LPL - LPL-ROHINI (NATIONAL REFERENCE LAB)
SECTOR - 18, BLOCK -E ROHINI
DELHI 110085

Name	:	**Z662**				**Collected**	: **1/9/2017 12:00:00AM**
						Received	: **1/9/2017 10:53:16AM**
Lab No.	:	**135091534**	**Age: 36 Years**	**Gender:**	**Female**	**Reported**	: **2/9/2017 10:41:47AM**
A/c Status	:	**P**	**Ref By : Dr. UNKNWON**			**Report Status**	: **Final**

Test Name	Results	Units	Bio. Ref. Interval
LEUKEMIA GENETIC PROFILE– ANY SIX MARKERS, PCR QUALITATIVE			
AML ETO GENE REARRANGEMENT t(8;21) (q22;q22);(RUNX1;RUNX1T1), PCR, QUALITATIVE (Real Time PCR)	**Positive**		

Note

1. Sensitivity of the assay is 0.01% when copies of ABL detected is 100,000
2. Limit of detection is 10 copies of 8;21 fusion gene transcripts per PCR
3. This is an in-house developed assay designed as per EAC (Europe Against Cancer) protocol
4. Test conducted on Whole blood / Bone Marrow

Comments

Cytogenetic aberrations play a central role in the classification of AML. These aberrations are detected in 50-70% cases of AML by using standard techniques. The AML1(CBFA2, RUNX1)-ETO (MTG8) gene fusion results from the t(8;21)(q22;q22), which is the commonest chromosomal rearrangement associated with AML, being detected in approximately 8% of AML cases in children and young adults. Most t(8;21) positive AML's are de novo leukemias - vast majority being M2 FAB subtype. This translocation creates chimeric genes encoding fusion proteins that interfere with the function of CBFα and block the maturation of myeloid cells.

Uses

- For diagnostic identification of AML having morphological, imuunophenotypic or clinical features strongly suggestive of translocation 8;21
- For prognostic evaluation - Presence of this translocation is associated with a favorable prognosis

BCR-ABL GENE REARRANGEMENT, PCR QUALITATIVE (Real Time PCR)		
BCR-ABL gene rearrangement	**Positive**	
Type of Translocation	Major	

Note

1. Sensitivity of the assay is 0.01% when copies of ABL detected is 100,000
2. Limit of detection is 10 copies of BCR-ABL fusion gene transcripts per PCR
3. This is an in-house developed assay designed as per EAC (Europe Against Cancer) protocol
4. This test detects Major (M) gene rearrangements namely- e13a2 & e14a2 and Minor (m) gene arrangement e1a2. This test does not detect micro gene rearrangement e19a2

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Test Name	Results	Units	Bio. Ref. Interval

5. Test conducted on Whole blood / Bone Marrow

Comments

Chronic Myeloid Leukemia (CML) is the commonest myeloproliferative neoplasm and possibly the commonest adult leukemia in India. This clonal stem cell disorder is characterized by a proliferation of myeloid cells at all stages of differentiation and the t(9:22) (q34:q11) leading to formation of BCR-ABL fusion gene. Cytogenetic and molecular studies are vital for the diagnosis of CML by using detection procedures for Philadelphia chromosome. The abnormality is present in over 95% patients of CML while remainder 5% have complex or variant translocations involving additional chromosomes. Major gene rearrangements are detected in CML while minor gene rearrangement may be detected in ALL.

Uses

- To detect & monitor therapy in CML patients.
- As a prognostic marker in ALL patients. Presence of BCR-ABL gene rearrangement is associated with poor prognosis.

FLT3 GENE MUTATION
(Real Time PCR and fragment analysis)

FLT3-ITD	**Positive**
FLT3-TKD	Negative

Note

1. This test detects FLT3-internal tandem duplication (ITD) by fragment analysis on a capillary electrophoresis system and FLT3-tyrosine kinase domain mutation(TKD) D835 by real time PCR
2. This is an in-house developed assay
3. Test conducted on Whole blood / Bone Marrow

Comments

FLT3 mutations are class I mutations that occur in 30% AML with normal karyotype, AML with t(15;17), AML with t(6;9) and in AML with mutated NPM1. The mutations are most commonly internal tandem duplications. There are also cases of point mutations within the tyrosine kinase domain. The presence of FLT3 mutations confers a poorer prognosis than in similar cases without FLT3 mutations.

