Cd60 (GTG > GAG)/Hb Cagliari mutation was found in scanning of Î²-thalassemia alleles from patients of East Kalimantan, Indonesia

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Journal Name	MOLECULAR GENETICS AND METABOLISM REPORTS
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Abstract	a:4:{i:0;s:315:"Background and purpose: Thalassemia is a genetic disorder with a fairly high prevalence worldwide. Three to 10% of Indonesian people are estimated to be carriers for thalassemia. This study was intended to figure out the spectrum of genetic mutations of patients with thalassemia in Samarinda City, East Kalimantan.";i:1;s:332:"Methods: The research subjects consisted of 31 beta-thalassemia patients registered with the Association of Thalassemia Patients' Parents (POPTI) of Samarinda. DNAs were extracted from the patients' blood samples then amplified by the direct sequencing technique with polymerase chain reaction to analyze beta-globin gene mutations.";i:2;s:502:"Result: The study results show that the male/female ratio was 51.6%:48.4%, the patients' ages ranged from 4 years to 56 years with an average age of 14 years, and the dominant ethnic group was Javanese (64.5%). The DNA analysis yielded 7 types of mutant alleles, namely Cd26/HbE (GAG > AAG) at 48.4%, IVS-1-5 (G > C) at 14.5%, IVS-1-2 (T > C) at 12.9%, Cd35 (-C) at 8.1%, IVS-1-1 (G > T) at 6.5%, and, the least frequently encountered mutant alleles, Cd30 (AGG > ACG) and Cd60 (GTG > GAG) each at 3.2%.";i:3;s:90:"Conclusion: This study discovered unreported mutant in Indonesia, namely Cd60 (GTG > GAG).";}
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