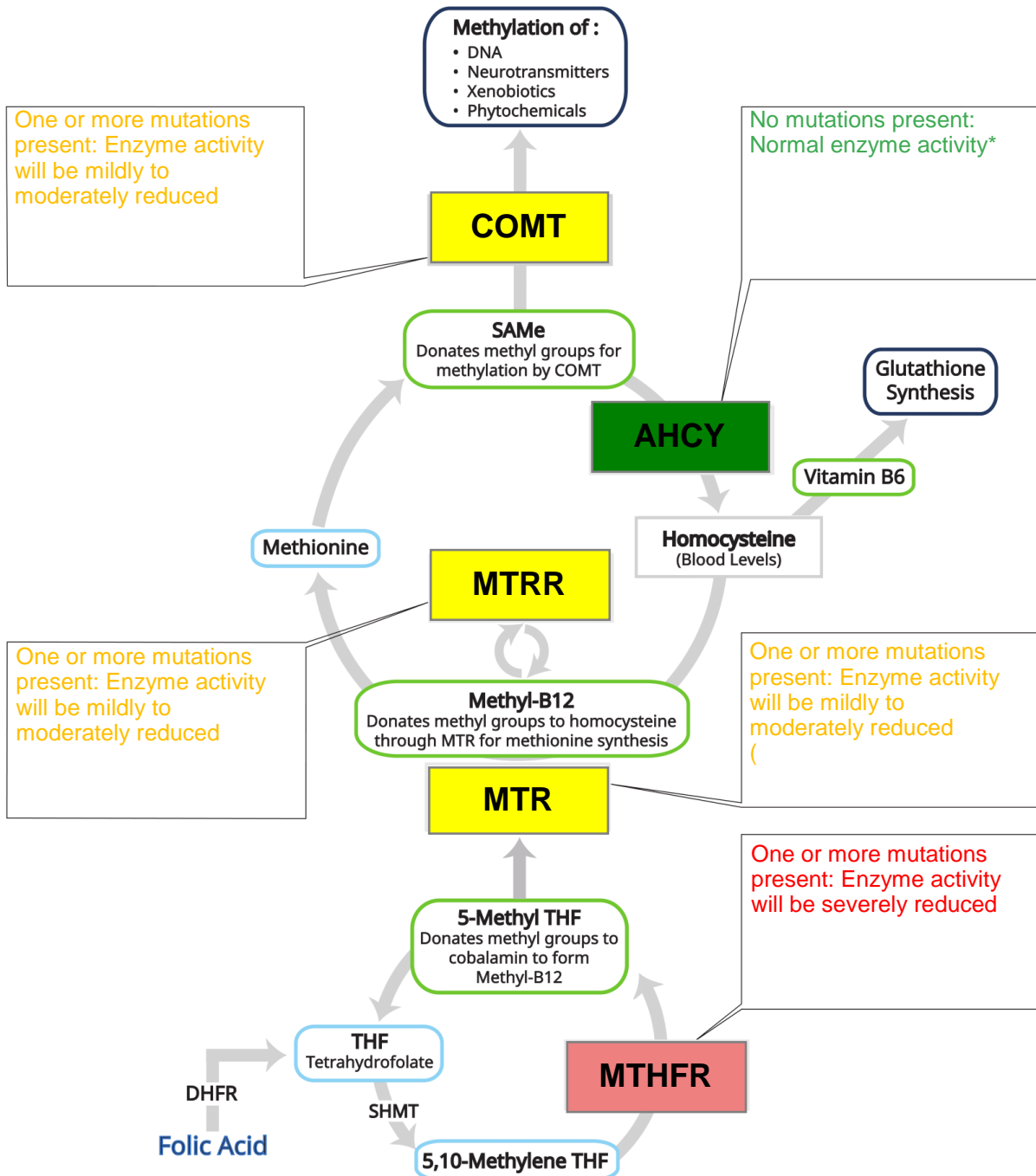


**METHYLATION DETOXIFICATION CYCLE**

**Methylation Detoxification Cycle:**



\* Note that mutations other than those tested may contribute to the decrease in the enzyme activity.



## GENOMIC\_ASSESSMENTS

SWAB, Buccal

Result

Range

Units

### Advanced MethylDetox Profile

#### MTHFR Gene Mutation

MethyleneTetraHydroFolate Reductase (MTHFR) Gene Mutation.

MTHFR Gene Mutation (A1298C): Homozygous for the mutation.

MTHFR Gene Mutation (C677T): Mutation not found.

#### Comment:

The patient has two copies of the MTHFR A1298C mutation.

