FINAL DIAGNOSIS:

Coexistence of Inversion 16 and t(9;22) (Philadelphia Chromosome)in a case of CML-AP.

DISCUSSION:

Chronic myelogenous leukemia (CML) is a clonal hematopoietic disorder characterized by t(9;22) (q34;q11.2) translocation, that results in the generation of an abnormal chromosome 22 (Philadelphia-Ph+ chromosome), and BCR-ABL1 fusion oncogene. Cytogenetic clonal evolution in CML, the occurrence of additional chromosomal abnormalities in addition to the Ph+ chromosome, is associated with disease progression and is considered a marker that defines the blast phase of CML-BP (WHO 5th Ed.). The occurrence of additional cytogenetic alterations other than t (9;22) is observed in up to 80% of cases of CML-BP. The most common additional cytogenetic abnormalities include trisomy 8, an extra copy of the Ph chromosome, 3q26 rearrangements, monosomy 7/del(7q), i(17)(q10), trisomy 21, minus Y, and trisomy 19. *CBFB* rearrangement, particularly *CBFB-MYH11* fusion, resulting from inv(16)(p13.1q22) or less commonly t(16;16)(p13.1;q22), is an acute myeloid leukemia (AML)-defining alteration that is associated with a favorable outcome. The co-occurrence of BCR-*ABL1* fusion and *CBFB* rearrangement is extremely rare and is associated with poor prognosis. .

CONCLUSION:

This case illustrate the importance of a comprehensive diagnostic work-up and importance of flow cytometry and cytogenetics in identifying additional abnormalities of clinical significance in follow up cases.