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THE AMERICAN SOCIETY OF HEMATOLOGY
26th Annual Meeting

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DELETIONS AND DUPLICATIONS OF α OR ζ GLOBIN GENES IN CHILDREN WITH SICKLE CELL ANEMIA.

A.E. Felice*, V.C. McKie*, K. McKie* and T.H.J. Huisman. Comp. Sickle Cell Center, Depts. of Cell & Molecular Biology and Pediatrics, Medical College of Georgia and Medical Research Service, V.A. Medical Center, Augusta, GA.

DNA samples from 345 Black children, aged 0-18 years with SS, SC and Sβ+Thal were tested with restriction endonucleases and α or ζ globin DNA probes. A -3.7 kb type of α-Thal-2 (or -α/) was found in 34% with -α/αα and 4% with -α/-α (gene frequency = 0.20). None had the -4.2 kb type of α-Thal-2. The incidence of α-Thal-2 was the same among SS, SC and Sβ+Thal patients. It did not vary among children and adolescents of different ages in the first two decades of life. Hematological effects depended on the age of the patient. Anemia was equal in severity up to the age of 6 yrs but older children with α-Thal-2 had higher Hb levels. The MCV and RBC values were typical of thalassemia since birth. Hb F levels were lower in SS/SC patients with α-Thal-2. The triplicated α-globin genotype was found in only one SS patient. A 10.4 kb deletion of the 3' end of the ζ_2 gene, the entire inter ζ DNA and the 5' end of the ψζ gene was noted in 8 of the 345 (= 2.3%) patients. The proportion of Hb S (av; 41.5%) and the MCV (av; 89 fl) in AS probands suggest that the ζ deletion is associated with normal expression of α globin genes.

The three haplotypes in different combinations could account for certain aspects of the heterogeneity of Sickle Cell Anemia.

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