

# MTHFR

# SPOT ON INFINITI™


*The Automated Multiplexing MDx Solution*



## Product Design

- ▶ The INFINITI™ MTHFR Assay is designed to identify patients with genetic variants of the MTHFR gene.
- ▶ The INFINITI MTHFR Assay utilizes the MTHFR Intellipac™, MTHFR Amp Mix and MTHFR BioFilmChip™ Microarray.
- ▶ The INFINITI MTHFR Assay is automated by the 510(k) cleared INFINITI Analyzer.
- ▶ Clinical Studies to support a 510(k) application are currently in progress.

## Benefits

	VERSATILITY	◆	Simultaneous Multiplexed determination of 2 genetic variants for MTHFR
	EFFICIENCY	◆	Rapid turnaround time enhances workflow efficiency
	AGILITY	◆	<i>Load N Go</i> automation with the INFINITI Analyzer
	INTEGRITY	◆	Replicate determinations on a single BioFilmChip Microarray ensure quality results

## Genetic Variants

MTHFR: A1298C and C677T

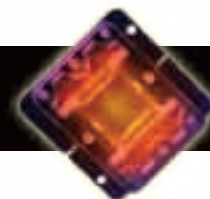
## Sample Type and Volume

0.2 - 2.0 ml of Peripheral whole blood in EDTA (purple-top) tube  
50 ng DNA / reaction

## Product Information

Product No.	Product Name	Description	Pack Size
01 104	INFINITI MTHFR BioFilmChip	12 BioFilmChips/magazine	4 Magazines / pack
01 204	INFINITI MTHFR Intellipac	24 tests/IntelliPac	2 Intellipac / pack
01 304	INFINITI MTHFR Amp Mix	250 ul/vial	4 vials / pack

Please contact AutoGenomics to obtain product information and for product status updates.



## Clinical Relevance<sup>2</sup>

- ▶ Methylenetetrahydrofolate reductase (MTHFR) is a key enzyme in the metabolism of homocysteine.<sup>1</sup>
- ▶ Elevated homocysteine levels have been associated with fetal neural tube defects (NTDs), miscarriages and also indicate an increased risk factor for blood clots, arteriosclerosis and strokes in both men and women.<sup>1</sup>
- ▶ The most common MTHFR mutation, C677T results in a thermolabile enzyme and decreased production of folate, a cofactor required for homocysteine remethylation. This MTHFR mutation is extremely common:<sup>1</sup>
  - 44% of the population have the normal enzyme
  - 44 % are heterozygous for the mutation (i.e. have 1 variant gene). These individuals have some normal enzyme and some of the thermolabile variant of the enzyme.
  - 12 % are homozygous for the mutation (i.e. have 2 variant genes). All of these individuals' enzyme is the thermolabile variant.
- ▶ The presence of a second mutation in the MTHFR gene, A1298C, in conjunction with C677T, has been associated with decreased MTHFR activity and hyperhomocysteinemia. Frequency of the A1298C mutation is reported to be as high as 30% in the general Caucasian population. (Heterozygosity or homozygosity for A1298C alone does not result in hyperhomocysteinemia.)<sup>1</sup>

## Clinical Utility<sup>1</sup>

- ▶ Testing for mutations in MTHFR is useful in identifying a genetic etiology for persistent hyperhomocysteinemia.
  - Also, in individuals known to have other genetic thrombophilic factors (e.g., factor V Leiden), detection of MTHFR mutations signifies a dramatically increased risk for venous thrombosis.<sup>2</sup>

## References

1. Ilan Weisberg, et.al., A Second Genetic Polymorphism in Methylenetetrahydrofolate Reductase (MTHFR) Associated with Decreased Enzyme Activity, *Molecular Genetics and Metabolism*, Volume 64, Issue 3, July 1998, Pages 169-172.
2. Salomon, O., et.al., Single and Combined Prothrombotic Factors in Patients With Idiopathic Venous Thromboembolism, *Arterioscler Thromb Vasc Biol.* 1999;19:511-518.