

Complex three-way Philadelphia translocation t(6;9;22) (p23;q34;q11.2) along with unusual t(1;4): A chronic myeloid leukemia case report

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, the third chromosome involved with chromosome 9 and 22. The segment of the 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. In this report, we present a unique and complex Ph translocation involving three chromosomes (6;9;22) in a 25-year-old female who was clinically diagnosed as CML. Chromosome analysis using 24 h and 48 h unstimulated culture with GTG-banding was performed

according to standard protocol. A total of 20 metaphases analyzed from the bone marrow culture of the patient and karyotyping was performed on the basis of International System for Human Cytogenetic Nomenclature (2016). The FISH analysis was performed according to the standard protocol from sample for confirmation of BCR/ABL gene using dual color fusion FISH probe (Zytovision Probe). All cell's images were captured using Olympus fluorescent microscope BX-61 equipped with a CCD camera and analyzed using Bioview FISH software.

Chromosome analysis revealed 46,XX,t (1;4)(p32;p16),t(6;9;22) (p23;q34;q11.2). In the addition of three-way complex translocation t(6;9;22), another unusual translocation t(1;4) (p32;p16) was observed. The FISH was performed to confirm the presence of BCR/ABL fusion gene, which was found to be in all cells. To the best of 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.

M. Kumar, Bekleshwar Salona, Shiv Murti Kumar, Akshay Gore, Shivani Sharma

Department of Cytogenetics, CORE Diagnostics, Gurgaon, 122016, Haryana, India

Correspondence to: Dr. Shivani Sharma, Email: shivani.sharma@corediagnostics.in

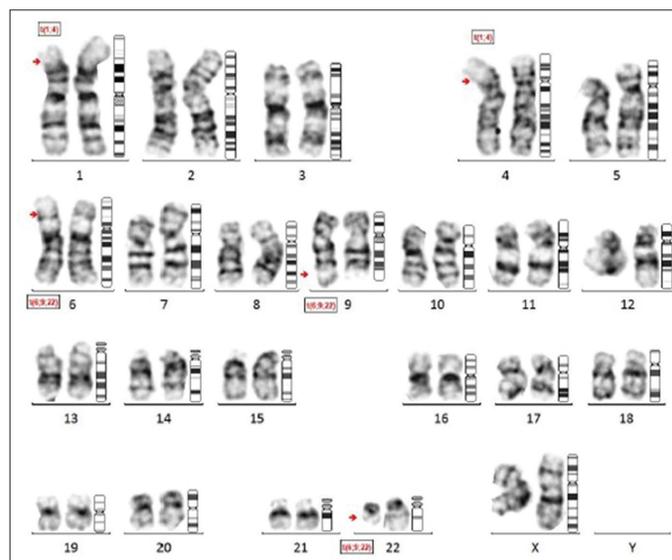


Figure 1: Female karyotype with 46,XX,t(1;4)(p32;p16),t(6;9;22)(p23;q34;q11.2)

How to cite this article: Kumar M, Salona B, Kumar SM, Gore A, Sharma S. Complex three-way Philadelphia translocation t(6;9;22) (p23;q34;q11.2) along with unusual t(1;4): A chronic myeloid leukemia case report. Int J Mol ImmunoOncol 2018;3:66.

Source of Support: Nil. **Conflict of Interest:** None declared.