INTISARI

SUPRIADI, A, 2018, DETEKSI MUTASI SEKUEN EKSON 1 GEN BETA GLOBIN PADA PASIEN TALASEMIA BETA MAYOR DI RSUD DR. SOEROTO NGAWI DENGAN METODE *POLYMERASE CHAIN REACTION SINGGLE STRAND CONFORMATION POLYMORPHISM*

Beta talasemia merupakan gangguan hematologis autosomal resesif yang secara genetis mengakibatkan berkurangnya sintesis beta globin di hemoglobin. Beta talasemia sebagian besar disebabkan mutasi titik, insersi atau delesi dalam gen beta globin yang terletak kromosom 11.

Penelitian ini merupakan penelitian deskriptif yang mengkaji tentang tipe dan letak mutasi pada gen beta globin individu talasemia beta mayor. Selain itu, penelitian ini juga mengkaji hubungan perubahan asam amino dengan struktur protein beta globin. Metode yang digunakan yaitu isolasi, *polymerase chain reaction-single stranded conformational polymorphism* (PCR-SSCP) dan sekuensing gen beta globin dari 9 individu talasemia beta mayor dengan menggunakan primer spesifik. Analisis data dilakukan dengan membandingkan hasil sekuensing dengan sekuen gen yang telah ada di Gene Bank menggunakan software genestudio dan Mega 6.0.

Hasil penelitian menunjukan mutasi pada ekson 1 gen beta globin terletak pada nukleotida 59 (Cd2) perubahan (CAT➔CAC) bertipe *silent mutation*, nukleotida ke 129 (Cd26) mutasi dari asam glutamat menjadi lisin (GAG➔AAG) tipe *missense mutation*, nukleotida ke 129 (Cd26) mutasi asam glutamat menjadi kodon stop (GAG➔TAG) tipe *nonsense mutation*, nukleotida ke 147 (IVS1-nt5) mutasi (G➔C) bertipe *splice site mutation*. Mutasi pada Cd 26 menyebabkan perubahan asam amino penyusun protein.

Kata kunci : talasemia beta mayor, ekson 1, deteksi mutasi, PCR-SSCP

ABSTRACT

SUPRIADI, A, 2018, DETECTION MUTATION OF EXON 1 THE BETA GLOBIN GENE IN BETA THALASSEMIA MAJOR PATIENTS IN RSUD DR. SOEROTO NGAWI WITH *POLYMERASE CHAIN REACTION SINGGLE STRAND CONFORMATION POLYMORPHISM* (PCR-SSCP) METHOD.

Beta-thalassaemia is an autosomal resesif haematological disorder resulting in a genetically deficient synthesis of the beta globin chain in haemoglobin. It is mostly caused by point mutations, a small deletions or insertions within the beta globin gene wich is located as a cluster on the short arm of chromosome 11.

This research is a descriptive study that examines the type and location of mutations in beta globin gene of thalassemia beta major patienes. In addition, this study also examines the relationship of amino acid changes in the protein structure of beta globin. Methods used are isolation, polymerase chain reaction-single stranded conformational polymorphism (PCR-SSCP) and beta globin gene sequencing of 9 samples thalassemia beta major patients using primer specific. Data analysis will be done by comparing the results of sequencing and the gene sequence that has existed in GeneBank using software GeneStudio and Mega 6.0.

The results showed that mutations in exon1 beta globin gene at nucleotide 59 (cd2) changes (CAT➔CAC) type silent mutations, at nucleotide 129 (Cd26) mutations from asam glutamate to lysine (GAG➔AAG) type missense mutation, at nuckleotide 129 (Cd26) mutation from asam glutamate to stop codon (GAG➔TAG) type nonsense mutation, and nuckleotide 147 (IVS1-nt5) mutations (G➔C) type splice site mutation. Mutation at Cd26 change amino acid in the protein structure of beta globin.

Key words : thalassaemia beta major, exon 1, detection mutation, PCR-SSCP