Uses

- For prognostic evaluation of AML

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Test Name	Results	Units	Bio. Ref. Interval
NPM1 GENE MUTATION (Real Time PCR)	**Positive**		

Note

1. This test detects A, B, D types of NPM1 mutations
2. This is an in-house developed assay
3. Test conducted on Whole blood / Bone Marrow

Comments

Evaluation of NPM1 is recommended in cases of cytogenetically normal AML as detection of these mutations in the absence of a FLT3 mutation confers a good prognosis and thus NPM1 mutations should always be interpreted with results of FLT3 mutations. NPM1 mutations are class II mutations which impair hematopoietic differentiation and subsequent apoptosis. They do not occur in combination with the recurrent cytogenetic abnormalities in AML and may represent a biologically distinct entity. More than 10 types of NPM mutations have been reported; types A, B, D together constitute >95% of the mutations.

Uses

- For prognostic evaluation of AML

PML-RARA GENE REARRANGEMENT, PCR QUALITATIVE (Real Time PCR)	
PML - RARA Type bcr 1	**Positive**
PML - RARA Type bcr 2	Negative
PML - RARA Type bcr 3	Negative

Note

1. Sensitivity of the assay is 0.01% when copies of ABL detected is 100,000
2. Limit of detection is 10 copies of PML-RARA fusion gene transcripts per PCR
3. This is an in-house developed assay designed as per EAC (Europe Against Cancer) protocol
4. This test detects 3 types of translocations - bcr 1, bcr 2 & bcr 3
5. Test conducted on Whole blood / Bone Marrow

Comments

Acute Promyelocytic Leukemia (APL) is characterized by a unique reciprocal chromosomal translocation t(15;17) (q22;q11-12) and its underlying fusion gene PML / RARA rearrangement. The fusion is seen between

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Test Name	Results	Units	Bio. Ref. Interval

Promyleocytic (PML) gene on chromosome 15 and RARA gene on chromosome 17. Based on PML breakpoint location, the PML RARA transcripts subtype bcr 1 & bcr 2 (Long transcript type) and bcr 3 (Short transcript type) may be formed.

Uses

- For diagnostic identification of PML RARA in cases of Acute Promyelocytic Leukemia
- To assess molecular resistance & predict response to treatments containing ATRA and / or ATO

t (12; 21) (p13;q22); ETV6-RUNX1(TEL-AML1) (Real Time PCR)	**Positive**		

Comments

Precursor B-cell Acute Lymphoblastic Leukemia (ALL) accounts for 85% Acute leukemias in children and 20% in adults. Most patients with ALL show an abnormal clone by conventional cytogenetic studies. The common chromosome translocations in pediatric ALL include t(1;19) (q23 ;p13.3); TCF3-PBX1(E2A-PBX1), t(12; 21) (p13;q22); ETV6-RUNX1(TEL-AML1) & MLL gene rearrangement. All these translocations are important to detect as they are important prognostic markers. Detection of translocation t(12;21) which is commonest in B-ALL is associated with good prognosis in children.

Uses

- Quantifying disease before treatment and after therapy for patients with ALL
- Assessing residual disease after treatment

t (4;11) (q21;q23); (MLL-AF4) (Real Time PCR)	Negative		

Comments

Precursor B-cell Acute Lymphoblastic Leukemia (ALL) accounts for 85% Acute leukemias in children and 20% in adults. Most patients with ALL show an abnormal clone by conventional cytogenetic studies. The common chromosome translocations in pediatric ALL include t(1;19)(q23;p13.3);TCF3-PBX1(E2A- PBX1), t(12;21) (p13;q22); ETV6-RUNX1(TEL-AML1) & MLL gene rearrangement. All these translocations are important to detect as they are important prognostic markers. The t(4;11)(q21;q23) results in the MLL-AF4 fusion gene and is the most frequent MLL translocation in ALL but is rare in AML. Detection of translocation t(4;11) is seen in 4-6% cases in children and adults. It is generally associated with a poor prognosis.

