Single Nucleotide Polymorphisms in PON1 Gene and their Relation with PON1 Activity in Vokkaliga Population of Mysore, South India

Jyothi Mallaiah, Rajashekara Reddy Ravuri*, Elizabeth A. Frank and Cletus J. M. D'Souza

Department of Studies in Biochemistry, Manasagangotri, University of Mysore, Mysore 570 006, Karnataka, India
Telephone: +91- 0821 2419621, E-mail: jyothm@gmail.com
*Anthropoligical Survey of India, Southern Regional Centre, Bogadi II Stage North, Mysore 570 026, Karnataka, India

KEYWORDS Phenotype, PON1, HDL, SNPs, Vokkaliga

ABSTRACT The cardio protective functions of HDL are attributed to the HDL-associated proteins like Paraoxonase 1 (PON1). PON1 is associated with HDL through a N-terminal signal peptide. HDL-associated PON1 alone is cardio protective. Indians have normal HDL levels and yet a high risk of cardiovascular diseases. Hence, the presence of polymorphisms in the promoter region and first exon of PON1 were investigated in 256 individuals to find out whether the polymorphisms could be the cause of the high risk. Out of these 256 individuals, 195 were from Vokkaliga community and the rest were from other communities. Whole blood was collected, plasma and cells were separated. Plasma was used for determination of PON1 activity and for lipid analysis. Cells were used for DNA extraction. DNA was sequenced from -1246 to +203 of the PON1 gene. Nine single nucleotide polymorphisms (SNPs) were identified of which three were new SNPs, two of which were in the 5' flanking region of the putative mRNA for the protein but none in the first exon. Our results show that the low active PON1 phenotype was predominant in the Vokkaliga population.