DETEKSI LIMA MUTASI UMUM GEN PENGKODE GLOBIN ALFA DENGAN *MULTIPLEX PCR*

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ABSTRAK

α-thalassemia merupakan salah satu kelainan genetik hemoglobin yang disebabkan oleh penurunan produksi rantai α-globin akibat delesi atau mutasi satu atau lebih dari empat gen α-globin yang terletak pada kromosom 16. Terdapat lima macam mutasi umum penyebab α-thalassemia yang paling sering dijumpai di populasi Asia Tenggara, yaitu mutasi delesi yang terdiri dari $-\alpha^{3,7}$, $-\alpha^{4,2}$, dan $-^{SEA}$ serta mutasi non-delesi yang terdiri dari Hb Adana dan Hb CS. Pada penelitian ini dilakukan deteksi terhadap lima macam mutasi tersebut dengan metode *Multiplex PCR* pada 305 subyek yang merupakan populasi umum Daerah Istimewa Yogyakarta. Hasil penelitian menunjukkan bahwa ditemukan delesi $-\alpha^{3,7}$ sebanyak 14 dari 305 subyek (4,6%), pada non-delesi Hb Adana sebanyak 2 dari 305 subyek (0,7%) sementara delesi $-\alpha^{4,2}$, $--^{SEA}$, dan non-delesi Hb CS tidak ditemukan pada populasi umum Daerah Istimewa Yogyakarta.

Kata kunci : α-thalassemia, mutasi umum, *Multiplex PCR*

DETECTION ON FIVE COMMON MUTATIONS ON ALPHA GLOBIN CODING GENE BY USING MULTIPLEX PCR

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ABSTRACT

α-thalassemia is one of the genetic hemoglobin disorders caused by decreased production of α-globin chains due to the deletions or mutations of one or more of four α-globin genes located on chromosome 16. There are five common mutations that are frequently found in the Southeast Asian population, namely deletion mutations consisting of $-\alpha^{3.7}$, $-\alpha^{4.2}$, and $--^{SEA}$ and non-deletion mutations consisting of Hb Adana and Hb CS. In this study, the detection of these five mutations was performed using Multiplex PCR method on 305 subjects who represent the general population of the Special Region of Yogyakarta. The results show that deletion $-\alpha^{3.7}$ was found in 14 out of 305 subjects (4.6%), while non-deletion Hb Adana was found in 2 out of 305 subjects (0.7%). However, deletion $-\alpha^{4.2}$, $--^{SEA}$, and non-deletion Hb CS mutations were not found in the general population of the Special Region of Yogyakarta.

Keywords : α-thalassemia, mutation, Multiplex PCR