DETEKSI MUTASI IVSI-5 (G>C) DAN IVSI-1 (G>T) GEN PENGKODE β-GLOBIN PADA PASIEN β-THALASSEMIA DI RSUD TIDAR KOTA MAGELANG

Nafis Muhimmatul 'Ulya
18/423357/BI/09991

Dosen Pembimbing: Dr. Niken Satuti Nur Handayani, M.Sc.

INTISARI

Thalassemia merupakan salah satu penyakit hemolitik herediter yang disebabkan kurangnya atau tidak adanya sintesis rantai globin sebagai penyusun utama struktur molekul hemoglobin. Rendahnya sintesis rantai globin diakibatkan oleh mutasi atau delesi pada gen α-globin atau β-globin. Mutasi pada gen α-globin disebut dengan α-Thalassemia, sedangkan mutasi pada gen β-globin disebut β-thalassemia. Kelompok gen β-globin terletak pada lengan pendek kromosom 11. Mutasi IVSI-5 (G>C) dan IVSI-1 (G>T) merupakan mutasi gen β-globin yang umum ditemukan di etnik Jawa-Sunda. Tujuan penelitian ini adalah mendeteksi dan mengetahui persentase jenis mutasi IVSI-5 (G>C) dan IVSI-1 (G>T) pada 61 sampel darah pasien β-thalassemia di RSUD Tidar Kota Magelang yang melakukan transfusi rutin. Metode deteksi yang digunakan adalah *Amplification Refractory Mutation System-Polymerase Chain Reaction* (ARMS-PCR). Hasil penelitian menunjukkan jenis mutasi IVSI-5 (G>C) dan IVSI-1 (G>T) dapat dideteksi pada gen β-globin sampel darah pasien β-Thalassemia yang diteliti. Mutasi tersebut diperoleh sebesar 60,65% dan 13,11%.

Kata Kunci: β-thalassemia, β-globin, IVSI-5 (G>C), IVSI-1 (G>T), ARMS-PCR

DETECTION OF IVSI-5 (G>C) AND IVSI-1 (G>T) MUTATIONS IN β-GLOBIN CODING GENE OF β-THALASSEMIA PATIENTS AT TIDAR MAGELANG CITY HOSPITAL

Nafis Muhimmatul 'Ulya
18/423357/BI/09991

Supervisor: Dr. Niken Satuti Nur Handayani, M.Sc.

ABSTRACT

Thalassemia is a hereditary hemolytic disease caused by a deficiency or no synthesis of globin chains as the main constituent of the molecular structure of hemoglobin. Low globin chain synthesis results from mutations or deletions in the α-globin or β-globin genes. Mutations in the α-globin gene are called α-thalassemia, while mutations in the β-globin gene are called β-thalassemia. The β-globin gene cluster is located in the short arm of chromosome 11. IVSI-5 (G>C) and IVSI-1 (G>T) mutations are common β-globin gene mutations found in the Javanese-Sundanese ethnicity. The purpose of this study was to detect and determine the percentage of IVSI-5 (G>C) and IVSI-1 (G>T) mutations in 61 blood samples of β-thalassemia patients at Tidar Magelang City Hospital who have routine transfusions. The Amplification Refractory Mutation System-Polymerase Chain Reaction (ARMS-PCR) is the detection method. The results showed that IVSI-5 (G>C) and IVSI-1 (G>T) mutations could be detected in the β-globin gene of blood samples of β-thalassemia patients studied. The mutations obtained were 60.65% and 13.11%, respectively.

Keyword: β-thalassemia, β-globin, IVSI-5 (G>C), IVSI-1 (G>T), ARMS-PCR