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Effect of group of genetic markers on induction of fetal hemoglobin and disease severity in hemoglobinopathies

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As higher HbF levels in patients with β-thalassemia or sickle cell anemia are associated with milder phenotypes, several groups are exploring the factors that regulate the switch from fetal to adult hemoglobin and its therapeutic potential. The contributions of cis-acting elements to HbF production are critical to understand the variable clinical phenotypes of β-thalassemia and sickle cell anemia. Recently, there has been considerable progress made in defining the loci or genomic regions that may often be involved in the regulation of the switch from fetal to adult hemoglobin. In the study we enrolled 79 β-thalassemia homozygous, 11 sickle cell anemia and 14 sickle-β-thalassemia individuals. Fifty age and sex matched healthy individuals were enrolled as the normal control group. Our objective was to study the effect of cis-DNA haplotypes, motifs or polymorphisms (Pre G γ globin gene haplotypes, Aγ-δ intergenic region haplotypes XmnI and (AT)x(T)y polymorphisms, β-LCR HS2 and HS3 site motifs) that may contribute to higher HbF levels and a milder clinical course. We found that a combination of T haplotype of the Aγ–δ intergenic region, TAG Pre-Gγ haplotype, presence of the XmnI polymorphism along with the (AT)9(T)5 motif constitutes a topography that co-relates with raised HbF levels which may contribute in ameliorating the disease severity.

Biography

Anita Nadkarni has completed her PhD in year 1995 from University of Mumbai and Postdoctoral studies from the National Institute of Advanced Industrial Science and Technology (AIST), Tsukuba Japan. She is the Deputy Director of National Institute of Immunohematology. She has made an outstanding contribution to the field of molecular genetics of thalassemia and other hemoglobinopathies during the past two decades. She is the recipient of many national and international awards. She has published more than 110 papers in reputed journals and has been serving as an Editorial Board Member of repute.

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