INTERPRETATION:

Chromosome analysis revealed abnormal Male karyotype with the presence of a Four-way translocation involving the chromosome 2, 9, 22, and 17 at band 2p11, 9q34, 22q11.2 and 17p11 (Complex Ph chromosome).

DISCUSSION:

Chronic myeloid leukemia (CML) is a clonal myeloproliferative disorder and genetically characterized by the presence of the Philadelphia (Ph) chromosome, resulting from a balanced reciprocal translocation between the long arms of chromosome 9 and 22 at bands 9q34 and 22q11.2 [t(9;22) (q34;q11.2)]. This translocation leads to formation of a novel and chimeric BCR/ABL (break point cluster region-Abelson) gene in the breakpoint region of the derivative chromosome. The BCR/ABL fusion gene generated encodes an oncogenic protein tyrosine kinase which causes leukemogenesis of CML and is a target of tyrosine kinase inhibitors.

80 – 90% of CML cases have classical t (9;22) Philadelphia positive, however, in <5% of cases in CML, Complex Ph translocations have been observed. In few cases, submicroscopic, masked Ph translocation, cryptic rearrangements and complex t(9;22) is not detectable by G-banding. These types of cryptic and complex rearrangements can be detected by fluorescence in situ hybridization (FISH) and molecular analysis. Four way translocations involving 9 and 22 is very rare. There is no particular treatment guidelines for these CML patients. In this case, patient responded well to Imatinib. As per our experience patients with variant translocation do not differ significantly as compare to classical translocation.