DETEKSI MUTASI INTRON 1 IVS-1 NT 5 (G>C) GEN PENGKODE β-GLOBIN PADA *CARRIER* β-THALASSEMIA DENGAN PCR-RFLP

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INTISARI

Penelitian ini dilakukan untuk mengetahui keberadaan mutasi IVS-1 nt 5 pada sejumlah sampel darah yang secara hematologis didiagnosis sebagai *carrier* β-thalassemia. *Carrier* β-thalassemia adalah suatu penyakit herediter yang tidak dapat diamati dengan kenampakan luar karena pada kondisi heterozigot bersifat asimtomatik. Dalam pendataan sampel darah *carrier* β-thalassemia, peneliti bekerja sama dengan POPTI dan Laboratorium Klinik Prodia untuk melihat data hematologis setiap sampel darah seperti hematologi rutin (*Cell Blood Count, CBC*), nilai indeks korpuskular (Mean Corpuscular, MC), analisis hemoglobin dengan metode *High Performance Liquid Chromatography* (HPLC), dan gambaran darah tepi. Pada penelitian ini digunakan sampel berupa *frozen blood* 9 individu yang berdasarkan analisis hematologis terdiagnosis sebagai *carrier* β-thalassemia. DNA sampel darah diisolasi untuk selanjutkan dilakukan amplifikasi DNA sekuen yang spesifik menggunakan set primer spesifik sehingga menghasilkan amplikon berukuran 293 bp (*basepairs*). Selanjutnya DNA hasil amplifikasi tersebut diinkubasi dengan enzim restriksi *Cac*8I pada suhu 37^0C selama 6 jam di dalam *waterbath*. Interpretasi data dilakukan dengan membandingkan perbedaan ukuran dan jumlah fragmen digesti pada sampel normal (kontrol negatif) dengan sampel *carrier* β-thalassemia serta membandingkan persamaan ukuran dan jumlah fragmen antara sampel kontrol positif (mutasi IVS-1 nt 5) dengan sampel *carrier* β-thalassemia. Hasil penelitian menunjukkan 6 dari 9 individu *carrier* β-thalassemia dideteksi mengalami mutasi IVS-1 nt 5 dengan PCR-RFLP *(Polymerase Chain Reaction-Restriction Fragment Length Polymorphism)*. Sehingga dapat disimpulkan bahwa dengan PCR-RFLP dapat dideteksi terdapat 6 dari 9 individu *carrier* β-thalassemia mengalami mutasi pada IVS-1 nt 5 (G>C).

Kata kunci : β-thalassemia, RFLP, mutasi, enzim restriksi, hematologis

DETECTION MUTATION INTRON 1 IVS-1 NT 5 (G>C) IN β-GLOBIN ENCODING GENE OF *CARRIER* β-THALASSEMIA WITH PCR-RFLP

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ABSTRACT

This study was conducted to determine the presence of mutations IVS-1 nt 5 in a number of blood samples haematologically diagnosed as a carrier β-thalassemia. Carrier β-thalassemia is a hereditary disease whose characteristics can not be observed with outside appearance because the condition of heterozygotes is asymptomatic. In data collection of blood samples diagnosed with β-thalassemia carrier, the researchers collaborated with POPTI and Prodia Clinical Laboratory to view hematological data of each blood sample such as routine hematologic (Cell Blood Count, CBC), corpuscular index value (Mean Corpuscular, MC), hemoglobin analysis with methods of High Performance Liquid Chromatography (HPLC), and peripheral blood images. This study used a sample of frozen blood 9 individuals based on hematologic analysis which diagnosed as a carrier β-thalassemia. The DNA of blood sample was isolated for further specific DNA sequence amplification using a specific primer set to produce a 293 bp amps (basepairs). Then the amplified DNA was incubated with restriction enzyme Cac8I at 37 ^{0}C for 6 hours in the waterbath. Interpretation of data was done by comparing the difference in size and number of digestive fragments in normal / negative control samples with samples diagnosed with β-thalassemia carrier and comparing the equation of size and number of fragments between positive control samples of mutation IVS-1 nt 5 with the samples diagnosed with β-thalassemia carrier. The results showed 6 of 9 individuals diagnosed with β-thalassemia carrier were detected by IVS-1 nt 5 mutation by PCR-RFLP (Polymerase Chain Reaction-Restriction Fragment Length Polymorphism) technique. So it can be concluded that 6 of 9 individual carrier β-thalassemia have mutation in intron one that is IVS-1 nt 5.

Keywords: β-thalassemia, RFLP, mutation, restriction enzyme, hematologic