Molecular Hematopathology

Test Menu

Test/Procedure Name: Heme Fusion NGS Panel – Myeloid

Test Includes: Targeted RNA-based NGS assay for the detection of gene fusions involving 73 genes known to be recurrently altered in a variety of hematologic neoplasms, including acute myeloid leukemia and other myeloid disorders.

Turnaround Time: 7 to 10 business days

Request Form: Orderable in Copath, Epic, Allscripts

Special Instructions: Fresh samples should be delivered promptly to the Immunopathology/Molecular Hematopathology lab and stored in 4°C. Frozen cell isolates/pellets should remain frozen (in dry ice) during transport and delivery.

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:
- Fresh peripheral blood and bone marrow aspirates
  - >4 ml peripheral blood collected in EDTA (lavender top) tube
  - >2 ml bone marrow aspirate collected in EDTA (lavender top) tube
  - Transport ASAP at room temperature and store at 2°C – 6°C for up to 48 hours
- Fresh or frozen cell isolates/pellets from peripheral blood or bone marrow aspirates
  - >5 – 10 million cells of mononuclear cells isolated (fresh or frozen)
  - Transport fresh cells on wet ice and store at 2°C – 6°C for up to 24 hours
  - Transport frozen cells on dry ice and store at -80°C
- At least 1 µg of RNA
  - Transport on ice for up to 36 hours and store at -80°C

Rejection Causes: Insufficient specimen, clotted peripheral blood or bone marrow specimens, specimens collected in heparin tubes, formalin-fixed, paraffin-embedded tissues, fixed flow cells, leukopheresis products
Molecular Hematopathology Test Menu

Test/Procedure Name: Detection of Minimal Residual Diseases in B cell malignancies using IGH NGS Assay

Test Includes: Clonality detection in patients with lymphoid malignant diseases, including acute lymphoblastic leukemia (B-ALL), chronic lymphocytic leukemia (CLL) and mantel cell lymphoma (MCL); track tumor loads by combining PCR and NGS-based deep sequencing technologies

Turnaround Time: 10 to 14 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:
- Fresh peripheral blood and bone marrow aspirates
  - 10 ml peripheral blood collected in EDTA (lavender top) tubes
  - >2 ml bone marrow aspirate 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
- At least 2 µg of DNA
  - Transport on ice for up to 36 hours and store at 2°C – 6°C for up to 48 hours

Rejection Causes: Insufficient specimen, clotted peripheral blood or bone marrow specimens, specimens collected in heparin tubes, formalin-fixed, paraffin-embedded tissues, fixed flow cells, leukopheresis products
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: Fresh samples should be delivered promptly to the Immunopathology/Molecular Hematopathology lab and stored in 4°C.

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:
- Fresh peripheral blood and bone marrow aspirates
  - >1 ml bone marrow aspirate collected in EDTA (lavender top) tubes
  - >4 ml peripheral blood 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
- Fresh or frozen cell isolates/pellets from peripheral blood or bone marrow aspirates
  - >5 million cells of mononuclear cells isolated
  - Transport on ice for up to 36 hours
- 1 µg 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
Molecular Hematopathology Test Menu

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: Fresh samples should be delivered promptly to the Immunopathology/Molecular Hematopathology lab and stored in 4°C.

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:
- Fresh peripheral blood and bone marrow aspirates
  - >1 ml bone marrow aspirate collected in EDTA (lavender top) tubes
  - >4 ml peripheral blood 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

- Fresh or frozen cell isolates/pellets from peripheral blood or bone marrow aspirates
  - >5 million cells of mononuclear cells isolated
  - Transport on ice for up to 36 hours

- 1 µg 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
Molecular Hematopathology Test Menu

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. Fresh samples should be delivered promptly to the Immunopathology/Molecular Hematopathology lab and stored in 4°C.

