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Letter to the Editor

In the era of next generation sequencing, is the cytogenetic analysis still important in polycythemia vera?

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Dear Editor,

Polycythemia vera (PV) is a Philadelphia chromosome-negative myeloproliferative neoplasm (MPN) characterized by increased red blood cell production. WHO 2008 criteria for PV diagnosis are based on laboratory, morphological, and molecular findings. Microsatellite studies on chromosome nine identified acquired uniparental disomy (UPD) as a common defect in MPN. V617F mutation in the Janus kinase 2 gene (JAK2) present in 95% of PV and >50% of essential thrombocythemia (ET) and primary myelofibrosis (PMF) patients.[1] New target gene CBL is associated with aberrations of chromosome 11q.[2] CBL exons 8 and 9 (11q23.3) mutations seen commonly in PMF and post MPN and rarer in PV and ET.[1] Here we report a case of PV with 11q23 deletion with a stable disease.

A 70-year-old male presented with pruritus for 1 year. Hemoglobin was 211 g/L, hematocrit–68.5%, total leucocyte count–20.6 × 10^6/L, platelet count–470 × 10^9/L with neutrophilic leukocytosis. Bone marrow was hypercellular with trilineage hyperplasia. Serum erythropoietin was 1.2 mIU/ml and JAK2 V617F from peripheral blood was positive. Cytogenetic analysis showed 46, XY with 11q23 deletion in 100% cells [Figure 1]. He was started on biweekly phlebotomy, aspirin 75 mg once daily and hydroxyurea 500 mg once daily; maintaining a hematocrit of 45% and off phlebotomy for the last 8 months.

Cytogenetic abnormalities in PV being del (20q), del (13q), +8, +9 and chromosome 1 abnormalities and acquired UPD of 1p, 4q 7q, 9p, and 11q associated with homozygosity for mutations in MPL, TET2, EZH2, JAK2 and CBL respectively.[2] CBL mutations in myeloid malignancies are associated with 11q acquired UPD.[3] The present case compared with cases from other studies with chromosome 11 abnormalities in Table 1. It's apparent from the literature that PV with chromosome 11 abnormalities has a chance of progression to AML. Hence, our index case has to be kept on close follow-up. Next generation sequencing based identification of

(Continue on page 50...)

(Letter to the editor continue from page 46...)

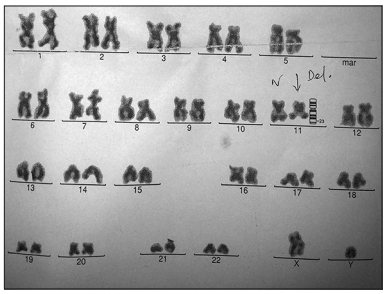

Figure 1: Cytogenetic analysis following unstimulated cell culture and GTG-banding revealed 46, XY, del (11q23) chromosomal pattern in all (100%) cells examined

Table 1: Comparison of present case with previous studies showing chromosome 11 abnormalities

Study	Number of cases showing aberration in chromosome 11 (11q)	Transformation	JAK2 positive status
Klampfl *et al.*[4]	3 (1)	N/A	N/A
Sever *et al.*[5]	2 (0)	Both cases	2/2
Gangat *et al.*[2]	1 (1)	N/A	N/A
Present case	1 (1)	No	1/1

N/A=Data not available, JAK2=Janus kinase 2

molecular markers indicating progression, are mostly not available in resource constrain countries like India. Therefore, one may still use conventional cytogenetics to indicate the molecular change responsible for progression.

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Conflicts of interest

There are no conflicts of interest.

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