Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	T-ALL
Molecular Abnormality:	Comprehensive flow cytometric immunophenotyping to include B, T and myeloid lineage markers
Test:	Comprehensive flow cytometric immunophenotyping to include B, T and myeloid lineage markers
Chromosome:	
Gene Symbol:	
Test Detects:	Protein expression
Methodology:	Flow cytometry
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	The initial immunophenotyping panel should be sufficiently comprehensive to establish a leukemia associated phenotype (LAP) that may include expression of non-lineage antigens. These LAP are useful in classification, particularly mixed lineage leukemias, and as a signature for minimal residual disease (MRD) detection.
Test Purpose:	Classification, Diagnostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5, ALL-A Page:12
Notes:	Typical Immunophenotype, T-ALL: TdT+. variable for all of the following: CD1a, CD2, CD3, CD4, CD5, CD7, CD8, CD34. •Pro-T-ALL: cCD3+, CD7+, CD1a-, CD2-, CD4-, CD8-, CD34+/-. Pre-T-ALL: cCD3+, CD7+, CD1a-, CD2+, CD4-, CD8-, CD34+/-. •Cortical T-ALL: cCD3+, CD7+, CD1a+, CD2+, CD4+, CD8+, CD34-. •Medullary T-ALL: cCD3+, sCD3+, CD7+, CD1a-, CD2+, CD4+ or CD8+, CD34- •ETP T-ALL: Lack of CD1a and CD8 expression, weak CD5 expression with less than 75% positive blasts, and expression of one or more of the following myeloid or stem cell markers on at least 25% of lymphoblasts: CD117, CD34, HLA-DR, CD13, CD33, CD11b, and/or CD65.

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 1 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL
Molecular Abnormality:	Comprehensive flow cytometric immunophenotyping to include B, T and myeloid lineage markers
Test:	Comprehensive flow cytometric immunophenotyping to include B, T and myeloid lineage markers
Chromosome:	
Gene Symbol:	
Test Detects:	Protein expression
Methodology:	Flow cytometry
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	The initial immunophenotyping panel should be sufficiently comprehensive to establish a leukemia associated phenotype (LAP) that may include expression of non-lineage antigens. These LAP are useful in classification, particularly mixed lineage leukemias, and as a signature for minimal residual disease (MRD) detection.
Test Purpose:	Classification, Diagnostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5, ALL-A Page:12
Notes:	Typical Immunophenotype B-ALL, not otherwise specified: CD10+, CD19+, CD79a+, cCD22+, sCD22+, CD24+, PAX5+, TdT+, variable CD20, variable CD34. • Early precursor B-ALL (pro-B-ALL): CD10-, CD19+, cCD79a+, cCD22+, TdT+. • Common B-ALL: CD10+. pre-B-ALL: cytoplasmic mu+, sIg-, CD10+/-. • Precursor B-ALL (pre-B-ALL): cytoplasmic mu+, sIg-, CD10+/-.

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 2 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL with recurrent genetic abnormalities
Molecular Abnormality:	Comprehensive flow cytometric immunophenotyping to include B, T and myeloid lineage markers
Test:	Comprehensive flow cytometric immunophenotyping to include B, T and myeloid lineage markers
Chromosome:	
Gene Symbol:	
Test Detects:	Protein expression
Methodology:	Flow cytometry
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	The initial immunophenotyping panel should be sufficiently comprehensive to establish a leukemia associated phenotype (LAP) that may include expression of non-lineage antigens. These LAP are useful in classification, particularly mixed lineage leukemias, and as a signature for minimal residual disease (MRD) detection.
Test Purpose:	Classification, Diagnostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5, ALL-A Page:12
Notes:	Typical Immunophenotype, B-ALL with recurrent genetic abnormalities: • Hyperdiploidy (DNA index >1.16; 51065 chromosome without structural abnormalities): CD10+, CD19+, CD34+, CD45- • Hypodiploidy (• t(9;22)(q34;q11.2); BCR-ABL1: CD10+, CD19+, TdT+, CD13+, CD33+, CD117-. • t(v;11q23); MLL rearranged: CD10-, CD19+, CD24-, CD15+ • t(12;21)(p13;q22); TEL-AML1: CD10+, CD19+, TdT+, CD13+, CD34+ • t(1;19)(q23;p13.3); E2A-PBX1: CD10+, CD19+, CD20 variable, CD34+/-, cytoplasmic mu+ • t(5;14)(q31;q32); IL-3-IGH: CD10+, CD19+

