GENOTYPE-PHENOTYPE CORRELATION OF BETA THALASSEMIA PATIENTS IN THE CITY OF SAMARINDA, EAST KALIMANTAN 2019

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Abstract	Thalassemia is a genetic blood disorder that is autosomal recessive and is quite common throughout the world. This study aims to determine the relationship of Hemoglobin beta (HBB) gene mutations types with clinical levels and hematological in the subjects of 31 thalassemia-beta patients in Samarinda City. Blood samples were taken from patients to obtain their DNA then amplified them with the Polymerase Chain Reaction and direct sequencing techniques to analyze the hemoglobin-beta gene mutation. Javanese ethnics is the most dominant in this study (64.5%) and the most common clinical levels is the moderate category (77.4%). The mean MCV and MCH values were 72 +/- 5,5 fL and 24 +/- 3,3 pg. DNA analysis found 8 types of mutant alleles including 48.4% of Cd26 / HbE (GAG>AAG), 14.5% of IVS-1-5 (G>C) 12.9% of IVS-1-2 (T>C,,8.1% of Cd35 (-C), 6.5% of IVS-1-1 (G>T) 3.2% of Cd30 (AGG>ACG), Cd60 (GTG>GAG) and Cd2 (CAT>CAC) are 1.6% each. This study found mutations that had not been previously reported in Indonesia, namely Cd60 (GTG>GAG) and Cd2 (CAT>CAC). Spearman rank statistical tests show there is no significant relationship between the two studied variables.
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