Title: Can certain clinical traits of myeloproliferative diseases be predicted by heterogenic patterns of the JAK2, MPL, and CALR genes?

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Abstract

Essential thrombocythemia (ET), polycythemia vera (PV), and primary myelofibrosis (PMF) are the three subtypes of myeloproliferative neoplasm (MPN), which is typically detected through mutation analysis in several key genes, including JAK2, MPL, and CALR. Using allele-specific PCR and sequencing, the mutations in the aforementioned genes in 50 MPN patients and 50 healthy volunteers were identified. According to the findings, there is a strong relationship between MPN and its subtypes and mutations (p 0.05). With the exception of ET, JAK2 (exon 14) mutation was associated with MPN and all of its subtypes. CALR (exon 9) type 1 mutation was only associated with ET, whereas CALR (exon 9) type 2 mutation was more common in MPN and PV (p 0.05). None of the mutations happened at the same time. There was no proof of a JAK2 (exon 12) mutation. Exons 12 and 10 of JAK2 and exons 9 and 2 of CALR may have mutations in Iranians with MPN, but exons 14 and 14 of JAK2 and MPL did not show any signs of a mutation throughout our analysis, disqualifying them as candidates for a diagnosis.

Keywords: *JAK2*, *MPL*, *CALR*, genes, exon, mutation, MPD, myeloproliferative neoplasm