FREQUENCY OF ALPHA-THALASSAEMIA IN THE ORANG SUNGAI AND BISAYA POPULATIONS IN EAST MALAYSIA

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Alpha-thalassaemia is an inherited autosomal recessive disorder which is commonly caused by deletions of the α-globin genes. As each individual possess four α-globin genes, the severity of α-thalassaemia therefore depends on the number of α-globin genes deleted. The common and rare mutations responsible for thalassaemia in the Malays and Chinese in West Malaysia have been well studied. However, there is a paucity of information in the extent of thalassaemia in East Malaysia. About 47.4% of the population of Sabah belong to one of 39 different indigenous groups. There are 28 different indigenous groups in Sarawak and they comprise about 50% of the population. This study aims to determine the types and frequency of α-thalassaemia in the Orang Sungai and Bisaya indigenous groups in Sabah and Sarawak respectively. Molecular analysis for deletional α-thalassaemia (-SEA, -α^3.7 and -α^4.2 deletions) were carried out using gap-PCR. Non-deletional α-thalassaemia - Haemoglobin Quong Sze (HbQS) and Haemoglobin Constant Spring (HbCS) - was determined by Amplification Refractory Mutation System (ARMS). The results showed a high frequency of α-thalassaemia in the Orang Sungai (43.8%) and Bisaya (38.5%). Alpha-thalassaemia in both groups was caused by the single -α^3.7 deletion. The SEA deletion (-SEA) was absent in the Bisaya group but confirmed in one Orang Sungai individual (1.6%; 1/64). The single leftward -α^4.2 deletion, HbCS and HbQS were absent in both populations. In conclusion, although the Orang Sungai and Bisaya are at little risk for the fatal condition, Bart’s hydrops foetalis, a high frequency of these people are α-thalassaemia carriers.