# ANALISIS *IN SILICO* STRUKTUR TIGA DIMENSI PROTEIN CD26/IVS I-5 PADA HbE/β-THALASSEMIA

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## INTISARI

β-thalassemia adalah kelainan genetik autosomal resesif yang disebabkan oleh terganggunya sintesis protein HBB atau β-globin. HbE/β-thalassemia disebabkan oleh *co-inheritance* varian hemoglobin E dan β-thalassemia. Hemoglobin E disebabkan oleh adanya subtitusi basa guanin (G) menjadi adenin (A) pada ekson 1 kodon 26 gen β-globin. Mutasi penyebab β-thalassemia yang paling umum berasosiasi dengan HbE adalah IVS I-5 (G>C) (Hernaningsih *et al*., 2022). Mutasi IVS I-5 (G>C) disebabkan transversi basa guanin (G) menjadi sitosin (C) pada nukleotida 5 intron 1 gen β-globin. Mutasi tersebut mengakibatkan terjadinya kesalahan pada *splicing* RNA dan menyebabkan terbentuknya tiga mRNA dewasa. Penelitian ini dilakukan dengan memprediksi protein HBB yang diproduksi oleh pasien HbE/β-thalassemia serta mengetahui pengaruhnya terhadap nilai hematologi dan frekuensi transfusi darah. Penelitian dilakukan secara *in silico* dengan memanfaatkan ColabFold, Chimera 1.17.3, dan Biovia Discovery Studio. Hasil penelitian menunjukkan terdapat tiga jenis protein HBB (HBB normal, HBB Cd 26, dan HBB IVS I-5) pada pasien HbE/β-thalassemia. Ketiga protein tersebut memiliki afinitas interaksi dengan protein HBA berbeda-beda. Keragaman afinitas interaksi ini memiliki pengaruh terhadap keberagaman nilai hematologi pasien HbE/β-thalassemia. Akan tetapi tidak ditemukan korelasi antara protein HBB mutan dengan frekuensi transfusi darah.

**Kata kunci**: β-thalassemia, HbE, 3D protein, *in silico*

# IN SILICO ANALYSIS OF THREE DIMENSIONAL STRUCTURE PROTEIN CD26/IVS I-5 IN HbE/β-THALASSEMIA

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## *ABSTRACT*

β-thalassemia is an autosomal recessive genetic disorder caused by decreased or absent synthesis of HBB or β-globin protein. HbE/β-thalassemia is caused by the co-inheritance of hemoglobin E and β-thalassemia. Hemoglobin E is caused by a subtitution of guanine (G) to adenine (A) in exon 1 codon 26 of the β-globin gene. The most common mutation causing β-thalassemia associated with HbE is IVS I-5 (G>C) (Hernaningsih et al., 2022). The IVS I-5 mutation (G>C) is caused by the transversion of a guanine base (G) to cytosine (C) at fifth nucleotide intron 1 β-globin gene. This mutation results in an error in RNA splicing and causes the formation of three mature mRNAs. This research aims to predict the HBB protein produced by people with HbE/β-thalassemia and know its effect on hematological values and frequency of blood transfusions. This research was carried out by in silico method using ColabFold, Chimera 1.17.3, and Biovia Discovery Studio. The results showed three types of HBB protein (normal HBB, HBB Cd 26, and HBB IVS I-5) in people with HbE/β-thalassemia. These three proteins have different interactions with the HBA protein. This variation in interaction affinity affects the diversity of hematological values in people with HbE/β-thalassemia. However, there is no correlation between the mutant HBB protein and the frequency of blood transfusions.

**Keyword**: β-thalassemia, HbE, 3D protein, *in silico*