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 5 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL with recurrent genetic abnormalities
Molecular Abnormality:	(1;19)(q23;p13.3) translocation
Test:	(1;19)(q23;p13.3) E2A-PBX1 translocation
Chromosome:	t(1;19)(q23;p13.3)
Gene Symbol:	TCF3-PBX1
Test Detects:	Translocation
Methodology:	Cytogenetics, FISH, RT-PCR
NCCN Category of Evidence:	2A
Specimen Types:	Bone marrow lymphoblasts, Peripheral blood lymphoblasts
NCCN Recommendation - Clinical Decision:	Diagnosis. Genetic Characterization. Optimal risk stratification and treatment planning requires testing marrow or peripheral blood lymphoblasts for specific recurrent genetic abnormalities using: • Karyotyping of G-banded metaphase chromosomes (cytogenetics); • Interphase fluorescence in situ hybridization (FISH) testing including probes capable of detecting the major recurrent genetic abnormalities; • Reverse transcriptase polymerase chain reaction (RT-PCR) testing for fusion genes (eg, BCR-ABL). Additional optional tests include: • Flow cytometric DNA index/ploidy testing (additional assessment for hyperdiploidy and hypodiploidy). Classification: Together these studies allow determination of the World Health Organization (WHO) ALL subtype and cytogenetic risk group.
Test Purpose:	Classification, Diagnostic, Prognostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5
Notes:	Subtypes: B-cell lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities include hyperdiploidy, hypodiploidy, and commonly occurring translocations: t(9;22)(q34;q11.2)[BCR-ABL1];t(v;11q23)[MLL rearranged]; t(12;21)(p13;q22)[TEL-AML1]; t(1;19)(q23;p13.3)[E2A-PBX1]; t(5;14)(q31;q32)[IL3-IGH; relatively rare]. Cytogenetic risk groups are defined as follows: Good risk: Hyperdiploidy (51-65 chromosomes and/or DNA index > 1.16; cases with trisomy of chromosomes 4,10, and 17 appear to have the most favorable outcome); t(12;21)(p13;q22):TEL-AML1. Poor risk: Hypodiploidy (<44 chromosomes and/or DNA index <0.81); t(v;11q23: MLL rearranged; t(9;22)(q34;q11.2): BCR-ABL1 (defined as high risk in the pre-TKI era); Complex karyotype (5 or more chromosomal abnormalities).

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 10 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL with recurrent genetic abnormalities
Molecular Abnormality:	(12;21)(p13;q22) translocation
Test:	(12;21)(p13;q22) TEL-AML1 translocation
Chromosome:	t(12;21)(p13;q22)
Gene Symbol:	ETV6-RUNX1
Test Detects:	Translocation
Methodology:	Cytogenetics, FISH, RT-PCR
NCCN Category of Evidence:	2A
Specimen Types:	Bone marrow lymphoblasts, Peripheral blood lymphoblasts
NCCN Recommendation - Clinical Decision:	Diagnosis. Genetic Characterization. Optimal risk stratification and treatment planning requires testing marrow or peripheral blood lymphoblasts for specific recurrent genetic abnormalities using: • Karyotyping of G-banded metaphase chromosomes (cytogenetics); • Interphase fluorescence in situ hybridization (FISH) testing including probes capable of detecting the major recurrent genetic abnormalities; • Reverse transcriptase polymerase chain reaction (RT-PCR) testing for fusion genes (eg, BCR-ABL). Additional optional tests include: • Flow cytometric DNA index/ploidy testing (additional assessment for hyperdiploidy and hypodiploidy). Classification: Together these studies allow determination of the World Health Organization (WHO) ALL subtype and cytogenetic risk group.
Test Purpose:	Classification, Diagnostic, Prognostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5
Notes:	Subtypes: B-cell lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities include hyperdiploidy, hypodiploidy, and commonly occurring translocations: t(9;22)(q34;q11.2)[BCR-ABL1];t(v;11q23)[MLL rearranged]; t(12;21)(p13;q22)[TEL-AML1]; t(1;19)(q23;p13.3)[E2A-PBX1]; t(5;14)(q31;q32)[IL3-IGH; relatively rare]. Cytogenetic risk groups are defined as follows: Good risk: Hyperdiploidy (51-65 chromosomes and/or DNA index > 1.16; cases with trisomy of chromosomes 4,10, and 17 appear to have the most favorable outcome); t(12;21)(p13;q22):TEL-AML1. Poor risk: Hypodiploidy (<44 chromosomes and/or DNA index <0.81); t(v;11q23: MLL rearranged; t(9;22)(q34;q11.2): BCR-ABL1 (defined as high risk in the pre-TKI era); Complex karyotype (5 or more chromosomal abnormalities).

