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University Malaysia Sarawak

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Transfusion-Dependent Thalassemia in Northern Sarawak: A Molecular Study to Identify Different Genotypes in the Multi-Ethnic Groups and the Importance of Genomic Sequencing in Unstudied Populations

Jin-Ai M.A. Tan[a] Saw-Sian Chin[b] Gek-Bee Ong[e] Mohamed N. Mohamed Unni[f] Ashley E.R. Soosay[c] Henry R. Gudum[d] Siew-Leng Kho[a] Kek-Heng Chua[a] Jang J. Chen[a] Elizabeth George[g]

[a]Department of Biomedical Science, Faculty of Medicine, University of Malaya, Kuala Lumpur, [b]Institute of Social Informatics and Technological Innovations, Departments of [c]Paraclinical Sciences, and [d]Pathology, Faculty of Medicine and Health Sciences, Universiti Malaysia Sarawak, and [e]Department of Paediatrics, Sarawak General Hospital, Kuching, [f]Department of Paediatrics, Hospital Miri, Miri, and [g]Department of Pathology, Faculty of Medicine and Health Sciences, Universiti Putra Malaysia, Serdang, Malaysia

Key Words

Chinese · Malays · Gene mutations · Indigenous groups · Sarawak · Thalassemia

Abstract

Background: Although thalassemia is a genetic hemoglobinopathy in Malaysia, there is limited data on thalassemia mutations in the indigenous groups. This study aims to identify the types of globin gene mutations in transfusion-dependent patients in Northern Sarawak. ***Methods:*** Blood was collected from 32 patients from the Malay, Chinese, Kedayan, Bisayah, Kadazandusun, Tagal, and Bugis populations. The α- and β-globin gene mutations were characterized using DNA amplification and genomic sequencing. ***Results:*** Ten β- and 2 previously reported α-globin defects were identified. The Filipino β-deletion represented the majority of the β-thalassemia alleles in the indigenous patients. Homozygosity for the deletion was observed in all Bisayah, Kadazandusun and Tagal patients. The β-globin gene mutations in the Chinese patients were similar to the Chinese in West Malaysia. Hb Adana (HBA2:c.179G>A) and the $-\alpha^{3.7}/\alpha\alpha$ deletion were detected in 5 patients. A novel 24-bp deletion in the α2-globin gene (HBA2:c.95 + 5_95 + 28delGGCTCCCTCCCCTGCTCCGACCCG) was identified by sequencing. Co-inheritance of α-thalassemia with β-thalassemia did not ameliorate the severity of thalassemia major in the patients. ***Conclusion:*** The Filipino β-deletion was the most common gene defect observed. Homozygosity for the Filipino β-deletion appears to be unique to the Malays in Sarawak. Genomic sequencing is an essential tool to detect rare genetic variants in the study of new populations.

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E-Mail karger@karger.com
www.karger.com/phg

Jin-Ai M.A. Tan
Department of Biomedical Science, Faculty of Medicine
University of Malaya
Kuala Lumpur 50603 (Malaysia)
E-Mail maryanne@um.edu.my