**MOLECULAR PATHOLOGY/CYTOGENETICS**

**Specimen Collection Link**

**Hours of operation:** 8:00 AM- 4:30 PM, Monday – Friday. A Molecular Path/Cytogenetics Lab Test Requisition (Form S/N 7480) must accompany all samples; testing will not be completed without this form. For any questions during routine working hours, contact the laboratory at extension 58545. For questions after normal hours of operation contact Clinical Lab office (x55700) and ask that Molecular or cytogenetics personnel on call be paged.

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### CYTOGENETICS

<table>
<thead>
<tr>
<th>Test Type</th>
<th>Specimen</th>
<th>Receiving Media</th>
<th>Transport</th>
<th>Turnaround Time</th>
<th>Special Instructions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood Chromosome Analysis: Constitutional</td>
<td>Blood</td>
<td>Green Top 4.0 ml (Sodium heparin)</td>
<td>Transport at room temperature. TAT: 14 days</td>
<td>None</td>
<td></td>
</tr>
<tr>
<td>Blood Chromosome Analysis: Oncology</td>
<td>Blood</td>
<td>Green Top 4.0 ml (Sodium heparin)</td>
<td>Transport at room temperature. TAT: 7-10 days</td>
<td>No recent transfusions.</td>
<td></td>
</tr>
<tr>
<td>Bone Marrow Chromosome Analysis: Oncology</td>
<td>Bone Marrow</td>
<td>Green Top 4.0 ml (Sodium heparin)</td>
<td>Transport at room temperature. TAT: 7-10 days</td>
<td>None</td>
<td></td>
</tr>
<tr>
<td>Tissue Chromosome Analysis: Lymph node, Tumor Solid Tumor</td>
<td>Lymph node Tissue Solid Tumor</td>
<td>Tissue</td>
<td>1 x 1 inch fragment sent in sterile RPMI transport media or Hanks BSS Transport at Room temperature ASAP TAT: 14-21 days</td>
<td>Store at room temperature. Refrigerate if held overnight.</td>
<td></td>
</tr>
<tr>
<td>Tissue Chromosome Analysis: Product of conception</td>
<td>Tissue Tissue Not exposed to formalin</td>
<td>Tissue</td>
<td>1 x 1 inch fragment sent in sterile RPMI transport media or Hanks BSS Transport at Room temperature ASAP TAT: 23-30 days</td>
<td>Store at room temperature. Refrigerate if held overnight.</td>
<td></td>
</tr>
<tr>
<td>Amniotic fluid Chromosome Analysis</td>
<td>Amniotic fluid by transabdominal</td>
<td>15 cc Screw-Top polypropylene centrifuge tube.</td>
<td>Transport at room temperature. TAT: 10-14 days</td>
<td>None</td>
<td></td>
</tr>
</tbody>
</table>
## FLUORESCENCE IN-SITU HYBRIDIZATION (FISH)

<table>
<thead>
<tr>
<th>Fluorescence in situ hybridization (FISH):</th>
<th>Blood</th>
<th>Amniotic fluid</th>
<th>Transport at room temperature. TAT: 2-4 days</th>
<th>NOTE: Helpful in assessing copy number of specific chromosomes present. Most probes available for all chromosomes.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prenatal</td>
<td>Green Top 4.0 ml (Sodium heparin)</td>
<td></td>
<td></td>
<td>None</td>
</tr>
<tr>
<td></td>
<td>Light Lavender: 2 ml. Lavender: 4 ml.</td>
<td>Sterile Screw Top Tube</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Transport at room temperature. TAT: 7 days</td>
<td></td>
<td>CALL laboratory for specific questions regarding probes</td>
<td></td>
</tr>
<tr>
<td>Fluorescence in situ hybridization (FISH):</td>
<td>Blood</td>
<td>Bone Marrow</td>
<td>Light Lavender: 2 ml. Lavender: 4 ml.</td>
<td>None</td>
</tr>
<tr>
<td>Oncology</td>
<td>Green Top 4.0 ml (Sodium heparin)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Transport at room temperature. TAT: 7 days</td>
<td>1 x 1 inch fragment sent in sterile Saline, RPMI, or Hanks BSS</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Fluorescence in situ hybridization (FISH):</td>
<td>Blood</td>
<td>Bone Marrow</td>
<td>Green Top 4.0 ml (Sodium heparin)</td>
<td>None</td>
</tr>
<tr>
<td>Microdeletion Syndromes</td>
<td>Blood</td>
<td>Bone Marrow</td>
<td>Light Lavender: 2 ml. Lavender: 4 ml.</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Amniotic fluid</td>
<td>Sterile Screw Top Tube</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Transport at room temperature. TAT: 7 days</td>
<td></td>
<td>NOTE: Probes for Prader-Willi/ Angelman, DiGeorge / VCFS, Cri-du-Chat, Williams, and other microdeletion syndromes.</td>
<td></td>
</tr>
</tbody>
</table>
## MOLECULAR PATHOLOGY

