

<b>Name</b> : Z775	<b>Collected:</b> 01-09-2017
<b>Lab No.</b> : 135091527 <b>Age</b> : 26 Years <b>Gender:</b> Female	<b>Received:</b> 01-09-2017
<b>A/C Status</b> :	<b>Reported:</b> 01-09-2017
<b>Ref by</b> : Unknown	<b>Report status:</b> Final

**Fluorescence in-situ Hybridization (FISH)**

**ALL FISH Panel**

**Specimen** : Bone Marrow

**Clinical Indication** : ? ALL

**Result** : nuc ish(BCR×2)(ABL×2)[200]  
 : nuc ish(5'MLL,3'MLL)×2(5'MLLcon 3'MLL×1)[168/200]  
 : nuc ish(TCF3×2)(PBX1×2)[200]  
 : nuc ish(ETV6×2),(RUNX1×2)[200]

**Interpretation** : Specimen is **positive** for 11q23(MLL gene rearrangement) in 84% cells  
 Specimen is negative for t(9;22)(q34;q11.2)(BCR/ABL), t(1;19)(q23;p13.3)  
 (PBX1/TCF3) & t(12;21)(p13;q22)(ETV6)(TEL)/RUNX1(AML1).

Probe: ZytoLight SPEC BCR/ABL1 Dual Color Dual Fusion Probe.

Interphase nuclei analyzed	Normal nuclei 2 Orange 2 Green signals	Abnormal nuclei 1 Orange 1 Green 2 Yellow signals
200	200	00

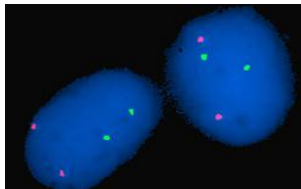
Cut off for the normal individual is 4%

Probe: LSI MLL Dual color, Break Apart Rearrangement probe

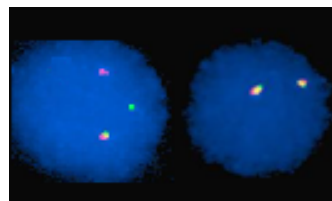
Interphase nuclei analyzed	Normal nuclei 2 Yellow	Abnormal nuclei 1 Orange 1 Green 1 yellow signals
200	32	168

Cut off for the normal individual is 3%

BCR/ABL1 Dual Color Dual Fusion



LSI MLL Dual color, Break Apart Rearrangement



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**Probe:** LSI PBX1 (1q23) S. Orange/LSI TCF3 (19p13.3) S. Green, Vysis, Abbott Molecular Inc.

Interphase nuclei analyzed	Normal nuclei 2 Orange 2 Green signals	Abnormal nuclei 1 Orange 1 Green 2 yellow signals
200	200	00

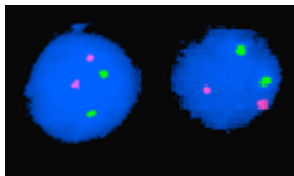
Cut off for the normal individual is 3%

**Probe:** Vysis LSI TEL(ETV6)(12p13) S. Green / LSI RUNX1(21q22) S. Orange Vysis, Abbott Molecular Inc.

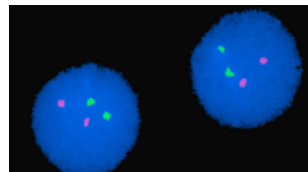
Interphase nuclei analyzed	Normal nuclei 2 Orange 2 Green signals	Abnormal nuclei 1 Orange 1 Green 2 yellow signals
200	200	00

Cut off for the normal individual is 3%

LSI PBX1 (1q23)/ TCF3 (19p13.3)



LSI TEL ETV6(12p13)/ LSI RUNX1(21q22)



**Comment:** t(9;22)(q34;q11.2) is the hallmark of almost all cases of Chronic Myeloid Leukemia (CML). In 5 to 10% of CML patients, the BCR/ABL gene fusion occurs in the absence of a cytogenetically detectable Ph chromosome as a result of more complex rearrangements. It is also seen in 25-30% cases of adult ALL and 2-5% of childhood ALL and is associated with an adverse outcome. Translocations involving the mixed-lineage leukemia (MLL) gene located at chromosomal band 11q23 occur in acute lymphocytic leukemia (ALL). t(1;19), found in 25% of childhood B-ALL with cytoplasmic  $\mu$  expression, fuses the transcription factor produced by *TCF3* at 19p13.3 with *PBX1* at 1q23. The t(12;21)(p13;q22) chromosomal translocation is the most common chromosomal rearrangement in childhood acute lymphoblastic leukaemia resulting in the fusion of the 5' section of the ETV6(TEL) gene on chromosome 12p13 to almost the entire RUNX1 (AML1) gene on chromosome 21q22. This translocation is present in approximately 30% of childhood pre-B-ALL and 3-4% of adult ALL. t(12;21) cannot be detected by standard cytogenetics unless a more complex rearrangement is present.

