Journal of Blood Research & Hematologic Diseases

Short Communication

A SCITECHNOL JOURNAL

Identification of JAK2 (V617F) mutation in Myeloproliferative Neoplasms by using Allele Specific Polymerase Chain Reaction (AS-PCR)

Khin La Pyae Tun

Yangon General Hospital, Myanmar

Abstract:

Myeloproliferative neoplasms (MPNs) is a group of clonal haematopoietic stem cell disorders characterized by the proliferation of one or more myeloid cell lineages. According to WHO classification, the Janus associated kinase 2 (JAK2) V617F mutation is the one of the major diagnostic criteria in BCR-ABL1 negative myeloproliferative neoplasms .The aim of this study is to detect the JAK2 (V617F) mutation in patients with myeloproliferative neoplasms to get accurate diagnosis and proper management. A total of 90 clinically diagnosed MPN patients attending to Department of Clinical Haematology ,Yangon General Hospital were enrolled in this study. The mean age was 53.4 ± 14 years which ranged from 16 to 81 years old and male and female ratio was 2.4:1. The identification of JAK2 (V617F) point mutation was found to be positive in 44/90 MPN patients (48.9%). According to MPN subtypes, the JAK2 mutation positivity was found in 19 out of 46 polycythemia vera patients (41.3 %), 17 out of 25 essential thrombocythemia patients (68%), 8 out of 15 primary myelofibrosis patients (53.3%), 0 of 4 others myeloproliferative neoplasms (0%). Confirmation of each of nine JAK2 mutation positive and negative samples were done by Sanger sequencing. The arterial or venous thrombotic attack was found in 32 /44 JAK 2 mutation positive cases (72.7%) and 12 /44 JAK2 mutation negative cases (27.3 %).The association between thrombotic attack and presence of JAK2 mutation was statistically significance with p=0.000. In our study, prevalence of JAK2 (V617F) mutation was lower in polycythemia vera patients compared to other studies which showed over 90% positivity of JAK2 mutation in PV . It might be due to the secondary polycythemia in which the chronic heavy smoking is the common cause. The diagnosis of myeloproliferative neoplasms mainly relies on the molecular genetics according to WHO classification. The Allele specific PCR reaction is sensitive, simple test and relatively cost effective. Therefore, the identification of JAK2 (V617F) somatic point mutation should be implemented as a routine diagnosis procedure for patients with chronic and suspected myeloproliferative neoplasms.

Biography:

Khin La Pyae Tun is a pathologist who works at Blood Research Division, Department of Medical Research, Myanmar. She has been working in research field for about 10 years. After getting her master degree in pathology, her great interest in haematology leads her to do research work focus on it. Since Myanmar is the developing county, diagnosis of haematological malignancy still rely mainly on haemato-clinical findings. Her wish to study gene muatation that can contribute towards the clinical diagnosis and management of the patients along with her research findings.