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 11 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL with recurrent genetic abnormalities
Molecular Abnormality:	(5;14)(q31;q32) translocation
Test:	(5;14)(q31;q32) IL3-IGH translocation
Chromosome:	t(5;14)(q31;q32)
Gene Symbol:	IL3-IGH
Test Detects:	Translocation
Methodology:	Cytogenetics, FISH, RT-PCR
NCCN Category of Evidence:	2A
Specimen Types:	Bone marrow lymphoblasts, Peripheral blood lymphoblasts
NCCN Recommendation - Clinical Decision:	Diagnosis. Genetic Characterization. Optimal risk stratification and treatment planning requires testing marrow or peripheral blood lymphoblasts for specific recurrent genetic abnormalities using: • Karyotyping of G-banded metaphase chromosomes (cytogenetics); • Interphase fluorescence in situ hybridization (FISH) testing including probes capable of detecting the major recurrent genetic abnormalities; • Reverse transcriptase polymerase chain reaction (RT-PCR) testing for fusion genes (eg, BCR-ABL). Additional optional tests include: • Flow cytometric DNA index/ploidy testing (additional assessment for hyperdiploidy and hypodiploidy). Classification: Together these studies allow determination of the World Health Organization (WHO) ALL subtype and cytogenetic risk group.
Test Purpose:	Classification, Diagnostic, Prognostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5
Notes:	Subtypes: B-cell lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities include hyperdiploidy, hypodiploidy, and commonly occurring translocations: t(9;22)(q34;q11.2)[BCR-ABL1];t(v;11q23)[MLL rearranged]; t(12;21)(p13;q22)[TEL-AML1]; t(1;19)(q23;p13.3)[E2A-PBX1]; t(5;14)(q31;q32)[IL3-IGH; relatively rare]. Cytogenetic risk groups are defined as follows: Good risk: Hyperdiploidy (51-65 chromosomes and/or DNA index > 1.16; cases with trisomy of chromosomes 4,10, and 17 appear to have the most favorable outcome); t(12;21)(p13;q22):TEL-AML1. Poor risk: Hypodiploidy (<44 chromosomes and/or DNA index <0.81); t(v;11q23: MLL rearranged; t(9;22)(q34;q11.2): BCR-ABL1 (defined as high risk in the pre-TKI era); Complex karyotype (5 or more chromosomal abnormalities).

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 12 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL with recurrent genetic abnormalities
Molecular Abnormality:	(9;22)(q34;q11.2) translocation
Test:	(9;22)(q34;q11.2) BCR-ABL1 translocation
Chromosome:	t(9;22)(q34;q11.2)
Gene Symbol:	BCR-ABL1
Test Detects:	Translocation
Methodology:	Cytogenetics, FISH, RT-PCR
NCCN Category of Evidence:	2A
Specimen Types:	Bone marrow lymphoblasts, Peripheral blood lymphoblasts
NCCN Recommendation - Clinical Decision:	Diagnosis. Genetic Characterization. Optimal risk stratification and treatment planning requires testing marrow or peripheral blood lymphoblasts for specific recurrent genetic abnormalities using: • Karyotyping of G-banded metaphase chromosomes (cytogenetics); • Interphase fluorescence in situ hybridization (FISH) testing including probes capable of detecting the major recurrent genetic abnormalities; • Reverse transcriptase polymerase chain reaction (RT-PCR) testing for fusion genes (eg, BCR-ABL). Additional optional tests include: • Flow cytometric DNA index/ploidy testing (additional assessment for hyperdiploidy and hypodiploidy). Classification: Together these studies allow determination of the World Health Organization (WHO) ALL subtype and cytogenetic risk group.
Test Purpose:	Classification, Diagnostic, Prognostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5
Notes:	Subtypes: B-cell lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities include hyperdiploidy, hypodiploidy, and commonly occurring translocations: t(9;22)(q34;q11.2)[BCR-ABL1];t(v;11q23)[MLL rearranged]; t(12;21)(p13;q22)[TEL-AML1]; t(1;19)(q23;p13.3)[E2A-PBX1]; t(5;14)(q31;q32)[IL3-IGH; relatively rare]. Cytogenetic risk groups are defined as follows: Good risk: Hyperdiploidy (51-65 chromosomes and/or DNA index > 1.16; cases with trisomy of chromosomes 4,10, and 17 appear to have the most favorable outcome); t(12;21)(p13;q22):TEL-AML1. Poor risk: Hypodiploidy (<44 chromosomes and/or DNA index <0.81); t(v;11q23: MLL rearranged; t(9;22)(q34;q11.2): BCR-ABL1 (defined as high risk in the pre-TKI era); Complex karyotype (5 or more chromosomal abnormalities).

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 13 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL with recurrent genetic abnormalities
Molecular Abnormality:	(v;11q23) translocation
Test:	(v;11q23) MLL rearranged
Chromosome:	t(v;11q23)
Gene Symbol:	MLL
Test Detects:	Translocation
Methodology:	Cytogenetics, FISH, RT-PCR
NCCN Category of Evidence:	2A
Specimen Types:	Bone marrow lymphoblasts, Peripheral blood lymphoblasts
NCCN Recommendation - Clinical Decision:	Diagnosis. Genetic Characterization. Optimal risk stratification and treatment planning requires testing marrow or peripheral blood lymphoblasts for specific recurrent genetic abnormalities using: • Karyotyping of G-banded metaphase chromosomes (cytogenetics); • Interphase fluorescence in situ hybridization (FISH) testing including probes capable of detecting the major recurrent genetic abnormalities; • Reverse transcriptase polymerase chain reaction (RT-PCR) testing for fusion genes (eg, BCR-ABL). Additional optional tests include: • Flow cytometric DNA index/ploidy testing (additional assessment for hyperdiploidy and hypodiploidy). Classification: Together these studies allow determination of the World Health Organization (WHO) ALL subtype and cytogenetic risk group.
Test Purpose:	Classification, Diagnostic, Prognostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5
Notes:	Subtypes: B-cell lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities include hyperdiploidy, hypodiploidy, and commonly occurring translocations: t(9;22)(q34;q11.2)[BCR-ABL1];t(v;11q23)[MLL rearranged]; t(12;21)(p13;q22)[TEL-AML1]; t(1;19)(q23;p13.3)[E2A-PBX1]; t(5;14)(q31;q32)[IL3-IGH; relatively rare]. Cytogenetic risk groups are defined as follows: Good risk: Hyperdiploidy (51-65 chromosomes and/or DNA index > 1.16; cases with trisomy of chromosomes 4,10, and 17 appear to have the most favorable outcome); t(12;21)(p13;q22):TEL-AML1. Poor risk: Hypodiploidy (<44 chromosomes and/or DNA index <0.81); t(v;11q23: MLL rearranged; t(9;22)(q34;q11.2): BCR-ABL1 (defined as high risk in the pre-TKI era); Complex karyotype (5 or more chromosomal abnormalities).