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**REPORT OF CHROMOSOME ANALYSIS FOR HEMATOLOGICAL MALIGNANCY**

**SPECIMEN** : Bone marrow

**INDICATION** : ? ALL

**MEDIUM USED** : RPMI– 1640, Hi-Karyol RPMI

**METHOD** : 24-hr unstimulated cultures with appropriate serum and antibiotics.

**BANDING RESOLUTION** : 450-550

**BANDING TECHNIQUE** : GTG (G bands by Trypsin and Giemsa)

**CYTOGENETIC PROFILE:**

Metaphases counted : 20

Metaphases analysed : 20

Metaphases karyotyped : 02

Metaphases photographed : 02

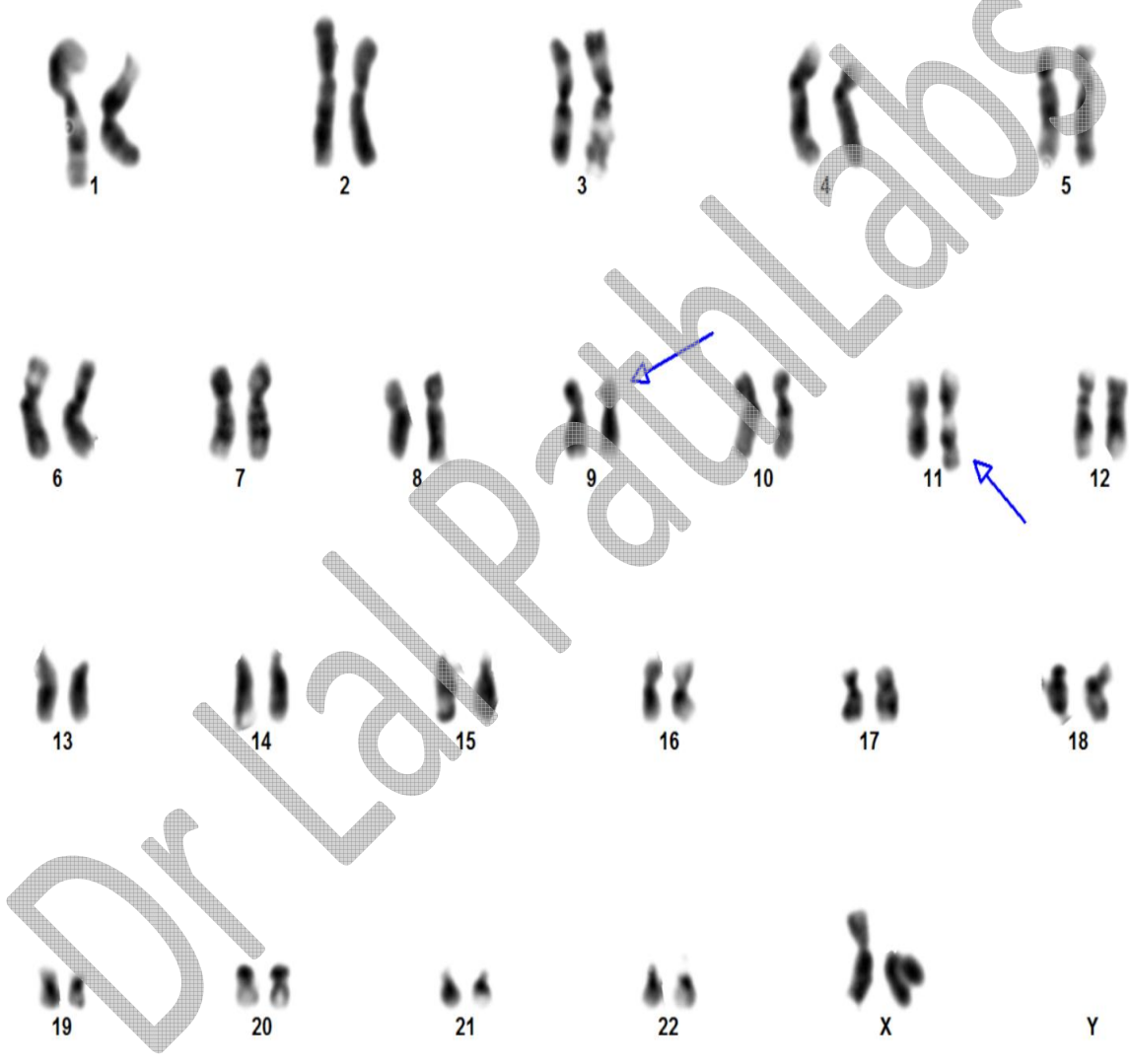
**KARYOTYPE** : 46,XX,t(9;11)(p22;q23)[15]/46,XX[5]

**INTERPRETATION** :

Fifteen of the 20 metaphases analysed are abnormal and show t(9;11). Kindly correlate these results with clinical and hematological findings.

**KARYOGRAM ATTACHED**

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*Saurabh*

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