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A single nucleotide polymorphism in the *SLC19A1* gene is associated with thoracic aortic aneurysms and dissection in Indian population

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Aim: Genetic susceptibility is an important risk factor for aortic wall degeneration and it leads to thoracic aortic aneurysm and dissection (TAAD). In many patients with TAD, the aorta progressively dilates and ultimately ruptures. The purpose of this study was to determine the single nucleotide polymorphism in six genes associated with thoracic aortic aneurysm and dissection patients in Indian population-a case-control study.

Methods: Genomic DNA was isolated from blood and aortic wall tissue of 66 patients with degenerative TAAD, and 67 control individuals. Six SNPs- rs819146, rs8003379, rs2853523, rs326118, rs3788205 and rs10757278 were genotyped using TaqMan SNP genotyping assays (Applied Biosystems, Foster city, Calif). The data was analyzed using STATA11.0 statistical software. Associations between polymorphisms and disease in tissue, blood and within gender were estimated with odds ratios and their 95% confidence intervals.

Results: The T allele frequency for the SNP on 21q22.3, 5' near gene as rs3788205 (-2174 C/T) was higher in male patients than in male controls (P=.049). Moreover, with adjustment for traditional cardiovascular risk factors (sex, age, hypertension, dyslipidemia, diabetes and smoking), the rs3788205 {odds ratio (OR) 0.41, 95% confidence interval (CI) 0.14 to 1.09} polymorphism was found to be an independent susceptibility factor for TAAD in males.

Conclusion: Our results suggest that a sequence variant on 21q22.3 is an important susceptibility locus that confers high cross-race risk for development of TAAD in Indian population.

Biography

Kalpnath is pursuing his PhD from All India Institute of Medical Sciences, New Delhi, India. He has published two papers. He has involved in the project entitled "Induction of therapeutic angiogenesis in limb ischemia by intra-arterial delivery of autologous bone-marrow derived stem cells" and "Transplantation of mesenchymal stem cells in animal models of myocardial infarction and Parkinson's disease" funded by Department of Biotechnology (DBT), Government of India.

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