myelofibrosis or transform into acute myelogenous leukemia (AML) or also can mutation in our population in Sudan.

and platelets count as a diagnostic test for patients with polycythemia in Sudan necessary investigation in patients who have high level of hemoglobin and infarction. In the longer term, these disorders can develop secondary to use other treatment with veinsection and know the frequency of JAK2 with PV frequently demonstrate a complex of hematological disorder and (PV, Secondary, Apparent polycythemia or idiopathic erythrocytosis). Patients Many polycythemia's Sudanese patients who are currently on phlebotomy therapy without known what the actually causes of Raised Hemoglobin levels 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 have a PV and could have a JAK2 exon 12 mutation which was not detected during research.

Rational
Polycythemia and Jak2 Mutation

Erythrocytosis defined as an increase in red cells (or blood hemoglobin) per unit volume. Polycythemia Vera is a chronic myeloproliferative disease characterized by an absolute increase in red cell mass and reflects an increase in red blood cell (RBC) production. Since the red cell count given on the complete blood count (CBC) reflects a ratio of red cells per unit volume, it is possible to have an increase in the RBC count (or hemoglobin or hematocrit) due to either an increase in the red cell mass (polycythemia) or a decrease in the plasma volume (relative erythrocytosis). Polycythemia can be a response to some other condition causing hypoxemia (secondary polycythemia), can result from inappropriate erythropoietin production due to renal disease or some neoplasms, or can represent an autonomous neoplastic condition (polycythemia Vera). Polycythemia Vera (sometimes called polycythemia rubra Vera) is a chronic myeloproliferative disorder

In 1992, JAK2 was mapped on the short arm of chromosome 9q24 by Pritchard and his colleagues; it has 140 kb spanning 25 exons to form 1352 amino acid JAK2 protein.

In 2005, four separate groups using different modes of discovery and different measurement techniques reported the presence of a novel somatic point mutation in the conserved autoinhibitory pseudokinase domain of the gene coding for the JAK2 protein in patients with classic MPNs. The mutation was noted to cause a valine-to-phenylalanine substitution at amino acid position 617 (V617F). Loss of autoinhibitory function caused by JAK2V617F results in constitutive activation of the kinase and in recruitment and phosphorylation of downstream signaling molecules (Kitamura T, Health Organization criteria as one of the following: PV (99 cases) and idiopathic erythrocytosis (34 cases smokers and 7 non-smokers). There is no clinically significant difference between low group in hematological department except in platelet count which were give a significant difference (P=0.006).

At present, finding the JAK2 mutation is diagnostic for MPD, given the high specificity of the mutation for clonal myeloid diseases. The observation in this study shows that the V617F mutation alteration is seen in 54.4% of patients who satisfied the WHO criteria for polycythemia Vera Patients. Our results showed close agreement with other research in other countries; 69.6% Parma-Italy 2009, 26% Cambridge-UK 2005, 47% China 2014

Conclusion
While the JAK2 mutation is already a diagnostic value, it is reasonable to consider that in the near future, it will also play a huge role in the choice of treatment and monitoring of erythrocytosis by giving the development of drugs inhibiting the JAK2 pathway.

JAK2V617F mutation was reported in a large proportion of Sudanese patients; this should be considered when investigations of patients with erythrocytosis are carried out, and JAK2V617F inhibitors should be implemented for those with the mutation.

Polycythemia's patients with JAK2 V617F negative should tested for JAK2 Exon12 Mutation.

Sequence analysis for patients with positive V617F mutation which will show the mutant peak and determined the frequency of the heterozygous and homozygous allele in Sudan.

References


