


**PATIENT**

PFirst PLast  
DOB: 01/01/72

**ORDERING PROVIDER**

Example Organization

**LABORATORY INFORMATION**

Lab ID: N8C9841  
Collection Date: 01/11/10  
Test Date: 01/21/10  
Report Date: 01/22/10

GENE MARKER	BALANCED	TEST RESULT	ENZYME ACTIVITY	COMMENT
CBS-C699T	C/C	C/C	○ Balanced	Balanced Activity
CBS-A360A	C/C	T/T	● Elevated	Possible increase in sulfates and ammonia, possible decreased levels of homocysteine and glutathione
CBS-N212N	C/C	C/C	○ Balanced	Balanced Activity

RISK ALLELE KEY: ○ No Risk ● Risk Allele Heterozygous ● Risk Allele Homozygous

**CBS BACKGROUND INFORMATION**

Cystathionine β-synthase (CBS) is a key enzyme that converts homocysteine to cystathionine, which is then converted to cysteine in a subsequent step.<sup>7</sup> The CBS enzyme also mediates the conversion of homocysteine into the antioxidant glutathione.<sup>6</sup> Certain mutations in the gene that encodes for CBS can cause an increase in the amount of the active enzyme.<sup>1,4,5</sup> When increased levels of the CBS enzyme are present, more of the substrate homocysteine is converted into cystathionine. This conversion process causes an increase in cysteine which creates by-products such as ammonia and sulfur as well as a decrease in glutathione.<sup>1,7</sup> At normal physiological levels, byproducts of the CBS enzyme are beneficial to the overall system.<sup>7-9</sup> When CBS enzyme levels are high, the cell will favor production of taurine over glutathione, and this will result in elevated levels of taurine.<sup>6</sup>

**CBS TREATMENT CONSIDERATIONS**

Supplement	Starting Dosage Range	Comments
B-12 hydroxycobalamin	500-1000 mcg a day	Unlike methylcobalamin, hydroxycobalamin B-12 will not increase the amount of methyl products in the folate pathway. Methylation supports the activity of the CBS enzyme. <sup>1,3</sup>

**Notice:** This information does not take into consideration patient health history, interaction with other medications or supplements, and/or allergies. It is the responsibility of the physician to determine appropriate dosing choices based on all clinical data.



## ADDITIONAL LIFESTYLE INTERVENTIONS

Minimize sulfur-rich foods such as eggs, onion, garlic, and cruciferous vegetables. Because the body may have increased sulfur by-products due to increased activity of the CBS enzyme, these foods may increase the already high amounts of sulfates present.

## BACKGROUND REFERENCES

1. Yasko, A. 2005. Genetic ByPass, Using Nutrition to Bypass Genetic Mutations. Matrix Development Publishing.
2. London, M. CBS Upregulation, Myth or Reality? MIT- Massachusetts Institute of Technology. <http://web.mit.edu/london/www/cbs.html>. [Date accessed May 2017]
3. McEvoy M. March 27 2013. Metabolic Gateways: CBS Mutations & Glutathione. Metabolic Healing Empowering Your Health. <https://metabolichealing.com/metabolic-gateways-cbs-gene-mutations-glutathione/>[Date accessed May 2017]
4. Wang L et al. Modulation of Cystathionine  $\beta$ -Synthase Levels Regulates Total Serum Homocysteine in Mice. American Heart Association Journal. 2004; 94:1318-1324.
5. Aras O et al. Influence of 699C-T and 1080C-T Polymorphisms of the Cystathionine  $\beta$ -synthase gene on Plasma Homocysteine Levels. Clin Gen. 2000; 58:455-459.
6. Yasko, A. 2009. Autism: Pathways To Recovery. Neurological Research Institute, LLC.
7. Stipanuk, M. and I. Ueki. Dealing with methionine/homocysteine sulfur: cysteine metabolism to taurine and inorganic sulfur. J Inheritt Metab Dis. 2011; 34(1):17-32.
8. Rumbelha, W. et al. Acute hydrogen sulfide-induced neuropathology and neurological sequelae: Challenges for translational neuroprotective research. Ann. N.Y. Acad. Sci. 2016; 1378:5-16.
9. Ingenbleek, Y. and H. Kimura., Nutritional essentiality of sulfur in health and disease. Nutrition Reviews. 2013; 71(7):413-432.

*This test detects only specific targeted genetic variations and there is a possibility that other genetic variants not detected by this test may be present. The DNA variants tested for in this report have been scientifically determined to be possible risk factors for the reported condition. The content of this report is provided for informational purposes only, not as a diagnostic tool. The report does not supersede the judgment of a qualified medical provider. This test is not a substitute for a comprehensive consideration of all factors that influence the maintenance of a healthy body. Genetic risk factors are not guarantees that you will develop a condition, and in many cases, the presence of a particular DNA variant may only play a minor role in your risk for disease, compared with environmental and lifestyle factors. This test is not FDA approved. The test's performance characteristics have been established and maintained by Kashi Clinical Laboratories under CLIA and CAP compliance.*

### Reported and Reviewed By:

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