Detection of the Janus Kinase 2 (*Jak-2*) V617F polymorphism in patients with polycythemia

***Ruluxy, P.**

Human Genetics Unit, Faculty of Medicine, Kynsey Road, Colombo 8.
*Corresponding E-mail: Ruluxypremendran@gmail.com

Polycythemia Vera (PV), primary polycythemia is a rare chromosome negative myeloproliferative neoplasm. A point mutation in the *JAK 2* gene, member of the tyrosine kinase family was recently identified and shown to be associated with several myeloproliferative disorders especially with Polycythemia Vera. PV is a stem cell disorder characterized by proliferation of hematopoietic cells. Several studies identified the *JAK 2* mutation resulting in the substitution of valine to phenylalanine at codon 617 (V617F). The main intention of this report is to detect the JAK -2- V617F polymorphism in a group of Polycythemia Vera patients. In order to that a simple and sensitive method was developed to detect the mutation via Polymerase Chain Reaction (PCR). Accordingly, 10 patients' DNA samples were selected, amplified using *JAK 2* PCR assay according to the conditions: 94 °C for 5 minutes; 25 cycles of 94 °C for 30 seconds, 55 °C for 30 seconds, and 72 °C for 30 seconds; and 72 °C for 7 minutes. Then 10 µl of each PCR product was loaded and visualized at 60V for 30 minutes. After the methodology was repeated twice, the results were compared using *JAK 2* positive and negative controls. It is concluded that according to the 10 patients' samples 30 % of the patients with polycythemia Vera have mutated *JAK-2* gene using the forward primer and reverse primer specific for mutant allele and forward primer of wild type.

Keywords: *JAK 2*, mutation, Polycythemia Vera, PCR, Polymorphism