PREDIKSI STRUKTUR 3 DIMENSI DAN ANALISIS *IN SILICO* PROTEIN AKIBAT DELESI $\alpha^{4,2}$ PADA GEN α-GLOBIN

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INTISARI

Thalassemia merupakan kelainan genetik diakibatkan oleh mutasi pada gen globin. Mutasi delesi $\alpha^{4,2}$ merupakan mutasi *silent trait*, namun pada penelitian ini nilai hematologis MHC dan MCV lebih rendah dibandingkan dengan kisaran normal serta terdapat abnormalitas morfologi eritrosit. Tujuan penelitian ini adalah menentukan titik potong delesi $\alpha^{4,2}$, memprediksi dan menganalisis efek mutasi delesi $\alpha^{4,2}$ terhadap protein hemoglobin dikaitkan dengan gangguan fisiologi penderita. DNA sampel darah disekuensing menggunakan metode Sanger. Proses *alignment* dilakukan dengan *Clustal Omega*, prediksi dan analisis struktur hemoglobin dilakukan dengan *CHIMERA* dan *Discovery Studio*. Titik potong delesi $\alpha^{4,2}$ berada diantara basa nomor 26 sekuens *box* X2 dan basa nomor 347 sekuens *box* X1. Delesi $\alpha^{4,2}$ memiliki struktur protein yang sama dengan protein normal baik residu ataupun seluruh ikatan penstabil protein. Nilai MCH dan MCV yang rendah serta abnormalitas morfologi eritrosit merupakan konsekuensi kurangnya produksi subunit α yang diakibatkan mutasi. Hasil analisis menyimpulkan delesi 1 gen yakni $\alpha^{4,2}$ tetap menghasilkan struktur hemoglobin yang normal dengan fungsi yang normal.

Kata kunci : eritrosit, hemoglobin, delesi $\alpha^{4,2}$, struktur protein, patologi thalassemia

THE THREE DIMENSIONAL STRUCTURE PREDICTION AND IN SILICO ANALYSIS OF PROTEIN DUE TO DELETION OF $\alpha^{4.2}$ IN α-GLOBIN GENE

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ABSTRACT

Thalassemia is a genetic disorder caused by mutations in the globin gene. The $\alpha^{4.2}$ deletion mutation is a silent trait mutation, but in this study the hematological value of MHC and MCV is lower than the normal range and there are morphological abnormalities of erythrocytes. The aim of this study is to determine the breakpoint of $\alpha^{4.2}$ deletion, predict and analyze the effect of the $\alpha^{4.2}$ deletion mutation on the hemoglobin protein associated with physiological disorders of the patient. DNA of blood sample was sequenced by using the Sanger method. The alignment process was carried out with Clustal Omega, prediction and analysis of the structure of hemoglobin is done by CHIMERA and Discovery Studio. The breakpoint for deletion $\alpha^{4.2}$ was found in base number 26 of box X2 sequence and base number 347 of box X1 sequence. The $\alpha^{4.2}$ deletion showed the same protein structure as a normal protein either residue or all protein stabilizing bonds. Low MCH and MCV values and morphological abnormalities of erythrocytes were a consequence of the lack of α subunit production caused by mutations. The results of the analysis concluded that deletion of 1 gene, $\alpha^{4.2}$, produces a normal hemoglobin structure with normal function.

Keywords: erythrocyte, hemoglobin, $\alpha^{4.2}$ deletion, protein structure, pathology of thalassemia