ANALISIS SEKUEN EKSON 2 GEN BETA GLOBIN PADA *CARRIER* BETA THALASSEMIA

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ABSTRAK

Thalassemia merupakan kelainan genetik autosomal resesif yang disebabkan oleh mutasi gen pembentuk hemoglobin. Mutasi dapat terjadi pada gen alpha (α) globin atau gen beta (β) globin. Jumlah penderita thalassemia di Indonesia semakin meningkat, dan diprediksi *carrier* thalassemia di Indonesia berkisar 6–10%. Pengetahuan tentang jenis mutasi diperlukan untuk mengetahui adanya hubungan dengan variasi klinis *carrier* β thalassemia, dan menjadi dasar dalam pengobatan secara genetik. Empat individu dengan mutasi pada ekson 2 gen β globin, yang terdeteksi dengan *polymerase chain reaction-single stranded conformational polymorphism* (PCR-SSCP), digunakan sebagai sampel dalam penelitian ini. Penelitian ini bertujuan untuk mengetahui jenis dan letak mutasi pada ekson 2 gen β globin pada *carrier* β thalassemia menggunakan metode sekuensing. Penelitian dilakukan dengan mengisolasi DNA darah subjek penelitian. Hasil isolasi DNA diamplifikasi menggunakan primer spesifik dengan metode PCR. Amplikon hasil PCR kemudian disekuensing menggunakan metode Sanger (*dideoxynucleotide chain terminator*) melalui Laboratorium *1st Base*. Analisis data dilakukan dengan *software* Chromas Lite 2.1 dan BioEdit 7.1.9. Hasil menunjukkan 2 subjek penelitian mengalami mutasi pada ekson 2 gen β globin, sedangkan 2 subjek penelitian lainya tidak ditemukan adanya mutasi pada ekson 2, dan justru ditemukan pada intron 1, yaitu mutasi substitusi tranversi pada nukleotida ke-5 (IVS1-5 (G→C)). Jenis mutasi yang ditemukan pada ekson 2 gen β globin adalah mutasi *frameshift* atau *nonsense*, akibat delesi nukleotida tunggal pada kodon 35 (Cd 35 (del C)).

Kata kunci: β thalassemia, mutasi, ekson 2, gen β globin

SEQUENCE ANALYSIS OF THE SECOND EXON BETA GLOBIN GENE IN BETA THALASSEMIA CARRIER

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ABSTRACT

Thalassemia is an autosomal recessive genetic disorder caused by mutations in the genes which form hemoglobin. Mutations can occur in alpha (α) globin gene or beta (β) globin gene. The number of patients with thalassemia in Indonesia is increasing, and it is predicted that around 6-10% of the population are carriers of thalassemia in Indonesia. Knowledge of the types of mutations is needed to determine their relationship with clinical variation in β thalassemia carriers and the basis for genetic treatment. Four individuals with mutations in the second exon β globin gene, which is detected by polymerase chain reaction-single stranded conformational polymorphism (PCR-SSCP), are used as a sample in this research. The purpose of this research was to determine the type and location of mutations in the second exon β globin gene in β thalassemia carriers using sequensing method. The research was conducted by isolating DNA from the research subject's blood. The isolated DNAs were amplified using specific primers with the PCR method. The PCR products were sequenced using Sanger method (*dideoxynucleotide chain terminator*) through the 1st Base Laboratory. Data analysis was performed by Chromas Lite 2.1 and BioEdit 7.1.9 software. The results showed that two research subjects had a mutation in the second exon β globin gene, while the other two research subjects did not reveal any mutations in the second exon, and it was found in the first intron, which is a substitutions transversion mutation at nucleotide 5th (IVS1-5 (G → C)). The type of mutation found in the second exon β globin gene is a frameshift or nonsense mutation resulting from the deletion of a single nucleotide at codon 35 (Cd 35 (del C)).

Keywords: β thalassemia, mutation, β globin, second exon