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 14 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL with recurrent genetic abnormalities
Molecular Abnormality:	Flow cytometric DNA cell cycle analysis
Test:	Flow cytometric DNA cell cycle analysis
Chromosome:	
Gene Symbol:	
Test Detects:	Hyperdiploidy, Hypodiploidy
Methodology:	Flow cytometry
NCCN Category of Evidence:	2A
Specimen Types:	Bone marrow lymphoblasts, Peripheral blood lymphoblasts
NCCN Recommendation - Clinical Decision:	Diagnosis. Genetic Characterization. Optimal risk stratification and treatment planning requires testing marrow or peripheral blood lymphoblasts for specific recurrent genetic abnormalities using: • Karyotyping of G-banded metaphase chromosomes (cytogenetics); • Interphase fluorescence in situ hybridization (FISH) testing including probes capable of detecting the major recurrent genetic abnormalities; • Reverse transcriptase polymerase chain reaction (RT-PCR) testing for fusion genes (eg, BCR-ABL). Additional optional tests include: • Flow cytometric DNA index/ploidy testing (additional assessment for hyperdiploidy and hypodiploidy). Classification: Together these studies allow determination of the World Health Organization (WHO) ALL subtype and cytogenetic risk group.
Test Purpose:	Classification, Diagnostic, Prognostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5
Notes:	Subtypes: B-cell lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities include hyperdiploidy, hypodiploidy, and commonly occurring translocations: t(9;22)(q34;q11.2)[BCR-ABL1];t(v;11q23)[MLL rearranged]; t(12;21)(p13;q22)[TEL-AML1]; t(1;19)(q23;p13.3)[E2A-PBX1]; t(5;14)(q31;q32)[IL3-IGH; relatively rare]. Cytogenetic risk groups are defined as follows: Good risk: Hyperdiploidy (51-65 chromosomes and/or DNA index > 1.16; cases with trisomy of chromosomes 4,10, and 17 appear to have the most favorable outcome); t(12;21)(p13;q22):TEL-AML1. Poor risk: Hypodiploidy (<44 chromosomes and/or DNA index <0.81); t(v;11q23: MLL rearranged; t(9;22)(q34;q11.2): BCR-ABL1 (defined as high risk in the pre-TKI era); Complex karyotype (5 or more chromosomal abnormalities).

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 17 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL with recurrent genetic abnormalities
Molecular Abnormality:	Hyperdiploidy
Test:	Hyperdiploidy
Chromosome:	
Gene Symbol:	
Test Detects:	Hyperdiploidy
Methodology:	Cytogenetics, FISH
NCCN Category of Evidence:	2A
Specimen Types:	Bone marrow lymphoblasts, Peripheral blood lymphoblasts
NCCN Recommendation - Clinical Decision:	Diagnosis. Genetic Characterization. Optimal risk stratification and treatment planning requires testing marrow or peripheral blood lymphoblasts for specific recurrent genetic abnormalities using: • Karyotyping of G-banded metaphase chromosomes (cytogenetics); • Interphase fluorescence in situ hybridization (FISH) testing including probes capable of detecting the major recurrent genetic abnormalities; • Reverse transcriptase polymerase chain reaction (RT-PCR) testing for fusion genes (eg, BCR-ABL). Additional optional tests include: • Flow cytometric DNA index/ploidy testing (additional assessment for hyperdiploidy and hypodiploidy). Classification: Together these studies allow determination of the World Health Organization (WHO) ALL subtype and cytogenetic risk group.
Test Purpose:	Classification, Diagnostic, Prognostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5
Notes:	Subtypes: B-cell lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities include hyperdiploidy, hypodiploidy, and commonly occurring translocations: t(9;22)(q34;q11.2)[BCR-ABL1];t(v;11q23)[MLL rearranged]; t(12;21)(p13;q22)[TEL-AML1]; t(1;19)(q23;p13.3)[E2A-PBX1]; t(5;14)(q31;q32)[IL3-IGH; relatively rare]. Cytogenetic risk groups are defined as follows: Good risk: Hyperdiploidy (51-65 chromosomes and/or DNA index > 1.16; cases with trisomy of chromosomes 4,10, and 17 appear to have the most favorable outcome); t(12;21)(p13;q22):TEL-AML1. Poor risk: Hypodiploidy (<44 chromosomes and/or DNA index <0.81); t(v;11q23: MLL rearranged; t(9;22)(q34;q11.2): BCR-ABL1 (defined as high risk in the pre-TKI era); Complex karyotype (5 or more chromosomal abnormalities).

