

PreventionGenetics

Disease Prevention through Genetic Testing

A FAX to: _____

FAX Number: _____

Cerebral Cavernous Malformation1 (CCMI) KRITI Mutation Detection by Sequencing.

Cerebral cavernous malformations (CCM) are congenital vascular anomalies of the brain that can cause significant neurological disabilities, including intractable seizures and hemorrhagic stroke. One locus (of 3 identified so far) for autosomal dominant CCM (CCMI) maps to chromosome region 7q21-q22. Using a genomic sequence-based positional cloning strategy, *KRITI*, encoding a protein that interacts with the Krev-1/rap1a tumor suppressor, has been identified as the *CCMI* gene.

A single mutation is common to most Mexican-American families with CCM (+/-70%). This C>T transition in exon 10 changes a *GLN* to a premature termination codon. Other Mexican-American and non-Hispanic Caucasian *CCMI* kindreds harbor other mutations in other exons of the *KRITI* gene. Identification of these other mutations has potential clinical significance for presymptomatic diagnosis of CCM in this population and their families. The identification of a *KRITI* mutation entails extraction of genomic DNA from blood, PCR amplification of 16 separate exons of the *KRITI* gene, sequencing and electrophoretic separation of product, and comparison of this patient specific sequence with "typical" sequence..

SPECIMEN REQUIREMENTS

Collect a minimum of 1 to 10 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube). Whole blood collected in Na Heparin is acceptable but not preferred.

Ship whole blood specimens at ambient temperature. Do not freeze blood tube.

During hot summer months, include a frozen ice pack in the shipping container. Do not allow the ice pack to come in direct contact with the specimen tubes. In Winter, include an unfrozen ice pack to help moderate extremes in temperature. The DNA in whole blood is stable for at least 48 hours at 21°C, 5-7 days at 4°C.

CPT Codes and Cost

CCMI Mutation Test DNA Seq		\$1,410.00
Molec Diag, Ascertainment	83890	
Molec Diag, Isolation	83891	
Molecular Diag, Amplif x16	83898	
Mutat Id By Seq, Single Seg x16	83904	
Molecular Diag, Separation	83894	
Interpretation And Report	83912	

CLIA Certificate of Accreditation Info.

CLIA ID number: **Pending** (LAP#: 7185561 AU ID: 1407125) Effective: **12/28/03**

Applied for by: **Dr. Eric W. Johnson, Director, Molecular Diagnostics and BioBanking**

Ship to:

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