



## Molecular Hematopathology

### Test Menu

**Test/Procedure Name:**

- *BCR-ABL1* (p210) Detection by Real Time PCR using International Standardization (IS)
- Quantitative *BCR-ABL1* p190 Transcript Detection using Real-Time PCR

**Test Includes:** Highly sensitive and accurate detection of *BCR-ABL1* translocations associated with chronic myeloid leukemia (CML) and B-acute lymphoblastic leukemia (B-ALL); monitoring residual leukemia post-therapy.

**Turnaround Time:** 4 business days

**Request Form:** Orderable in CoPath, Epic, Allscripts

**Special Instructions:** None

### Logistics

Send Specimens to:

- Location: Immunopathology, ST-715
- Phone Number: 212-746-2442

Specimen Testing:

- Location: Molecular Hematopathology, K-502
- Phone Number: 212-746-6485

**Availability:** Monday – Friday, 9 AM – 6 PM, except holidays

### Acceptable Specimens:

- $\geq 1$  ml of fresh bone marrow aspirate ( $\geq 2$  ml preferred) or peripheral blood ( $\geq 4$  ml preferred) collected in EDTA (lavender top) tubes
  - Transport ASAP at room temperature for up to 24 hours; store at 2°C – 6°C for up to 48 hours
- $\geq 5$  million cells of mononuclear cells isolated (fresh or frozen)
  - Transport on ice for up to 36 hours
- 1  $\mu$ g of total RNA upon RNA extraction
  - Transport on ice for up to 36 hours and -80°C for storage

**Rejection Causes:** Insufficient specimen, clotted peripheral blood or bone marrow specimens, specimens collected in heparin tubes, formalin-fixed, paraffin-embedded tissues

**Test/Procedure Name:** RT-PCR Detection of *PML-RARA* Translocation, qualitative

**Test Includes:** Detection of the t(15;17) fusion *PML-RARA* with suspected acute promyelocytic leukemia (APL); monitoring residual leukemia after therapy.

**Turnaround Time:** 4 business days (for residual disease); 1 business day (for diagnostic)

**Request Form:** Orderable in CoPath, Epic, Allscripts

**Special Instructions:** None

**Logistics**

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- Phone Number: 212-746-2442

Specimen Testing:

- Location: Molecular Hematopathology, K-502
- Phone Number: 212-746-6485

**Availability:** Monday – Friday, 9 AM – 6 PM, except holidays

**Acceptable Specimens:**

- $\geq 1$  ml of fresh bone marrow aspirate ( $\geq 2$  ml preferred) or peripheral blood ( $\geq 4$  ml preferred) collected in EDTA (lavender top) tubes
  - Transport ASAP at room temperature for up to 24 hours; store at 2°C – 6°C for up to 48 hours
- $\geq 5$  million cells of mononuclear cells isolated (fresh or frozen)
  - Transport on ice for up to 36 hours
- 1  $\mu\text{g}$  of total RNA upon RNA extraction
  - Transport on ice for up to 36 hours and -80°C for storage

**Rejection Causes:** Insufficient specimen, clotted peripheral blood or bone marrow specimens, specimens collected in heparin tubes, formalin-fixed, paraffin-embedded tissues

**Test/Procedure Name:** Quantification of *NPM1* Type A Mutation by Real-Time PCR

**Test Includes:** Quantitative, real-time PCR based assay for the detection of *NPM1* Type A mutation in acute myeloid leukemia (AML) and other myeloid neoplasms

**Turnaround Time:** 4 business days

**Request Form:** Orderable in Epic

**Special Instructions:** For patients with previously known *NPM1* type A mutation

**Logistics**

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- Location: Immunopathology, ST-715
- Phone Number: 212-746-2442

Specimen Testing:

