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GENETIC ORIGINS AND CLINICAL DESCRIPTION OF SICKLE CELL DISEASE IN SRI LANKA

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ABSTRACT

Sickle cell disease (SCD) is found at a low prevalence in Sri Lanka. A recent hospital based survey identified about 50 patients in the country. The clinical spectrum of SCD in Sri Lanka has not been studied and its genetic origin remains unknown. This study was conducted to identify the genetic origin (s) of Sickle haemoglobin (Hb S) and to carry out a clinical description of SCD in Sri Lankan patients. Patients were recruited from Ragama, Anuradhapura, Hambantota and Kurunegala thalassaemia centres. All patients were examined and clinical details were recorded. Genetic analyses were performed to identify the haplotype of HbS, *Xmn I* polymorphism, α plus thalassaemia and β-mutations. A total of 49 SCD patients were studied. Ages ranged from 5 – 47 years (mean 20.4). 45 (91.8%) patients were Sinhalese and the rest were Muslims. 43 (87.7%) patients had sickle- β-thalassaemia (SBT) and 6 were homozygous (HbSS). Joint pain was the commonest symptom among SCD patients. Clinical presentations in SBT varied from those with none to frequent crises. Clinical management varied with only 21 (42.86%) patients being on hydroxyurea. Most patients had not inherited the known disease ameliorating genetic factors with 47 (95.9%) patients not having α plus thalassaemia and 44 (89.8%) patients not having *Xmn I* polymorphism at (+/+) framework. Three Sickle haplotypes were identified including; Arab-Indian, Benin and Bantu. There appears to be at-least three genetic origins of HbS in Sri Lanka. SCD is extremely clinically variable in Sri Lanka. The reason for this variation needs further study, as most patients seem not to have common inherited disease ameliorating modifiers.

Keywords: Sickle, Sri Lanka, Haplotype, Origin, Clinical