Gambaran Hematologi Thalassemia Alfa Delesi satu gen pada mahasiswa Fakultas Kedokteran Universitas Indonesia = One gene Deletion Alpha Thalassemia Hematology profile in Faculty of Medicine University of Indonesia students / Nenny Puspandari

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Abstrak

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Thalassemia adalah kelainan herediter yang disebabkan oleh mutasi pada gen globin dan memiliki fenotip yang bervariasi. Terdapat empat fenotip thalassemia,
yaitu silent carrier yang gambaran hematologinya dapat normal, -thalassemia trait dengan gambaran hematologi yang mungkin mikrositik, penyakit HbH dan Hb Bart’s
hydrops fetalis. Tujuan penelitian ini adalah untuk mendapatkan gambaran hematologi thalassemia delesi satu gen pada pemeriksaan penapisan thalassemia
pada 266 mahasiswa FKUI. Dilakukan deteksi delesi satu gen globin menggunakan polymerase chain reaction multipleks, sedangkan kadar HbA2 diperiksa dengan
metode high performance liquid chromatography. Didapatkan 10 (3,7%) subjek mengalami delesi satu gen globin , dan seluruhnya berupa delesi 3,7 kb heterozigot.
Gambaran hematologi dari subjek penelitian yang mengalami delesi satu gen didapatkan kadar hemoglobin normal berkisar 12,0 – 14,6 g/dL, nilai VER bervariasi
berkisar 75,5 – 82,8 fL, nilai HER 26,0 – 28,0 pg, nilai RDW 13,8 – 23,7%.
Morfologi eritrosit didapatkan 67% mikrositik hipokrom dan kadar HbA2 dalam batas normal dengan rentang 2,6 – 3,1 %.
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ABSTRACT
Alpha thalassemia is a hereditary disorder caused by alpha globin gene mutation and has various phenotype. There are four clinical phenotypes of alpha thalassemia, silent carrier which may have normal hematology parameters, -thalassemia trait which have only microcytic hypochromia, HbH disease and Hb Bart’s hydrops fetalis. The aim of this study is to get hematology profile of one gene deletion alpha thalassemia
in the thalassemia trait screening in 266 students of faculty of medicine UI. Detection of one gene deletion was investigated in these subjects by multiplex polymerase chain reaction. HbA2 was measured using high performance liquid chromatography. There are 10 (3.7%) subject who have one gene deletion, and all of the mutation are heterozigous 3.7 kb deletion. Hematology profile of one gene deletion subject in this study, hemoglobin are normal with the range 12,0 – 14,6 g/dL, MCV 75,5 – 82,8 fL, MCH 26,0 – 28,0 pg, RDW 13,8 – 23,7%. Erytrocyte morphology 67% microcytic hipochromic and HbA2 value 2,6 – 3,1 %.