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:
- The patient should be known to previously have had a $NPM1$ Type A mutation ($NPM1$, NM_002520, Exon 11, c.860_863dupTcTG, p.W288Cfs*)
- Fresh peripheral blood and bone marrow aspirates
  - >1 ml bone marrow aspirate collected in EDTA (lavender top) tubes
  - >4 ml peripheral blood 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
- Fresh or frozen cell isolates/pellets from peripheral blood or bone marrow aspirates
  - >5 million cells of mononuclear cells isolated
  - Transport on ice for up to 36 hours
- 1 µg 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
Molecular Hematopathology Test Menu

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:
- 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:
- >1 ml of fresh bone marrow aspirate or peripheral blood (>2 ml preferred) 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
- >5 million cells of mononuclear cells isolated (fresh or frozen)
- 1 ug of total DNA upon DNA extraction
- Transport on ice for up to 36 hours and store at 2°C – 6°C for up to 48 hours

Rejection Causes: Insufficient specimen, formalin-fixed, paraffin-embedded tissues
Molecular Hematopathology Test Menu

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:
• >1 ml of fresh bone marrow aspirate or peripheral blood (>2 ml preferred) collected in EDTA (lavender top) tubes
  o Transport at room temperature for up to 24 hours and store at 2°C – 6°C for up to 48 hours
• >5 million cells of mononuclear cells isolated (fresh or frozen)
• 1 ug of total DNA upon DNA extraction
• Transport on ice for up to 36 hours and store at 2°C – 6°C for up to 48 hours

Rejection Causes: Insufficient specimen, formalin-fixed, paraffin-embedded tissues
Molecular Hematopathology Test Menu

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:
- 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:**
- ≥1 ml of fresh bone marrow aspirate or peripheral blood (≥2 ml preferred) 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
- ≥5 million cells of mononuclear cells isolated (fresh or frozen)
- 1 ug of total DNA upon DNA extraction
- Transport on ice for up to 36 hours and store at 2°C – 6°C for up to 48 hours
- 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 (5-10µm thickness)
  - Transport and store at room temperature

**Rejection Causes:** Insufficient specimen
Molecular Hematopathology Test Menu

**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**

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:**
- 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 36 hours and store at 2°C – 6°C for up to 48 hours
- 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
- 1 ug of total DNA upon DNA extraction
  - Transport on ice for up to 36 hours and store at 2°C – 6°C for up to 48 hours

**Rejection Causes:** Insufficient specimen, formalin-fixed, paraffin-embedded tissues
Molecular Hematopathology Test Menu

Test/Procedure Name:
- PCR Detection of \textit{IGH} Gene Rearrangement using BIOMED-2 Assay (FR3)
- PCR Detection of \textit{IGK} Gene Rearrangement using BIOMED-2 Assay
- PCR Detection of \textit{TCRG} Gene Rearrangement using TCRG version 2 Assay
- PCR Detection of \textit{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

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:
- Formalin-fixed, paraffin-embedded tissues
  - Section area equivalent to 0.5 cm x 0.5 cm
  - Collect 5-10 sections in a 1.5 ml centrifuge tube (10 \( \mu \text{m} \) thickness), or
  - Minimum 1 glass slide with 0.5 cm x 0.5 cm section area (unstained or H&E stained, preferably without coverslip, 10 \( \mu \text{m} \) thickness)
  - Transport and store at room temperature
- Frozen sections
  - Collect 5-10 sections in 1 15 ml tube (20 \( \mu \text{m} \) thickness)
  - Transport on dry ice and store at -70°C
- Fresh or frozen tissues
  - Fresh: tissue size equivalent to 0.5 cm\(^3\) 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 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)

Rejection Causes: Insufficient specimen
Molecular Hematopathology Test Menu

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

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:
- Formalin-fixed, paraffin-embedded tissues
  - Section area equivalent to 0.5 cm x 0.5 cm
  - Collect 5-10 sections in a 1.5 ml centrifuge tube (10 µm thickness), or
  - Minimum 1 glass slide with 0.5 cm x 0.5 cm section area (unstained or H&E stained, preferably without coverslip, 10 µm thickness)
  - Transport and store at room temperature
- Frozen sections
  - Collect 5-10 sections in 1 15 ml tube (20 µm thickness)
  - Transport on dry ice and store at -70°C
- Fresh or frozen tissues
  - Fresh: tissue size equivalent to 0.5 cm³ 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 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)

Rejection Causes: Insufficient specimen
Molecular Hematopathology Test Menu

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
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:
- >1 ml of fresh bone marrow aspirate or peripheral blood (>2 ml preferred) collected in EDTA (lavender top) tubes
- >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