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 19 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	B-ALL with recurrent genetic abnormalities
Molecular Abnormality:	Hyperdiploidy
Test:	Hyperdiploidy
Chromosome:	
Gene Symbol:	
Test Detects:	Hypodiploidy
Methodology:	Cytogenetics, FISH
NCCN Category of Evidence:	2A
Specimen Types:	Bone marrow lymphoblasts, Peripheral blood lymphoblasts
NCCN Recommendation - Clinical Decision:	Diagnosis. Genetic Characterization. Optimal risk stratification and treatment planning requires testing marrow or peripheral blood lymphoblasts for specific recurrent genetic abnormalities using: • Karyotyping of G-banded metaphase chromosomes (cytogenetics); • Interphase fluorescence in situ hybridization (FISH) testing including probes capable of detecting the major recurrent genetic abnormalities; • Reverse transcriptase polymerase chain reaction (RT-PCR) testing for fusion genes (eg, BCR-ABL). Additional optional tests include: • Flow cytometric DNA index/ploidy testing (additional assessment for hyperdiploidy and hypodiploidy). Classification: Together these studies allow determination of the World Health Organization (WHO) ALL subtype and cytogenetic risk group.
Test Purpose:	Classification, Diagnostic, Prognostic
When to Test:	
Guideline Page with Test Recommendation:	ALL-1 Page:5
Notes:	Subtypes: B-cell lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities include hyperdiploidy, hypodiploidy, and commonly occurring translocations: t(9;22)(q34;q11.2)[BCR-ABL1];t(v;11q23)[MLL rearranged]; t(12;21)(p13;q22)[TEL-AML1]; t(1;19)(q23;p13.3)[E2A-PBX1]; t(5;14)(q31;q32)[IL3-IGH; relatively rare]. Cytogenetic risk groups are defined as follows: Good risk: Hyperdiploidy (51-65 chromosomes and/or DNA index > 1.16; cases with trisomy of chromosomes 4,10, and 17 appear to have the most favorable outcome); t(12;21)(p13;q22):TEL-AML1. Poor risk: Hypodiploidy (<44 chromosomes and/or DNA index <0.81); t(v;11q23: MLL rearranged; t(9;22)(q34;q11.2): BCR-ABL1 (defined as high risk in the pre-TKI era); Complex karyotype (5 or more chromosomal abnormalities).

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 21 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	
Molecular Abnormality:	ABL1 kinase domain mutation analysis
Test:	ABL kinase domain mutation analysis
Chromosome:	9q34.1
Gene Symbol:	ABL1
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Consider *ABL* gene mutation testing for Ph+ ALL (AYA), Ph+ ALL (Adult) for relapse/refractory disease.
Test Purpose:	Treatment decision
When to Test:	
Guideline Page with Test Recommendation:	ALL-7 Page:11, ALL-D 3 of 4 Page:20
Notes:	

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 27 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	
Molecular Abnormality:	Abnormal immunophenotypes
Test:	Flow cytometry
Chromosome:	
Gene Symbol:	
Test Detects:	Protein expression
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Year 1 (every 1-2 months), bone marrow aspirate as indicated. If bone marrow aspirate is done: comprehensive cytogenetics, FISH, flow cytometry and consideration of molecular tests.
Test Purpose:	Surveillance
When to Test:	
Guideline Page with Test Recommendation:	ALL-7 Page:11
Notes:	

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 28 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	
Molecular Abnormality:	Abnormal immunophenotypes
Test:	Multicolor flow cytometry
Chromosome:	
Gene Symbol:	
Test Detects:	Abnormal immunophenotypes
Methodology:	Flow cytometry
NCCN Category of Evidence:	2A
Specimen Types:	Sampling of bone marrow mononuclear cells is preferred
NCCN Recommendation - Clinical Decision:	Minimal Residual Disease assessment. Timing of MRD assessment: Upon completion of intial induction; Additional timepoints may be useful depending on the regimen used
Test Purpose:	Monitoring
When to Test:	
Guideline Page with Test Recommendation:	ALL-3 Page:7, ALL-4 Page:8, ALL-5 Page:9, ALL-6 Page:10, ALL-F Page:23
Notes:	

