



Molecular Diagnostic Laboratory
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IGHV Hypermutation

Indications for Molecular Testing

Hypermutation status provides prognostic information for patients with chronic lymphocytic leukemia (CLL) and small lymphocytic lymphoma (SLL). Hypermutation (mutated) is defined as $\geq 2\%$ difference from the germline IGHV gene and $<2\%$ difference is considered evidence of no somatic hypermutation (unmutated). There is a clear distinction in the median survival of patients with and without hypermutation. Hypermutation of the IGHV gene is strongly predictive of a better prognosis while lack of mutation predicts a poor prognosis.

Testing Methodology

Polymerase Chain Reaction (PCR) amplification of the conserved framework (FR) and joining (J) regions of the Immunoglobulin Heavy Chain gene followed by Sequencing Analysis. This is a laboratory-developed test performed with consensus primers supplied as analyte specific reagents from InVivoScribe Technologies. (PCR is utilized pursuant to a license agreement with Roche Molecular Systems, Inc.)

Specimen Requirements

Peripheral Blood--1 lavender-top (EDTA) tube. Invert several times to mix blood.

Bone Marrow--Place 1-2 mL of anticoagulated bone marrow in a lavender-top (EDTA) tube. Invert several times to mix bone marrow.

Frozen Tissue--10 mm³ of fresh frozen tissue in sterile, plastic container. Forward frozen tissue on dry ice.

Formalin-Fixed, Paraffin-Embedded (FFPE) Tissue--Twenty 10 micron sections of FFPE tissue in a sterile, microcentrifuge tube.

Do not freeze blood, bone marrow, or FFPE, forward promptly at ambient temperature to the following address:

Molecular Diagnostic Laboratory
Barnes-Jewish Hospital, Institute of Health
Mail Stop 90-28-344
425 South Euclid Avenue, Room 5970
St. Louis, MO 63110

Clinical information must be provided with specimen referral in order to correctly interpret test results.

Current Pricing

Contact Lab Customer Service for current pricing 314 362-1470.

CPT code: 81263

Miller, JE, Wilson, SS, Jaye, DL, and Kronenberg, M. Molecular Diagnostics, 1999;4:101-7.

InVivoScribe Technologies: IGH Gene Clonality Assay, 2004.

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Hamblin TJ, Davis Z, Gardiner A, Oscier DG, Stevenson FK. Unmutated Ig V(H) genes are associated with a more aggressive form of chronic lymphocytic leukemia. Blood 1999;94:1848-54.

Cook GP, Tomlinson IM. The human immunoglobulin VH repertoire. Immunology Today 1995;16:237-42.

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