Molecular Scanning of Î²-Thalassemia in the Southern Region of Central Java, Indonesia; a Step Towards a Local Prevention Program

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Abstract	Thalassemia is the most prevalent genetic blood disorder worldwide, and particularly prevalent in Indonesia. The purpose of this study was to determine the spectrum of beta-thalassemia (beta-thal) mutations found in the southern region of Central Java, Indonesia. The subjects of the study included 209 b-thal Javanese patients from Banyumas Residency, a southwest region of Central Java Province. DNA analysis was performed using polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP), amplification refractory mutation system (ARMS), and the direct sequencing method. The results showed that 14 alleles were found in the following order: IVS-I-5 (G > C) (HBB: c.92 + 5G > C) 43.5%, codon 26 (Hb E; HBB: c.79G > A) 28.2%, IVS-I-1 (G > A) (HBB: c.92 + 1G > A) 5.0%, codon 15 (TGG > TAG) (HBB: c.47G > A) 3.8%, IVS-I-1 (G > T) (HBB: c.92 + 1G > T) 3.1%, codon 35 (-C) (HBB: c.110delC) 2.4%. The rest, including codons 41/42 (-TTCT) (HBB: c.126_129delCTTT), codons 8/9 (+G) (HBB: c.27_28insG), codon 19 (AAC > AGC) (HBB: c.59A4G), codon 17 (AAG > TAG) (HBB: c.52A > T), IVS-I-2 (T > C) (HBB: c.92 + 2T > C), codons 123/124/125 (-ACCCCACC) (HBB: c.370_378delACCCCACCA), codon 40 (-G) (HBB: c.123delG) and Cap +1 (A > C) (HBB: c.-50A > C), accounted for up to 1.0% each. The most prevalent alleles would be recommended to be used as part of b-thal screening for the Javanese, one of the major ethnic groups in the country.
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