



## ImmunoGenomic® Profile

**ImmunoGenomic® Profile** evaluates genetic variations, called single nucleotide polymorphisms (SNPs), in genes that modulate immune and inflammatory activity. Polymorphisms affect the levels and activity of the cytokines. These variations can affect balance between cell-mediated (TH-1) and humoral (TH-2) immunity, reveal potential defects in immune system defense, and stimulate mechanisms leading to chronic, overactive inflammatory responses.

### The test uncovers potential genetic susceptibility to:

- Asthma
- Certain Cancers
- Infectious Diseases
- Arthritis
- Heart Disease
- *Helicobacter pylori* infection (cause of ulcers)
- Autoimmune Disorders
- Allergy
- Bone Inflammation
- Inflammatory Bowel Disease
- Osteopenia

### Chronic Inflammation

- IL-1 $\beta$  (interleukin-1beta)

This gene affects the duration and intensity of the acute inflammatory response. The polymorphism leads to increased production of IL-1 $\beta$ , hypochlorhydria, increased susceptibility to *H. pylori* infection, and gastritis and gastric cancer in *H. pylori*-infected individuals.

### TH-1 Cytokines (Viral Infection & Cancer)

- TNF- $\alpha$  (tumor necrosis factor-alpha)

Polymorphisms of this gene affect cell-mediated immunity, increasing production of the pro-inflammatory cytokine TNF-alpha. This can promote or exacerbate chronic conditions such as arthritis, osteoporosis, and asthma.

### TH-2 Cytokines (Allergy, Asthma, and Atopy)

- IL-4 (interleukin-4)
- IL-6 (interleukin-6)
- IL-10 (interleukin-10)
- IL-13 (interleukin-13)

TH-2 cytokines promote humoral immunity, including the synthesis of IgE. They are generally regarded as anti-inflammatory; however, excessive activity of some of these cytokines may promote the development of allergic conditions such as atopy, asthma, and hypersensitivity. Low levels may result in chronic inflammatory conditions characterized by a TH-1 response. IL-6 has both pro- and anti-inflammatory properties; this cytokine drives the acute phase response and can promote chronic inflammation and progression in autoimmune disease.

#### • Specimen Requirements:

- Buccal-Swab
- or

#### • Before Patient Takes this Test:

- See instructions inside test kit for more details



# ImmunoGenomic Profile

## GENOVATIONS

Patient: **SAMPLE PATIENT**

Age: 49

Sex: F

MRN:

Order Number:

Completed: February 22, 2008

Received: February 01, 2008

Collected: January 30, 2008

### Immuno Genomic Profile Results

This profile identifies genetic single nucleotide polymorphisms associated with increased risk of developing defects in immune competence and surveillance. Immune system polymorphisms have been associated with increased risk of asthma, atopy, osteopenia, arthritis, heart disease, auto-immunity and infectious diseases.

#### Chronic Inflammation

**IL-1?**: Interleukin 1-beta, produced mainly by blood monocytes, mediates the panoply of host inflammatory reactions collectively known as acute phase response. Polymorphisms in IL-1? may predispose individuals to chronic inflammatory conditions by upregulating COX2 activity and prostaglandin production. Other effects include hypochlorhydria, predisposition to H. pylori infection and gastric cancer.

#### TH-1 Cytokine (Viral Infection & Cancer)

**TNF-?**: Tumor necrosis factor-alpha is a pro-inflammatory cytokine that can contribute to arthritis, asthma and osteoporosis. Polymorphisms of TNF-? inappropriately activate inflammatory response and increase TNF-? production.

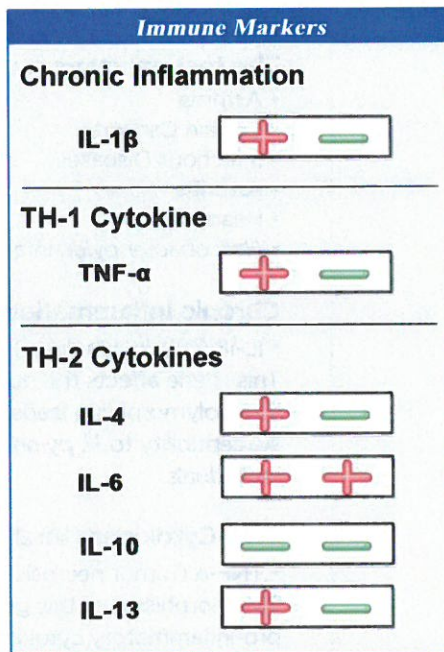
#### TH-2 Cytokines (Allergy, Asthma & Atopy)

**IL-4**: Polymorphisms in Interleukin-4 lead to increased IL-4 production and to decreased barrier function in lung epithelial cells causing a hyper-responsiveness to antigen stimulus, leading to increased risk and severity of bronchial asthma.

**IL-6**: Interleukin-6 contributes to inflammatory response and also affects adipose tissue metabolism, lipoprotein lipase activity, and hepatic triglyceride secretion. This particular SNP has been associated with elevations in serum triglycerides in response to carbohydrate intake and decreased levels of HDL cholesterol.

**IL-10**: Interleukin-10 has an inhibitory effect on TH-1 cytokine production. Polymorphisms in IL-10 may affect the risk of frequent viral infections, cancer and auto-immune diseases such as rheumatoid arthritis or lupus (SLE).

**IL-13**: Interleukin-13 acts to promote IgE synthesis and IgE-based mucosal inflammation typical of atopy and bronchial asthma. These SNPs are associated with increased IL-13 production and activity.



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 biplex 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%.

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DIAGNOSTICS

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### Related Phenotype Assessments

#### Baseline:

To assess baseline expression of immune-related genetic tendency

#### Follow-up:

To regularly monitor therapeutic interventions that modify genetic expression

- Allergy Antibody Assessment (foods and inhalants)
- Comprehensive Cardiovascular Assessment (urine)
- Oxidative Stress (blood or urine)
- H. Pylori Stool Antigen

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

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