- Location: Molecular Hematopathology, K-502
- Phone Number: 212-746-6485

**Availability:** Monday – Friday, 9 AM – 6 PM, except holidays

**Acceptable Specimens:**

- The patient should be known to previously have had a *NPM1* Type A mutation (*NPM1*, NM\_002520, Exon 11, c.860\_863dupTcTG, p.W288Cfs\*)
- $\geq 1$  ml of fresh bone marrow aspirate ( $\geq 2$  ml preferred) or peripheral blood ( $\geq 4$  ml preferred) collected in EDTA (lavender top) tubes
  - Transport ASAP at room temperature for up to 24 hours; store at 2°C – 6°C for up to 48 hours
- $\geq 5$  million cells of mononuclear cells isolated (fresh or frozen)
  - Transport on ice for up to 36 hours
- 1  $\mu$ g of total RNA upon RNA extraction
  - Transport on ice for up to 36 hours and -80°C for storage

**Rejection Causes:** Insufficient specimen, clotted peripheral blood or bone marrow specimens, specimens collected in heparin tubes, formalin-fixed, paraffin-embedded tissues

**Test/Procedure Name:** *JAK2* Genotyping by ARMS-PCR

**Test Includes:** Qualitative, sensitive ARMS-PCR assay used to detect the *JAK2* V617F mutation

**Turnaround Time:** 4 business days

**Request Form:** Orderable in CoPath, Epic

**Special Instructions:** None

**Logistics**

Send Specimens to:

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- Phone Number: 212-746-2442

Specimen Testing:

- Location: Molecular Hematopathology, K-502
- Phone Number: 212-746-6485

**Availability:** Monday – Friday, 9 AM – 6 PM, except holidays

**Acceptable Specimens:**

- $\geq 1$  ml of fresh bone marrow aspirate or peripheral blood ( $\geq 2$  ml preferred) collected in EDTA (lavender top) tubes
- $\geq 5$  million cells of mononuclear cells isolated (fresh or frozen)
- 1 ug of total DNA upon DNA extraction
- Transport at room temperature for up to 24 hours and store at 2°C – 6°C for up to 48 hours (longer storage compromises integrity of nucleic acids, which may lead to amplification failure)

**Rejection Causes:** Insufficient specimen, formalin-fixed, paraffin-embedded tissues

**Test/Procedure Name:** Rapid PCR Detection of *FLT3* ITD and TKD Mutations

**Test Includes:** Rapid and sensitive detection of *FLT3* ITD and TKD mutations in acute myeloid leukemia (AML) and other myeloid malignancies

**Turnaround Time:** 2 business days

**Request Form:** Orderable in CoPath, Epic

**Special Instructions:** None

**Logistics**

Send Specimens to:

- Location: Immunopathology, ST-715
- Phone Number: 212-746-2442

Specimen Testing:

- Location: Molecular Hematopathology, K-502
- Phone Number: 212-746-6485

**Availability:** Monday – Friday, 9 AM – 6 PM, except holidays

**Acceptable Specimens:**

- $\geq 1$  ml of fresh bone marrow aspirate or peripheral blood ( $\geq 2$  ml preferred) collected in EDTA (lavender top) tubes
- $\geq 5$  million cells of mononuclear cells isolated (fresh or frozen)
- 1 ug of total DNA upon DNA extraction
- Transport at room temperature for up to 24 hours and store at 2°C – 6°C for up to 48 hours (longer storage compromises integrity of nucleic acids, which may lead to amplification failure)

**Rejection Causes:** Insufficient specimen, formalin-fixed, paraffin-embedded tissues

**Test/Procedure Name:** MYD88 L265P Mutation Detection by ddPCR

**Test Includes:** Detection of the MYD88 p.L265P point mutation in samples in order to facilitate the identification and classification of cases with differential diagnosis of lymphoplasmacytic lymphoma, marginal zone lymphoma and B-cell lymphomas with plasmacytic differentiation as well as identify some subtypes of DLBCL.

