Prothrombin)
- PAI-1 (Plasminogen activator inhibitor-1)
- Factor 5 (Leiden)

These genetic variants focus on estrogen's interaction with some of the key constituents of the clot formation and fibrinolysis process, such as clotting factors and inhibitors of fibrinolysis. The SNPs can over-activate blood clotting processes, increasing the risk of sudden cardiac events such as thromboses, heart attacks, and strokes, especially among women take supplemental estrogens and oral contraceptives.

Cardiovascular

- Apo E (Apoliprotein E)
- TNF-α
- MTHFR
- IL-6

These genes affect how the body processes cholesterol, responds to inflammation, and metabolizes B vitamins. The presence of these SNPs can increase risk of cardiovascular disease, hyperlipidemia, and blood vessel damage.

Osteoporosis

- VDR
- TNF-α
- IL-6

Osteoporosis SNPs relate to estrogen's influence on inflammation, bone resorption, vitamin D function and bone collagen formation.

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

GENOVA DIAGNOSTICS®

Estrogenomic Profile

GENOVA DIAGNOSTICS

63 Zillicoa Street
Asheville, NC 28801

Patient: **SAMPLE PATIENT**
Age: 44
Sex: F
MRN:

Order Number:
Completed: January 31, 2008
Received: January 16, 2008
Collected: January 14, 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

GENOVATIONS™

GALA RMS 3079

Related Phenotype Assessments

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

- ***Essence*** *(Women's Hormonal Health Assessment)*
- ***Estrogen Metabolism Plus*** *(urinary hormone metabolites)*
- ***Comprehensive Cardiovascular Assessment*** *(urine)*
- ***Bone Resorption*** *(urine)*
- ***Oxidative Stress*** *(blood or urine)*
- ***Detoxification Profile*** *(urine or saliva)*
- ***Metabolic Analysis Profile*** *(urine)*
- ***Amino Acids Analysis*** *(blood or urine)*

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