The provenance of the JAK2 V617F exon 14 mutation in sudanese patients with chronic myeloproliferative neoplasms

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The understanding of the pathogenesis of myeloproliferative neoplasms has been aided significantly by the discovery of the JAK2 V617F exon 14 mutation and provided with additional capabilities for analysing and managing this type of disease. The aim of this study was to determine the frequency of JAK2 V617F exon 14 mutation in Sudanese patients with myeloproliferative disorders referred to Fedail Hospital and Radioisotopes Centre Khartoum (RICK) in Khartoum State-Sudan., and to investigate the differences of laboratory parameters between patients with JAK2 V617F exon 14 positive myeloproliferative neoplasms (MPNs) and JAK2 V617F exon 14 wild type MPNs.

Materials and Methods: A total of 166 patients with MPNs; 76 with polycythemia Vera (PV), 76 with essential thrombocythemia (ET) and 14 with primary myelofibrosis (PMF), and 11 healthy individuals were conducted from 2014 to 2018. DNA was isolated from peripheral blood leukocytes by QIAamp mini kit, and JAK2 V617F exon 14 mutation gene detected by quantitative real-time PCR (qRT-PCR) technology (Quant Studio 12K Flex) using TaqMan® Mutation Detection Assay and Sanger sequencing to confirm the results of TaqMan and to identify the type allele of mutations.

Results: The JAK2 V617F exon 14 was detected in 61.3% in all MPNs patients. The prevalence of JAK2 V617F exon 14 mutations was 68.6% in PV, 50% in ET and PMF patients. Mutation was not detected in 11 healthy adult people. The presence of JAK2 V617F exon 14 mutations was not associated with total WBCs count and PLTs count for PV patients, whoever the mutation positively correlates with high total RBCs count ($p = .005$), Hb concentration ($p = .018$) and HCT ($p = .016$) in PV patients, and with high total WBC count ($p = .000$) in ET patients. A JAK2 V617F exon 14 Sanger sequencing was done for 114 of the 166 patients to confirm the results of TaqMan and to identify the type allele of mutations; 64 PV, 38 ET and 12 PMF. The majority of JAK2 V617F exon 14 positive ET and PMF patients were heterozygous, while there is no JAK2 V617F exon14 homozygous allele was detected in PV patients.

Conclusions: The JAK2 V617F exon 14 mutation could be frequently detected in the Sudanese patients with MPNs, the vast majority of polycythemia patients and around half of the essential thrombocythemia and primary myelofibrosis have the mutation.

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