**Turnaround Time:** 4 business days

**Request Form:** Orderable in CoPath, Epic, Allscripts

**Special Instructions:** None

**Logistics**

Send Specimens to:

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Specimen Testing:

- Location: Molecular Hematopathology, K-502
- Phone Number: 212-746-6485

**Availability:** Monday – Friday, 9 AM – 6 PM, except holidays

**Acceptable Specimens:**

- $\geq 1$  ml of fresh bone marrow aspirate or peripheral blood ( $\geq 2$  ml preferred) collected in EDTA (lavender top) tubes
- $\geq 5$  million cells of mononuclear cells isolated (fresh or frozen)
- 1 ug of total DNA upon DNA extraction
- Formalin-fixed, paraffin-embedded tissues
  - Minimum 2% tumor
  - Section area equivalent to at least 0.5 cm x 0.5 cm
  - Collect 5 to 10 serial sections in a 1.5 ml microcentrifuge tube (10 $\mu$ m thickness)
  - Transport and store at room temperature
- Transport at room temperature for up to 24 hours and store at 2°C – 6°C for up to 48 hours (longer storage compromises integrity of nucleic acids, which may lead to amplification failure)

**Rejection Causes:** Insufficient specimen

**Test/Procedure Name:** *IDH1* and *IDH2* Multiplex Mutation Detection by ddPCR

**Test Includes:** Screen for various *IDH1* and *IDH2* hotspot mutations (codons *IDH1* p.R132, *IDH2* p.R140 and *IDH2* p.R172) in AML patient samples by digital droplet PCR.

**Turnaround Time:** 4 business days

**Request Form:** Orderable in CoPath, Epic, Allscripts

**Special Instructions:** None

**Logistics**

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Specimen Testing:

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- Phone Number: 212-746-6485

**Availability:** Monday – Friday, 9 AM – 6 PM, except holidays

**Acceptable Specimens:**

- Peripheral blood and bone marrow aspirates (as fresh, frozen in cryopreservation media or DMSO, or frozen cell pellets)
  - A minimum of 1 mL of PB or BM aspirate collected in EDTA (lavender top) tubes
  - Transport at room temperature for up to 24 hours and store at 2°C – 6°C for up to 48 hours (longer storage compromises integrity of nucleic acids, which may lead to amplification failure)
- Cells and other fluids
  - A minimum of 10 – 20 million cells are required
  - Can be fresh or frozen (preserved as a pellet or in DMSO-containing frozen media)
  - Fresh: transport on wet ice and store at 2°C – 6°C in media for up to 48 hours
  - Frozen: transport on dry ice and store at -70°C

**Rejection Causes:** Insufficient specimen, formalin-fixed, paraffin-embedded tissues

**Test/Procedure Name:**

- PCR Detection of *IGH* Gene Rearrangement using BIOMED-2 Assay (FR3)
- PCR Detection of *IGK* Gene Rearrangement using BIOMED-2 Assay
- PCR Detection of *TCRG* Gene Rearrangement using BIOMED-2 Assay
- PCR Detection of *BCL2-IGH* Translocation using BIOMED-2 Assay

**Tests Include:** Genomic DNA-based detection of gene rearrangements and reciprocal chromosomal translocations associated with lymphoid neoplasms

**Turnaround Time:** 4 business days

**Request Form:** Orderable in CoPath, Epic, Allscripts

**Special Instructions:** None

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**Availability:** Monday – Friday, 9 AM – 6 PM, except holidays