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 29 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	
Molecular Abnormality:	Chromosomal abnormalities
Test:	Comprehensive cytogenetics
Chromosome:	
Gene Symbol:	
Test Detects:	Chromosomal abnormalities
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Year 1 (every 1-2 months), bone marrow aspirate as indicated. If bone marrow aspirate is done: comprehensive cytogenetics, FISH, flow cytometry and consideration of molecular tests.
Test Purpose:	Surveillance
When to Test:	
Guideline Page with Test Recommendation:	ALL-7 Page:11
Notes:	

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 30 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	
Molecular Abnormality:	Chromosomal abnormalities
Test:	FISH
Chromosome:	
Gene Symbol:	
Test Detects:	
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Year 1 (every 1-2 months), bone marrow aspirate as indicated. If bone marrow aspirate is done: comprehensive cytogenetics, FISH, flow cytometry and consideration of molecular tests.
Test Purpose:	Surveillance
When to Test:	
Guideline Page with Test Recommendation:	ALL-7 Page:11
Notes:	

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 31 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	
Molecular Abnormality:	Fusion genes and clonal rearrangements
Test:	Real time quantitative PCR
Chromosome:	
Gene Symbol:	
Test Detects:	Fusion genes and clonal rearrangements
Methodology:	RQ-PCR
NCCN Category of Evidence:	2A
Specimen Types:	Sampling of bone marrow mononuclear cells is preferred
NCCN Recommendation - Clinical Decision:	Minimal Residual Disease assessment. Timing of MRD assessment: Upon completion of intial induction; Additional timepoints may be useful depending on the regimen used
Test Purpose:	Monitoring
When to Test:	
Guideline Page with Test Recommendation:	ALL-3 Page:7, ALL-4 Page:8, ALL-5 Page:9, ALL-6 Page:10, ALL-F Page:23
Notes:	

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 32 | Under Review

Disease Description:	Acute Lymphoblastic Leukemia
Specific Indication:	
Molecular Abnormality:	TPMT gene polymorphisms
Test:	TPMT gene polymorphisms
Chromosome:	6p22.3
Gene Symbol:	TPMT
Test Detects:	Polymorphisms
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	For patients receiving 6-MP, consider testing for TPMT gene polymorphisms, particularly in patients that develop severe neutropenia after starting 6-MP.
Test Purpose:	Treatment decision
When to Test:	
Guideline Page with Test Recommendation:	ALL-D 1 of 4 Page: 18, ALL-D 2 of 4 Page: 19
Notes:	

Guideline: Acute Lymphoblastic Leukemia v.1.2014, as of 12/4/2014 10:22:20 AM NCCN Reference ID: 33 | Under Review

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	CEBPA mutation
Test:	CEBPA mutation
Chromosome:	19q13.1
Gene Symbol:	CEBPA
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Evaluation for acute leukemia. Bone marrow with cytogenetics (karyotype +/- FISH) and molecular analysis (KIT, FLT3-ITD, NPM1, and CEBPA mutations).
Test Purpose:	Diagnostic, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6
Notes:	Molecular abnormalities (KIT, FLT3-ITD, NPM1, and CEBPA mutations) are important for prognostication in a subset of patients (category 2A) and may guide therapeutic intervention (category 2B) These are useful for patients with normal karyotype (especially FLT3-ITD, NPM1 mutations) or core binding factor leukemia (especially c-KIT mutation). Multiplex gene panels and sequencing assays are available for the assessment of other molecular abnormalities that may have prognostic impact in AML. If a test is not available at your institution, consult pathology about preserving material from the original diagnostic sample for future use at an outside reference lab after full cytogentic data are available.

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 34

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	FLT3 internal tandem duplication (ITD) mutation
Test:	FLT3 internal tandem duplication (ITD) mutation
Chromosome:	13q12
Gene Symbol:	FLT3
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Evaluation for acute leukemia. Bone marrow with cytogenetics (karyotype +/- FISH) and molecular analysis (KIT, FLT3-ITD, NPM1, and CEBPA mutations).
Test Purpose:	Diagnostic, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6
Notes:	Molecular abnormalities (KIT, FLT3-ITD, NPM1, and CEBPA mutations) are important for prognostication in a subset of patients (category 2A) and may guide therapeutic intervention (category 2B) These are useful for patients with normal karyotype (especially FLT3-ITD, NPM1 mutations) or core binding factor leukemia (especially c-KIT mutation). Multiplex gene panels and sequencing assays are available for the assessment of other molecular abnormalities that may have prognostic impact in AML. If a test is not available at your institution, consult pathology about preserving material from the original diagnostic sample for future use at an outside reference lab after full cytogentic data are available.

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 35

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	KIT mutation
Test:	C-KIT mutation
Chromosome:	4q12
Gene Symbol:	KIT
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Evaluation for acute leukemia. Bone marrow with cytogenetics (karyotype +/- FISH) and molecular analysis (KIT, FLT3-ITD, NPM1, and CEBPA mutations).
Test Purpose:	Diagnostic, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6
Notes:	Molecular abnormalities (KIT, FLT3-ITD, NPM1, and CEBPA mutations) are important for prognostication in a subset of patients (category 2A) and may guide therapeutic intervention (category 2B) These are useful for patients with normal karyotype (especially FLT3-ITD, NPM1 mutations) or core binding factor leukemia (especially c-KIT mutation). Multiplex gene panels and sequencing assays are available for the assessment of other molecular abnormalities that may have prognostic impact in AML. If a test is not available at your institution, consult pathology about preserving material from the original diagnostic sample for future use at an outside reference lab after full cytogentic data are available.

