sample type: **BUCCAL**

GENOVATIONS®

NeuroGenomic™ Profile

NeuroGenomic™ Profile evaluates single nucleotide polymorphisms (SNPs) in genes that modulate methylation, glutathione conjugation, oxidative protection (and the potential to evaluate vascular oxidation).

The test uncovers potential genetic susceptibility to:

- Neurodegenerative Disorders
- Mood Disorders
- Detoxification Capacity
- Developmental Issues
- Oxidative Stress

Methylation

- MTHFR (methylenetetrahydrofolate reductase)
- COMT (catechol-O-methyltransferase)

These genes affect how homocysteine and methionine are metabolized to support formation of S-adenosylmethionine (SAMe). The ability to donate methyl groups affects neurologic function and is modifiable by proper B-vitamin intake.

Detoxification

- GSTM (glutathione-s-transferase, M, isoforum)
- GSTP (glutathione-s-transferase, P, isoforum)

These genes are responsible for detoxifying products of oxidative stress and carcinogens. Variants decrease detoxification capacity.

Oxidative Protection

- SOD-2 (Superoxide dismutase -2)

These genetic variants alter anti-oxidant enzyme activity and modify requirements for anti-oxidants.

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

GENOVA DIAGNOSTICS®

NeuroGenomic™ Profile

GENOVA DIAGNOSTICS

63 Zillicoa Street
Asheville, NC 28801

Patient: **SAMPLE PATIENT**
Age: 10
Sex: M
MRN:

Order Number:
Completed: February 11, 2008
Received: January 31, 2008
Collected: January 28, 2008

MTHFR	*5,10-methyltetrahydrofolate reductase : METHYLATION*
Location: Chromosome 1 **C677T** **Your Genotype:** + / −	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. **Health Implications** · Heterozygosity for both 677 (-/+) and 1298 (-/+) results in 50-60% reduction in MTHFR enzyme activity, low folate status, and increased risk of elevated homocysteine (and S-adenosylhomocysteine, or SAH) · MTHFR polymorphism-induced SAH elevations may disrupt neurotransmitter metabolism as well as synthesis of DNA, carnitine, and coenzyme Q10 · Increased risk of autism, depression, neural tube defects, cardiovascular disease, diabetic retinopathy, osteoporosis, and some cancers · Low folate status significantly increases risk of associated disorders **Treatment Options** · Ensure adequate intake of folate-rich green vegetables · Consider supplementation with folic acid (or folinic acid or 5-methyltetrahydrofolate), vitamins B2, B3, B6 (pyridoxal 5-phosphate), B12 (or methylcobalamin), and betaine (trimethylglycine)
A1298C **Your Genotype:** + / −	

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

- ***Optimal Nutritional Evaluation*** *(ONE)*
- ***Oxidative Stress Profile*** *(blood or urine)*
- ***Detoxification Profile*** *(urine)*
- ***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