**Acceptable Specimens:**

- Formalin-fixed, paraffin-embedded tissues
  - Section area equivalent to 0.5 cm x 0.5 cm collected in a 1.5 ml centrifuge tube
  - Transport and store at room temperature
- Frozen sections
  - Transport on dry ice and store at -70°C
- Fresh or frozen tissues
  - Fresh: tissue size equivalent to 0.5 cm<sup>3</sup> kept in cold PBS at 2°C to 6°C for up to 24 hours, transport on wet ice
  - Snap-frozen: at least 250 mg of tissue, transport in dry ice and store at -70°C
  - Mince tissue in ethanol can be transported and stored at room temperature
- Cells and other fluids
  - A minimum of 50,000 – 100,00 cells are required
  - Can be fresh or frozen (preserved as a pellet or in DMSO-containing frozen media)
  - Fresh: transport on wet ice and store at 2°C to 6°C in media
  - Frozen: transport on dry ice and store at -70°C
- Whole blood, bone marrow aspirates (fresh, frozen in cryopreservation media or DMSO, or frozen as dry pellets), bone marrow clot section and smear
  - A minimum of 2 ml of PB or BM aspirate collected in EDTA (lavender top) tubes
  - Transport at room temperature for up to 24 hours and store at 2°C – 6°C for up to 48 hours (longer storage compromises integrity of nucleic acids, which may lead to amplification failure)

**Rejection Causes:** Insufficient specimen



**Test/Procedure Name:**

- PCR Detection of *EBV* DNA
- PCR Detection of *KSHV* DNA

**Tests Include:** Genomic DNA-based detection of EBV and KSHV viruses

**Turnaround Time:** 4 business days

**Request Form:** Orderable in CoPath, Epic, Allscripts

**Special Instructions:** None

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Specimen Testing:

- Location: Molecular Hematopathology, K-502
- Phone Number: 212-746-6485

**Availability:** Monday – Friday, 9 AM – 6 PM, except holidays

**Acceptable Specimens:**

- Formalin-fixed, paraffin-embedded tissues
  - Section area equivalent to 0.5 cm x 0.5 cm collected in a 1.5 ml centrifuge tube
  - Transport and store at room temperature
- Frozen sections
  - Transport on dry ice and store at -70°C
- Fresh or frozen tissues
  - Fresh: tissue size equivalent to 0.5 cm<sup>3</sup> kept in cold PBS at 2°C to 6°C for up to 24 hours, transport on wet ice
  - Snap-frozen: at least 250 mg of tissue, transport in dry ice and store at -70°C
  - Mince tissue in ethanol can be transported and stored at room temperature
- Cells and other fluids
  - A minimum of 50,000 – 100,00 cells are required
  - Can be fresh or frozen (preserved as a pellet or in DMSO-containing frozen media)
  - Fresh: transport on wet ice and store at 2°C to 6°C in media
  - Frozen: transport on dry ice and store at -70°C
- Whole blood, bone marrow aspirates (fresh, frozen in cryopreservation media or DMSO, or frozen as dry pellets), bone marrow clot section and smear
  - A minimum of 2 ml of PB or BM aspirate collected in EDTA (lavender top) tubes
  - Transport at room temperature for up to 24 hours and store at 2°C – 6°C for up to 48 hours (longer storage compromises integrity of nucleic acids, which may lead to amplification failure)

**Rejection Causes:** Insufficient specimen

**Test/Procedure Name:** *CSF3R* Exon 14 and Exon 17 Mutation Testing

**Test Includes:** Detection of *CSF3R* mutations in myeloid malignancies

**Turnaround Time:** 4 business days

**Request Form:** Orderable in CoPath, Epic

**Special Instructions:** None

**Logistics**

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- Location: Molecular Hematopathology, K-502
- Phone Number: 212-746-6485

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**Acceptable Specimens:**

- $\geq 1$  ml of fresh bone marrow aspirate or peripheral blood ( $\geq 2$  ml preferred) collected in EDTA (lavender top) tubes
- $\geq 5$  million cells of mononuclear cells isolated (fresh or frozen)
- 1 ug of total DNA upon DNA extraction
- Transport at room temperature for up to 24 hours and store at 2°C – 6°C for up to 48 hours (longer storage compromises integrity of nucleic acids, which may lead to amplification failure)

**Rejection Causes:** Insufficient specimen, formalin-fixed, paraffin-embedded tissues