

About the BCR-ABL1 assay by real time polymerase chain reaction (RT-PCR)

The current TaqMan probe based assay detects the Major- e13a2/e14a2, minor e1a2 and micro e19a2 fusion transcripts of BCR-ABL1 gene in comparison to ABL1 as reference gene. Quantitative RT-PCR testing for all three BCR-ABL1 transcripts yielded positive results for both major and micro transcripts.

Transcript Variant	MAJOR	MICRO
Transcript copy number	12056	151568
ABL1 copy number	686647	686647
Ratio %	1.7557	22.0736
IS ratio %	0.9481	

Signals for BCR-ABL1 P230 (e19a2) micro were detected in leukocytes of the specimen. Additionally significant ratio of P210 major transcript was also detected in the specimen.

DISCUSSION

The co-occurrence of BCR-ABL1 fusion transcripts e19a2 and 13/14a2 is a rare event. The e19a2 encoding the p230 was earlier associated with neutrophilic CML but has been consistently reported in typical CML across all phases¹. The e19a2 falls among the rare BCR-ABL1 transcript variants and is the most common in this category. Even though associated with benign disease presentation and favorable prognosis, the clinical evidence remains ambiguous. Evidence from a Sudanese population study with cohort size of 43 CML patients reported the incidence of co-expression of e19a2 with e13a2/e14a2 to be only 2.3%. Such co-expression has also been reported by another Indian study in 2012 with an incidence of 0.48% ^{2.3}. It is still not completely understood how the presence of both p230 and p210 will impact the disease progression and needs further investigation.