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Apolipoprotein C3 (*Sst*I) Gene Variability in Northwest India: A Global Perspective

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ABSTRACT Apolipoprotein C3 plays an important role in the receptor mediated hydrolysis of triglyceride rich lipoproteins (TRLs) by inhibition of lipoprotein lipase (LPL), delayed clearance of which causes hypertriglyceridemia (HTG). Indians are considered to be more vulnerable to the adverse effects of hypertriglyceridemia and consequently its probable sequel of cardiovascular disorders. Several studies have revealed the association of rare allele (S2) of APOC3 (*Sst*I) polymorphism with dyslipidemias and coronary artery diseases. In order to investigate the role and relevance of this polymorphism in Northwest India, the present study aimed to investigate the genetic variation of 3' untranslated region of APOC3 (*Sst*I) in 312 individuals belonging to four endogamous groups (Banias, Brahmins, Jatsikhs and Khatris) of Punjab. Uncommon *S2 allele frequency was 22.6%, 22.5%, 22.7% and 26.2% in Banias, Brahmins, Jatsikhs and Khatris respectively. Higher heterozygosity of 0.39 in Khatris reflected their greater variation at this locus than the other populations. Chi-square analysis did not reveal any significant differences between these populations and other studies from North India ($P > 0.05$). Comparative analysis of 66 other populations across the world revealed large heterogeneity at this locus whereby, Mongoloid populations have the highest frequencies of *S2 allele (0.19 to 0.48) followed by Indians (0.18 to 0.29), Africans (0.04 to 0.27) and Caucasian populations (0.01 to 0.12). Genetic distance and multivariate analyses showed that Indian population is quite distinct from other Caucasian and Oriental populations. Clinal heterogeneity of predisposing *S2 allele in Asia showed an increasing cline ($y = 0.0043x + 0.1209$, $R^2 = 0.1162$) towards North. As this allele is associated with HTG and other cardiovascular complications, differential variation in different populations may have insightful implications for association and medical genetic studies.