# The Genetic Origins and Molecular Characterization of Sickle Cell Disease in Sri Lanka

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**Background**: Sickle cell disease (SCD) is globally the commonest monogenic disease. Although the incidence is not as common as in India, it is found in Sri Lanka too. A recent hospital based survey identified around 60 patients in the country but no detailed study of SCD have been done to-date. The genetic origin of Haemoglobin (Hb) S found in Sri Lanka is not yet known.

**Objective:** To determine the genetic origin and characterize genetic modifiers such as α+ thalassaemia, β- thalassaemia mutations and *Xmn I* polymorphism of SCD among Sri Lankan patients.

**Methodology**: Patients were recruited from Mahara (Ragama) and Kurunegala thalassaemia centers as part of an ongoing study. Capillary electrophoresis technique was used to identify and quantify Hb S and sickling test too was subsequently done. RFLP technique was used to determine the Sickle cell haplotype and *XmnI* polymorphism. Gap PCR and ARMS techniques were used to characterize common α gene deletions and β mutations respectively.

**Results:** A total of 25 SCD patients have been molecularly characterized to date. Majority of the SCD subjects were compound heterozygotes with β-thalassaemia and two were homozygous (HbSS). Mean Hb concentration was 8.4 g/dl (8.0-8.8: 95%CI) while the mean Hb S percentage was 63.6 (58.2-69.0: 95% CI). β mutation studies showed 60.8% of S-β thalassaemia patients had β+ severe type whereas, the rest had $\beta^0$ type. Common α+ gene deletions were not detected except for two cases with 3.7 kb deletions. None of the patients were +/+ for the *Xmn 1* polymorphism.

Twenty two patients were heterozygous for Arab-Indian haplotype while one patient was heterozygous for Benin haplotype. Two homozygous cases were homozygous for Benin haplotype. Xmn I polymorphism of Gγ gene was heterozygous in all Arab-Indian cases while it was negative in all the Benin cases.

**Conclusion**: There appears to be at least two genetic origins of Hb S in Sri Lanka. (Indian-Arab and the Benin). Most patients seem not to have inherited disease ameliorating genetic modifiers.

**Keywords:** Sickle cell, Origin, Haplotype, genotype, Sri Lanka

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