## **CORE** DIAGNOSTICS<sup>™</sup>

## **MOLECULAR FINDINGS:**

S.No	Genomic Alterations	Associated FDA Approved	Associated Non FDA	<b>Clinical trials</b>	Fusion Reads / VAF
		Therapies	Approved Therapies in trials		
1	BCR(14) - ABL1(2), p210 (Major)	Imatinib Mesylate, Nilotinib, Dasatinib, Bosutinib & Ponatinib	Flumatinib, Asciminib & Radotinib	8	40924 fusion reads
2	NPM1, p.W288Cfs*12	None	None	5	24.0% VAF

## Variant for uncertain significance (VUS)

Gene (Exon) [Transcript]	Variant	VAF
RUNX1(9) [NM_001754.5]	p.Ser369Trp (c.1106C>G)	20.1%

## **RT-PCR Results (Qualitative):**

 Initially, we tested the index patient with AML Multiplex Basic (BCR - ABL, PML RaRa, AML ETO, INV 16, NPM1, FLT3, KIT) by Qualitative analysis performed through Real Time PCR (FLT3- ITD detection through RT-PCR followed by Gel Electrophoresis) where BCR-ABL p210 transcripts (t(9;22)(q34:q11) (Major)) were detected along with NPM1 mutation

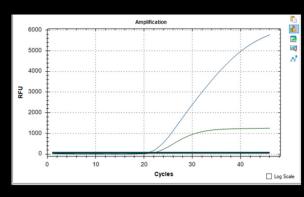


Figure 1: BCR ABL1 p210 by RT-PCR

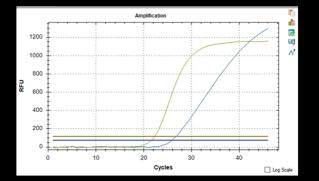


Figure 2: NPM1 mutation by RT-PCR