ABSTRACT

Raisa Tahira Rudiyani Putri, 24020221140016. **Detection of Mutations in *JAK2* Exon 12 Gene with V617F Negatife in Blood Cancer Patients (Essential Thrombocytosis and Primary Myelofibrosis)** (under the guidance of Hermin Pancasakti Kusumaningrum *and* Farmaditya Eka Putra).

Essential thrombocytosis (ET) and primary myelofibrosis (PMF) belong to the myeloferative neoplasm (MPN) disease group, a rare chronic blood cancer disease caused by somatic mutations of the *JAK2*, *CALR*, *MPL*, or the *JAK2* exon 12 gene. However, research on *JAK2* exon 12 gene mutations in the Indonesian patient population, especially in the ET and MFP patient subgroups has not been found explicitly. The purpose of this study was to amplify the *JAK2* exon 12 gene in ET and MFP patients with negative *JAK2* V617F, detect the presence or absence of mutations in the *JAK2* exon 12 gene based on amplification results using Allele Specific-PCR (AS-PCR), and based on the results of sequencing analysis. The research samples were DNA from ET & PMF V617F mutation negative patients at CEBIOR Laboratory from 2012-2025, with a total of 98 samples. The research was conducted by testing the quality and quantity of DNA, amplifying the *JAK2* exon 12 gene using AS-PCR, visualizing the samples by electrophoresis, sequencing and analyzing the sequencing results. The results showed that, from the amplification and sequencing results of patient samples with the *JAK2* exon 12 gene, there were no samples that had mutations in exon 12. However, in the results of the reverse sequence alignment analysis showed that some samples had nucleotide insertions with the same number and location. This requires further research. It can be concluded that mutations in the JAK2 exon 12 gene with negative V617F were not found among 89 TE & MFP patient samples.

***Keyword**: Essential Trombocytosis, Exon 12, Janus Kinase 2 (JAK2), Primary Myelofibrosis*