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 36

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	NPM1 mutation
Test:	NPM1 mutation
Chromosome:	5q35
Gene Symbol:	NPM1
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Evaluation for acute leukemia. Bone marrow with cytogenetics (karyotype +/- FISH) and molecular analysis (KIT, FLT3-ITD, NPM1, and CEBPA mutations).
Test Purpose:	Diagnostic, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6
Notes:	Molecular abnormalities (KIT, FLT3-ITD, NPM1, and CEBPA mutations) are important for prognostication in a subset of patients (category 2A) and may guide therapeutic intervention (category 2B) These are useful for patients with normal karyotype (especially FLT3-ITD, NPM1 mutations) or core binding factor leukemia (especially c-KIT mutation). Multiplex gene panels and sequencing assays are available for the assessment of other molecular abnormalities that may have prognostic impact in AML. If a test is not available at your institution, consult pathology about preserving material from the original diagnostic sample for future use at an outside reference lab after full cytogentic data are available.

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 37

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	Abnormal immunophenotypes
Test:	Immunophenotyping
Chromosome:	
Gene Symbol:	
Test Detects:	
Methodology:	Flow cytometry, IHC
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Evaluation for acute leukemia. Immunophenotyping and cytochemistry
Test Purpose:	Diagnostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 38

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	Mutations
Test:	Molecular studies (general)
Chromosome:	
Gene Symbol:	
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Marrow to document remission status upon hematologic recovery, including cytogenetics and molecular studies as appropriate.
Test Purpose:	Monitoring remission status
When to Test:	Post induction therapy, upon hematologic recovery
Guideline Page with Test Recommendation:	AML-8 Page:13, AML-9 Page:14, AML-D Page:24
Notes:	Relevant only in APL and Ph+ disease (AML-D) at this time

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 39

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	(9;22) translocation
Test:	(9;22) translocation
Chromosome:	t(9;22)
Gene Symbol:	BCR-ABL1
Test Detects:	Translocation
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor risk; Cytogenetics: t(9;22).
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 41

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	(16;16) translocation
Test:	(16;16) translocation
Chromosome:	t(16;16)
Gene Symbol:	CBFB-MYH11
Test Detects:	Translocation
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Favorable risk; Cytogenetics: Core binding factor: inv(16) or t(16;16) or t(8;21) Risk status: intermediate risk, Molecular abnormalities: t(16;16) with c-KIT mutation.
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 43

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	16 inversion
Test:	16 inversion
Chromosome:	inv(16)
Gene Symbol:	CBFB-MYH11
Test Detects:	Chromosome inversion
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Favorable risk; Cytogenetics: Core binding factor: inv(16) or t(16;16) or t(8;21).
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 45

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	CEBPA mutation
Test:	CEBPA mutation
Chromosome:	19q13.1
Gene Symbol:	CEBPA
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2B
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Favorable risk; Molecular abnormalities: Normal cytogenetics with isolated biallelic CEBPA mutation.
Test Purpose:	Treatment decision
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6, AML-10 Page:15, AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 46

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	CEBPA mutation
Test:	CEBPA mutation
Chromosome:	19q13.1
Gene Symbol:	CEBPA
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Favorable risk; Molecular abnormalities: normal cytogenetics with isolated biallelic CEBPA mutation
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 47

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	(6;9) translocation
Test:	(6;9) translocation
Chromosome:	t(6;9)
Gene Symbol:	DEK-NUP214
Test Detects:	Translocation
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Cytogenetics: t(6;9).
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 48

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	FLT3 internal tandem duplication (ITD) mutation
Test:	FLT3 internal tandem duplication (ITD) mutation
Chromosome:	13q12
Gene Symbol:	FLT3
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Molecular abnormalities: normal cytogenetics with FLT3-ITD mutation.
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 50

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	FLT3 internal tandem duplication (ITD) mutation
Test:	FLT3 internal tandem duplication (ITD) mutation
Chromosome:	13q12
Gene Symbol:	FLT3
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2B
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Molecular abnormalities: normal cytogenetics with FLT3-ITD mutation.
Test Purpose:	Treatment decision
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6, AML-10 Page:15, AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 51

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	FLT3 tyrosine kinase domain mutation D835
Test:	FLT3 tyrosine kinase domain mutation D835
Chromosome:	13q12
Gene Symbol:	FLT3
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	FLT3-ITD mutations are considered to confer a significantly poorer outcome in patients with normal karyotype. There is controversy as to whether FLT3-TKD mutations carry an equally poor prognosis
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 52

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	KIT mutation
Test:	C-KIT mutation
Chromosome:	4q12
Gene Symbol:	KIT
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2B
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Intermediate risk; Molecular abnormalities: t(8;21), inv(16), t(16;16) with c-KIT mutation.
Test Purpose:	Treatment decision
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6, AML-10 Page:15, AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 54

