DISCUSSION

Chronic myeloid leukemia (CML) is a myeloproliferative disorder and genetically characterized by the presence of the Philadelphia (Ph) chromosome, resulting from a balanced reciprocal translocation between chromosome 9 and 22 at bands 9q34 and 22q11.2 [t(9;22)(q34;q11.2)]. In the formation of the Ph chromosome, the specific region of the ABL oncogene is transposed from 9q34 to the specific region of the BCR gene on chromosome 22 to form a fusion gene BCR-ABL, which encodes a constitutively active protein, with tyrosine kinase activity. Vast majority of CML patients show classical Ph translocation, t(9;22) and 5-10% of cases are observed with variant Ph translocation. In variant Ph translocation, generally a third chromosome is involved, along with chromosome 9 and 22[4-5]. The segment of third chromosome usually translocates to the chromosome 9 at band 9q34. The formation of variant or complex Ph translocation is controversial topic. A wide array of additional chromosome involved in translocation with the t(9;22) has been described previously in CML patients[6-7]. In this report, we present a unique and complex Ph translocation involving three chromosomes (6;9;22).

Chromosome analysis revealed 46,XX,t(1;4)(p32;p16),t(6;9;22)(p23;q34;q11.2)[20]. In the addition of three way complex translocation t(6;9;22) another unusual translocation t(1;4)(p32;p16) was observed. The FISH assay was performed to confirm the presence of bcr/abl fusion gene, which was found to be present in all cells. To our knowledge, this is the first report with such a unique translocations in a Ph-positive CML patient involving three chromosomes along with clonal evolution with t(1;4) as a secondary abnormality.

Further clinical follow up and details of therapeutic response are warranted.