

ABL single nucleotide polymorphisms may masquerade as BCR-ABL mutations associated with resistance to tyrosine kinase inhibitors in patients with chronic myeloid leukemia

Thomas Ernst, Jana Hoffmann, Philipp Erben, Benjamin Hanfstein, Armin Leitner, Rüdiger Hehlmann, Andreas Hochhaus, and Martin C. Müller

III. Medizinische Klinik, Medizinische Fakultät Mannheim der Universität Heidelberg, Mannheim, Germany

Supplementary Table S1. Selection of 8 patients for whom single nucleotide polymorphisms (SNPs) were confirmed by ABL allele-specific mutation analysis.

Patient Number	Age (yrs)	Gender	Phase of disease	BCR-ABL transcript	Nucleotide position ¹	Nucleotide change	Amino acid change ²	Proportion of mutated ABL allele (%)	Proportion of mutated BCR-ABL allele (%)	Therapy and best response
1	29	female	CP	b2a2	58758	G/A	T240T	60	100	Imatinib status not available, good molecular response for 26 months on dasatinib
2	39	male	CP	b3a2	58778	A/G	K247R	40	100	Imatinib intolerant, good molecular response for 24 months on dasatinib
3	32	male	CP	b2a2	58778	A/G	K247R	70	0	Good molecular response on imatinib
4	59	male	lyBC	e1a2	68708	T/G	F311V	50	100	Imatinib resistant, no response on dasatinib, time to progression 1 month
5	45	male	CP	b2a2	68722	T/G	T315T	40	100	Good molecular response since 37 months on 600 mg imatinib
6	62	female	CP	b2a2	68736	A/G	Y320C	80	0	Imatinib resistant, complete hematologic response for 24 months on dasatinib
7	72	male	CP	b2a2	74901	A/G	E499E	40	100	Minor cytogenetic response on imatinib
8	66	female	CP	b2a2	74901	A/G	E499E	50	100	Imatinib resistant, complete hematologic response for 16 months on dasatinib

CP: chronic phase; lyBC: lymphoid blast crisis. ¹Nucleotide positions according to GenBank accession number U07563 for the ABL 1a splice variant. ²Amino acid residues are denoted with the single letter code and correspond to the ABL 1a variant.