LABORATORY ALLIANCE of Central New York, LLC

Methylmalonic Acid by Tandem Mass Spectrometry

Effective May 24, 2011, Laboratory Alliance of Central New York will begin testing for serum methylmalonic acid levels by liquid chromatography/tandem mass spectrometry (LC/MS/MS). The LC/MS/MS assay was developed by Laboratory Alliance and approved by NY State Department of Health. This assay is based on a published methodology and is performed on a Waters Alliance 2795 HPLC coupled to a Waters Quattro micro tandem mass spectrometer. This assay is highly sensitive and specific.

Clinical Significance:

The measurement of methylmalonic acid (MMA) is carried out to monitor intracellular vitamin B12 (cobalamin) status. Cobalamin is an essential co-factor in two enzymatic reactions: (1) the conversion of homocysteine to methionine; and (2) the conversion of MMA into succinic acid. Therefore a deficiency of cobalamin results in elevated serum concentrations of total homocysteine and methylmalonic acid. Both homocysteine and methylmalonic acid are sensitive markers of intracellular cobalamin status. Serum methylmalonic acid, however, is less influenced by extraneous factors such as age, sex, drugs, folate status, and others than is homocysteine. As a result, serum methylmalonic acid is a more specific marker of cobalamin status than is homocysteine.

Total cobalamin is a poor indicator of bioavailable cobalamin. Approximately 80% of total plasma cobalamin is bound to haptocorrin. This bound form of cobalamin is not taken up by cells. The remaining 20% of total plasma cobalamin is bound to transcobalamin. This bound form of cobalamin is available for cellular uptake and is therefore the bioactive fraction. Methods currently available for the determination of plasma cobalamin concentration measure both bound forms of cobalamin, and therefore may not provide an accurate representation of the intracellular cobalamin status. For this reason the measurement of serum or plasma MMA in addition to cobalamin is superior to the measurement of serum or plasma cobalamin alone in order to assess intracellular cobalamin status.

Cobalamin deficiency is relatively common in the elderly, and has recently been recognized to be a significant problem among neonates and infants. A deficiency of cobalamin can cause subacute combined degeneration of the spinal cord, which is a serious and often irreversible neurologic disorder. Moderately increased MMA is an early indicator of acquired cobalamin deficiency. A massive increase of serum MMA is indicative of methylmalonic acidemia, an inborn metabolic disorder.

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Test Code:	MMAS
Method:	LC/MS/MS
Specimen Requirements:	3.0 mL Serum collected in serum separator tube (1.0 mL minimum).
Unacceptable Conditions:	Plasma
Stability:	14 days refrigerated; 3 months frozen
Schedule of testing:	Monday, Thursday (reported: Tuesday and Friday)
CPT Code:	83921
Billing Code:	1010423
Storage and Transport:	Refrigerated

For More Information:
References are available upon request. For questions or concerns regarding this new test service, please contact Cheryl Haskins, MS, MT(ASCP)SC, Manager of Chemistry and Referral Testing, at 315-410-7014 or cherylhaskins@lacny.com.