



Duplication of Isoderivative Ph Chromosome with Tp53 Deletion in a Case of Imatinib Resistant CML

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ABSTRACT Philadelphia chromosome, a consequence of BCR/ABL1 fusion on chromosome 22, is present in ninety percent chronic myeloid leukemia cases and corresponds to good response to first-line Imatinib Mesylate, regardless of the disease phase. There still remains a subset of patients failing to respond to Imatinib. These patients often harbor cytogenetic abnormalities like BCR/ABL1 gene amplification and Tp53 deletion that are strongly associated with resistance. ABL1 domain mutation is another major cause of resistance. The researchers' report a case of CML, displaying resistance to Imatinib- the current standard of care, due to duplication of isoderivative Philadelphia chromosome, translocation t(17;22)(p11.2;q11.2) and Tp53 deletion. This case additionally also showed ABL1 domain mutation. Chromosomal aberrations in this patient were identified by conventional karyotype and FISH, ABL domain mutation by RQ-PCR and BCR/ABL1 fusion gene copy number by real time PCR. Identifying such chromosomal aberrations can help predict clinical outcome and modify treatment.