<table>
<thead>
<tr>
<th>B Cell Immunoglobulin Heavy Chain, Kappa Light Chain, or Lambda light chain gene rearrangement: by PCR</th>
<th>Blood, Bone marrow, or Tissue</th>
<th>Green Top 4.0 ml (Sodium heparin) Light Lavender: 2 ml. Lavender: 4 ml.</th>
<th>Transport blood or bone marrow at room temperature. Keep fresh tissue cold or frozen with dry ice. TAT blood or bone marrow: 7-10 days TAT paraffin tissue: 14-21 days</th>
<th>None</th>
</tr>
</thead>
<tbody>
<tr>
<td>T Cell receptor (Gamma, Beta, or Delta chain rearrangements): by PCR</td>
<td>Blood, Bone marrow, or Tissue</td>
<td>Green Top 4.0 ml (Sodium heparin) Light Lavender: 2 ml. Lavender: 4 ml. 0.5 gms. Fresh tissue in sterile Saline, RPMI Paraffin-embedded material</td>
<td>Transport blood or bone marrow at room temperature. Keep fresh tissue cold or frozen with dry ice. TAT blood or bone marrow: 7-10 days TAT paraffin tissue: 14-21 days</td>
<td>None</td>
</tr>
<tr>
<td><strong>Hypercoag. Panel: Includes Factor II, Factor V, MTHFR</strong></td>
<td><strong>Factor II (prothrombin) Mutation DNA Test:</strong> By PCR</td>
<td>Blood</td>
<td>Light Lavender: 2 ml. Lavender: 4 ml.</td>
<td>Transport at room temperature TAT: 7-10 days</td>
</tr>
<tr>
<td></td>
<td><strong>Factor V (Leiden) Mutation DNA Test:</strong> By PCR</td>
<td>Blood</td>
<td>Light Lavender: 2 ml. Lavender: 4 ml.</td>
<td>Transport at room temperature TAT: 7-10 days</td>
</tr>
<tr>
<td></td>
<td><strong>Methylenetetrahydrotolyl reductase (MTHFR) Mutation DNA Test: By PCR</strong></td>
<td>Blood</td>
<td>Light Lavender: 2 ml. Lavender: 4 ml.</td>
<td>Transport at room temperature TAT: 7-10 days NOTE: Both C677T and A1298C mutations assessed</td>
</tr>
<tr>
<td>Cytogenetic Translocations in Leukemia: e.g., t(9;22) BCR/ABL and t(15;17)PML/RARA</td>
<td>Blood or Bone marrow</td>
<td>Light Lavender: 2 ml. Lavender: 4 ml.</td>
<td>Transport at room temperature TAT: 7-10 days</td>
<td></td>
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</tr>
<tr>
<td>Hereditary Hemochromatosis Mutations (HFE) by PCR</td>
<td>Blood</td>
<td>Light Lavender: 2 ml. Lavender: 4 ml.</td>
<td>Transport at room temperature TAT: 7-10 days NOTE: H63D, C282Y, and S65C mutations assessed</td>
<td></td>
</tr>
<tr>
<td>JAK2 by PCR</td>
<td>Blood or Bone marrow</td>
<td>Green Top 4.0 ml (Sodium heparin) Light Lavender: 2 ml. Lavender: 4 ml.</td>
<td>Transport at room temperature TAT: 7-10 days</td>
<td></td>
</tr>
</tbody>
</table>

**Director Approval:**

| Stephen M. Bonsib, M.D./Director, Department of Pathology | Date |

**Division Approval:**

| Jaiyeola Thomas, M.D./Medical Director, Anatomic Division | Date |
| James Cotelingam, M.D./Medical Director, Clinical Division | Date |