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

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Objective: Genetic susceptibility is an important risk factor for aortic wall degeneration and its 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 6 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, USA). The data were 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=0.049). Moreover, with adjustment for traditional cardiovascular risk factors (sex, age, hypertension dyslipidemia diabetes and smoking), the rs3788205 (odd ratio (OD) 0.41, 95% confidence interval (CL) 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.

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Clinical and economic burden of congestive heart failure in a non-North American country

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The two components of disability-adjusted life years (DALYs), years of life lost (YLL) and years lived with disability (YLD) are underutilized in evaluating heart failure with reduced ejection fraction (HFrEF) and global burden of diseases (GBD). We aimed to describe both direct (medical) and indirect (morbidity and mortality) cost of CHF in high-income non-OECD Middle Eastern countries in relation to YLL and YLD. We used the World Health Organization GBD methodology to calculate DALYs, YLL, and YLD in 174 prospectively enrolled patients in a single center heart failure registry using 0.4 disability weight (DW) and 3% future age discounts. We reported the cost of hospitalization, re-hospitalization, and non-invasive and invasive procedures per 1,000 HFrEF patients in US dollars. Expressing results as per 1,000 HFrEF capita revealed DALYs of 1480±1909 vs. 2177±2547 (460, 3894) in females and males, respectively. The costs per HFrEF capita in US dollars (USD) were $909.00±676.1 for a single day hospital stay, $7,999 for single hospitalization, $12311±13840 for annual hospitalizations, $20486±22068 for all cause hospitalizations, and $37355±49336 from the time of diagnosis until death or recovery. In this study, HFrEF imposed a substantial economic and disability burden on the non-OECD non-North American Middle Eastern countries; however, males represented a higher economic burden than females.

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