Uses

- Quantifying disease before treatment and after therapy for patients with ALL
- Assessing residual disease after treatment

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Test Name	Results	Units	Bio. Ref. Interval
t (9;11) (p21-22;q23); (MLL-AF9) (Real Time PCR)	Negative		

Comments

Abnormalities of mixed lineage Leukemia (MLL) gene on chromosome band 11q23 can be detected in Acute Myeloid Leukemia (AML) and Acute Lymphoblastic Leukemia (ALL) as well as in therapy related AML particularly after treatment with DNA topoisomerase II inhibitors. Upto 50 different translocations involving the MLL gene have been reported. t(9;11)(p21-22;q23) resulting in MLL - AF9 fusion gene is the most common MLL abnormality in AML.

t (1;19) (q23 ;p13.3); TCF3-PBX1(E2A- PBX1) (Real Time PCR)	Negative		

Comments

Precursor B-cell Acute Lymphoblastic Leukemia (ALL) accounts for 85% Acute leukemias in children and 20% in adults. Most patients with ALL show an abnormal clone by conventional cytogenetic studies.The common chromosome translocations in pediatric ALL include t(1;19)(q23;p13.3); TCF3-PBX1(E2A-PBX1), t(12;21) (p13;q22); ETV6-RUNX1(TEL-AML1) & MLL gene rearrangement. All these translocations are important to detect as they are important prognostic markers. Detection of translocation t(1;19) shows improved outcome with intensive chemotherapy.

Uses

- Quantifying disease before treatment and after therapy for patients with ALL
- Assessing residual disease after treatment

t (11;19) (q23;p13.3); (MLL-ENL) (Real Time PCR)	**Positive**		

Comments

Precursor B-cell Acute Lymphoblastic Leukemia (ALL) accounts for 85% Acute leukemias in children and 20% in adults. Most patients with ALL show an abnormal clone by conventional cytogenetic studies. The common chromosome translocations in pediatric ALL include t(1;19) (q23;p13.3);TCF3-PBX1(E2A-PBX1), t(12;21) (p13;q22); ETV6-RUNX1 (TEL-AML1) & MLL gene rearrangement. All these translocations are important to detect as they are important prognostic markers. Translocation t(11;19) (q23;p13.3) generates MLL-ENL fusion gene which is observed with equal frequency in AML & ALL.

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Test Name	Results	Units	Bio. Ref. Interval

Uses

- Quantifying disease before treatment and after therapy for patients with ALL
- Assessing residual disease after treatment

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Test Name	Results	Units	Bio. Ref. Interval
INV 16 (p13q22) / t(16;16)(p13;q22), GENE REARRANGEMENT, PCR , QUALITATIVE (Real Time PCR)	**Positive**		

Note

1. Sensitivity of the assay is 0.01% when copies of ABL detected is 100,000
2. Limit of detection is 10 copies of Inv(16)(p13q22) / t(16;16)(p13;q22) with the CBFB-MYH11 fusion gene transcript per PCR
3. This is an in-house developed assay designed as per EAC (Europe Against Cancer) protocol
4. This tests detects A, D, and E transcripts but does not distinguish between them
5. Test conducted on Whole blood / Bone Marrow

Comments

Cytogenetic aberrations play a central role in the classification of AML. These aberrations are detected in 50-70% cases of AML by using standard techniques. Pericentric inversion of chromosome 16, inv(16) (p13q22), is found in about 4% of cytogenetically abnormal AML cases. Inv(16) or the rarer t(16;16)(p13;q22) creates chimeric genes encoding fusion genes that interfere with the function of CBF α and block the maturation of myeloid cells. Around 10 different CBFB-MYH11 FG transcripts have been reported. More than 85% of positive patients have the type A transcript; type D and E transcripts each represent nearly 5%, whereas all other types occur sporadically.

Uses

- For diagnostic identification of AML having morphological, imuunophenotypic or clinical features strongly suggestive of M4eo FAB subtype
- For prognostic evaluation - Presence of this translocation is associated with a favorable prognosis

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Sr. Consultant Pathologist

----------End of report ----------

Not in NABL scope