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	KIT mutation
Test:	C-KIT mutation
Chromosome:	4q12
Gene Symbol:	KIT
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Intermediate risk; Molecular abnormalities: t(8;21), inv(16), t(16;16) with c-KIT mutation.
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 55

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	Translocations
Test:	Translocations
Chromosome:	11q23
Gene Symbol:	MLL
Test Detects:	Translocation
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Cytogenetics: 11q23 abnormalities.
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 57

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	(9;11) translocation
Test:	(9;11) translocation
Chromosome:	t(9;11)
Gene Symbol:	MLLT3-MLL
Test Detects:	Translocation
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Intermediate risk; Cytogenetics: t(9;11).
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 59

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	NPM1 mutation
Test:	NPM1 mutation
Chromosome:	5q35
Gene Symbol:	NPM1
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2B
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Favorable risk; Molecular abnormalities: normal cytogenetics with NPM1 mutation in absence of FLT3 ITD.
Test Purpose:	Treatment decision
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6, AML-10 Page:15, AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 60

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	NPM1 mutation
Test:	NPM1 mutation
Chromosome:	5q35
Gene Symbol:	NPM1
Test Detects:	Mutation
Methodology:	
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Favorable risk; Molecular abnormalities: normal cytogenetics with NPM1 mutation in absence of FLT3 ITD.
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 61

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	3 inversion
Test:	3 inversion
Chromosome:	inv(3)
Gene Symbol:	RPN1-EVI1
Test Detects:	Chromosome inversion
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Cytogenetics: inv(3).
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 63

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	3 translocation
Test:	3 translocation
Chromosome:	t(3;3)
Gene Symbol:	RPN1-EVI1
Test Detects:	Translocation
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Cytogenetics: t(3;3).
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 65

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	(8:21) translocation
Test:	(8:21) translocation
Chromosome:	t(8;21)
Gene Symbol:	RUNX1-RUNX1T1
Test Detects:	Translocation
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Favorable risk; Cytogenetics: Core binding factor: inv(16) or t(16;16) or t(8;21)
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 67

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	Chromosomal abnormalities
Test:	Cytogenetics
Chromosome:	
Gene Symbol:	
Test Detects:	Chromosomal abnormalities
Methodology:	Cytogenetics, FISH
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Evaluation for acute leukemia. Bone marrow with cytogenetics (karyotype +/- FISH)
Test Purpose:	Diagnostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-1 Page:6
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 68

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	5 deletion
Test:	5 deletion
Chromosome:	-5
Gene Symbol:	
Test Detects:	Chromosome deletion
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Cytogenetics: -5.
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 70

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	5q deletion
Test:	5q deletion
Chromosome:	del(5q)
Gene Symbol:	
Test Detects:	Chromosome deletion
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Cytogenetics: del(5q).
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 72

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	7 deletion
Test:	7 deletion
Chromosome:	-7
Gene Symbol:	
Test Detects:	Chromosome deletion
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Cytogenetics: -7.
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 74

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	7q deletion
Test:	7q deletion
Chromosome:	del(7q)
Gene Symbol:	
Test Detects:	Chromosome deletion
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Cytogenetics: del(7q).
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 76

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	Chromosomal abnormalities
Test:	Cytogenetics
Chromosome:	
Gene Symbol:	
Test Detects:	Chromosomal abnormalities
Methodology:	Karyotype analysis
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Marrow to document remission status upon hematologic recovery, including cytogenetics and molecular studies as appropriate.
Test Purpose:	Monitoring remission status
When to Test:	Post induction therapy, upon hematologic recovery
Guideline Page with Test Recommendation:	AML-8 Page:13, AML-9 Page:14, AML-D Page:24
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 77

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	Chromosomal abnormalities, complex karyotype, monosomal karyotype
Test:	Cytogenetics
Chromosome:	
Gene Symbol:	
Test Detects:	Gross chromosomal abnormalities
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Poor-risk; Cytogenetics: Complex (≥ 3 clonal chromosomal abnormalities); Monosomal karyotype.
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/26/2015 2:54:54 PM NCCN Reference ID: 79

Disease Description:	Acute Myeloid Leukemia (AML)
Specific Indication:	
Molecular Abnormality:	Trisomy of chromosome 8
Test:	Trisomy of chromosome 8
Chromosome:	+8
Gene Symbol:	
Test Detects:	Trisomy
Methodology:	Cytogenetics
NCCN Category of Evidence:	2A
Specimen Types:	
NCCN Recommendation - Clinical Decision:	Risk status: Intermediate-risk; Cytogenetics: +8 alone.
Test Purpose:	Classification, Prognostic
When to Test:	Evaluation for acute leukemia
Guideline Page with Test Recommendation:	AML-A Page:20
Notes:	

Guideline: Acute Myeloid Leukemia (AML) v.1.2015, as of 2/13/2015 4:46:20 PM NCCN Reference ID: 81