This is associated with decreased enzyme activity, but no increase in plasma homocysteine levels nor increased risk for venous thrombosis.

MethyleneTetraHydroFolate Reductase (MTHFR) is a regulatory enzyme in folate-dependent homocysteine remethylation.

A common polymorphism in the MTHFR gene at position 677 is associated with a thermolabile enzyme with decreased activity. The prevalence of the homozygous mutation ranges from 8- 18% in various populations.

Clinically, homozygotes for the mutation have an increased risk of thromboembolism as well as premature vascular disease.

A second mutation (A1298C) has also been described. This mutation is associated with an increased risk of thromboembolism, when only found together with the C677T mutation.

#### Important:

1. If individual is being treated with antifolates and homocysteine levels are elevated, supporting literature strongly suggests supplementation with 5-MTHF.

Examples of antifolates include:

Methotrexate (Rheumatrex, Trexall), Pyrimethanine (Daraprim) , Premetrezed (Alimta), Trimethoprim, Proguani.

2. Use caution with individuals previously diagnosed with serotonin syndrome.

Genomic Recommendations: 5-MTHF 0.1mg - 1.0mg

#### MTR Gene Mutation

Methionine Synthase (MTR) Gene Mutation.

MTR Gene Mutation (C3518T): Mutation not found.

MTR Gene Mutation (A2756G): Heterozygous for the mutation.

#### Comment:

The patient is negative for the MTR C3518T mutation and heterozygous for the MTR A2756G mutation.

Enzyme effectiveness tends to be mildly to moderately reduced.

Mild tendency towards elevated homocysteine levels.

Genomic Recommendations: Methyl-B12 1.5mg - 6.0mg

**Sample Test Name**

Date Of Birth: 04 OCT 1972  
Sex: M  
Lab ID: #####

**Test Physician**

Dr. Edward Chan  
11 - 1, Wisma Laxton,  
Jalan Desa, Taman Desa, 58100.

**MTRR Gene Mutation**

Methionine Synthase Reductase (MTRR) Gene Mutation.

MTRR Gene Mutation (A66G): Heterozygous for the mutation.

**Comment:**

The patient is heterozygous for the MTRR A66G mutation.  
Enzyme effectiveness tends to be mildly to moderately reduced.

If in combination with the C677T polymorphism in MTHFR, MTRR genotypes AG (heterozygous) and GG (homozygous positive) influence total plasma homocysteine levels. Additionally, the combination of the genetic polymorphisms in MTRR and MTHFR is linked to an increase in DNA damage as measured by micronucleus frequency (MN). Use caution with individuals previously diagnosed with serotonin syndrome.

Genomic Recommendations: SAME 200mg - 600mg

**AHCY Gene Mutation**

s-Adenosylhomocysteine hydrolase (AHCY) Gene Mutation.

AHCY Gene Mutation (G32878481C): Mutation not found.

AHCY Gene Mutation (C112A): Mutation not found.

AHCY Gene Mutation (G367A): Mutation not found.

**Comment:**

The patient is negative for the AHCY G32878481C Gene Mutation, AHCY C112A Gene Mutation, AHCY G367A Gene Mutation.

Enzyme activity tends to be normal.

Relevant mutations are associated with decreased enzyme presence and/or impaired function leading to the elevated AdoHcy concentrations and impaired methylation potential. Studies show association between mutations resulting in poor methylation potentials, severe myopathies, developmental delays, and hypermethionemia.

**Sample Test Name**

Date Of Birth: 04 OCT 1972  
Sex: M  
Lab ID: #####

**Test Physician**

Dr. Edward Chan  
11 - 1, Wisma Laxton,  
Jalan Desa, Taman Desa, 58100.

**COMT Gene Mutation**

Catechol-O-Methyltransferase (COMT) Gene Mutation.

COMT Gene Mutation (G472A): Heterozygous for the mutation.

COMT Gene Mutation (G304A): Mutation not found.

**Comment:**

The patient is heterozygous for the COMT Gene Mutation G472A and negative for the COMT Gene Mutation G304A mutation.

Enzyme effectiveness tends to be mildly to moderately reduced.

Degradation of the following substances by methylation tends to be poor:

Catechol Estrogens, catechol xenobiotics, dietary phytochemicals, nucleotides, catechol amines (neurotransmitters).

Consumption of COMT inhibitors should be reduced (Green or black tea, quercetin).

Consider use of SAmE, following review of past history for serotonin syndrome.

Genomic Recommendations: SAmE 200mg - 600mg