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## Detection of Janus kinase 2 (JAK2V617F) in sudanese patients with polycythemia in Khartoum state, Sudan

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**Background & Aim**: Erythrocytosis is form of hematological diseases, the main members of which are polycythemia vera (PV) , familial polycythemia and idiopathic erythrocytosis. The molecular pathogenesis of these disorders is unknown, but tyrosine kinases have been implicated in several related disorders. We investigated the presence of the cytoplasmic tyrosine kinase JAK2 in patients with polycythemia and the hematological difference between these types in Sudanese patients.

**Methods:** We measure the full blood counts and obtained DNA samples from patients with erythrocytosis. The *JAK2V617F* mutation was detected from peripheral-blood samples. Allele-specific PCR were undertaken on subgroups of patients who satisfied the WHO criteria of polycythemia vera.

**Results**: A single point mutation (V617F) was identified in JAK2 in 31 (54.4%) of 57 patients with polycythemia and there are no significant differences of hemoglobin, hematocrit, and neutrophil between patients with the JAK2 mutation and who without the mutation. Polycythemia vera patients who are not detected for the mutation and patients with idiopathic erythrocytosis showing no difference except in platelet's count .

**Interpretation:** A single mutation of JAK2V617F was noted in more than half of patients with erythrocytosis. If the person is negative for JAK2V617F mutations, the person may still have a PV. The person could have a JAK2 exon 12 mutation which was not detected during research.

### Biography

Mouhanad Adam has completed his MSc from Alzaiem Alazhari University. He is the Director of Alshamel Clinical Lab. He was awarded a Master's degree in Hematology and blood transfusion with grade very good. He is very interested in the molecular and hematological studies as general.

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