MAT2A MUTATION ANALYSIS
JOHN WELSH CARDIOVASCULAR DIAGNOSTIC LABORATORY

MAT2A gene encodes the alpha methionine adenosyltransferase II, which catalyzes the production of Sadenosylmethionine (AdoMet) from methionine and ATP. AdoMet is the key methyl donor in cellular processes. MAT2A gene contains nine exons and spans about 6.3 kb of genomic distance, and has been mapped to chromosome 2p11.2. Liver cancer and glycine N-methyltransferase deficiency have been described to be associated with mutations in the MAT2A gene. It has recently been reported that mutations in this gene are also associated with thoracic aortic aneurysm patients. MAT2A mutations demonstrate autosomal dominant inheritance with a broad range of clinical severity both within and between families. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for MAT2A mutations. Individuals are tested by DNA sequencing of the coding exons of the MAT2A gene. We strongly recommend initial testing of a clearly affected individual, if available, in order to provide the greatest test sensitivity and clearest interpretation of results for subsequent family members. Genetic counseling is recommended for all individuals.

REASONS FOR REFERRAL

Molecular confirmation of the diagnosis of thoracic aortic aneurysm.

METHODOLOGY

Genomic DNA is analyzed for MAT2A mutations by DNA sequencing of the coding exons of the MAT2A gene, as well as the exon/intron junctions and a portion of the 5’ and 3’ untranslated region. Patient DNA is sequenced in both the forward and reverse orientations. If a mutation is identified, additional family members are analyzed only for the familial mutation by automatic fluorescent DNA sequencing.

SERVICE FEES

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<tr>
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<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tr>
<td>Index Case (Male or Female)</td>
<td>$800 per sample</td>
<td>81405</td>
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<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
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SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-9 of MAT2A.

SPECIMEN REQUIREMENTS

Blood (preferred): EDTA (purple-top) tubes: Adult: 5 cc Child: 5 cc Infant: 2-3 cc
Tissue: Frozen (preferred), RNAlater
Other Body Fluids or Formalin-fixed, Paraffin-embedded Tissue: Call to inquire

John Welsh Cardiovascular Diagnostic Laboratory • Section of Cardiology • Department of Pediatrics
Baylor College of Medicine • 1102 Bates Avenue, Suite 480.02 • Houston, TX 77030
PHONE: (832) 824-4155 • FAX: (832) 825-5159 • E-MAIL: yuxinf@bcm.edu
Web Site: www.bcm.edu/pediatrics/welsh