 ISSN 2362-0412

Minor Allele Frequencies of Single Nucleotide Variants Associated with Diabetic Peripheral Neuropathy in a Cohort of Sri Lankan Population

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ABSTRACT

There is increasing evidence that genetic factors could contribute to the development of Diabetic Peripheral Neuropathy (DPN). The objective of this study is to estimate the minor allele frequencies (MAF) of single nucleotide variants (SNV) associated with DPN in the Sri Lankan population and to compare the corresponding MAF in five different populations reported in the 1000 Genomes database. The genetic variants associated with DPN were identified by an extensive search of scientific literature published in PubMed and were annotated using the SnpEff software to filter the exonic and splice-site variants. MAF of the 11 selected variants in the Sri Lankan population were calculated from available data of 50 individuals in the genomic database in Human Genetics Unit, Faculty of Medicine, University of Colombo, Sri Lanka. The MAF of these variants were compared with the corresponding frequencies in 5 different populations reported in the 1000 Genomes phase 3 release (http://www.1000genomes.org) database. The MAF of exonic and splice-site variants of *BDKRB2*, *ADIPOQ*, *VEGFA* and *HSPA5* genes in the Sri Lankan population reported statistically significant ($p<0.05$) MAF with all the other globally represented populations of Americans of African Ancestry in SW USA (ASW), Bengali from Bangladesh (BEB), Utah residents with North and Western European Ancestry (CEU), Han Chinese in Beijing, China (CHB) and British in England and Scotland (GBR). *MTHFR* variant in CEU, CHB and GBR, *CYBA* variant in CHB and *PPARG* in ASW populations were reported statistically significant ($p<0.05$) MAF. *KCNJ11* and *APOE* gene variants of Sri Lankan population showed no statistically significant MAF with all the other globally represented populations. This study shows that MAF of some important exonic and splice-site variants of key genes associated with DPN in the Sri Lankan cohort had statistically significant differences compared to other global populations.

Keywords: *Diabetic peripheral neuropathy, minor allele frequency, exonic and splice site variants, diabetes mellitus, susceptible and prognostic genes.*