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Frequency of variants in genes associated with diabetic peripheral neuropathy in Sinhalese in Sri Lanka

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Background & Aim: Diabetic Peripheral Neuropathy (DPN) is one of the devastating complications of Type-2 Diabetes Mellitus (T2D) that presents in around 50% of cases. There is increasing evidence that genetic variants also contribute to the development of DPN. Ethnic differences in the distribution of these variants have also been reported. The objective of this study is to describe the Minor Allele Frequencies (MAF) of Single Nucleotide Variants (SNV) associated with the susceptibility and prognosis of DPN in Sinhalese in Sri Lanka and to compare the corresponding MAF in 5 different populations reported in the 1000 Genomes database.

Materials & Methods: The genetic variants associated with the susceptibility and prognosis of DPN were identified by an extensive search of scientific literature published in PubMed using the search terms 'Diabetic peripheral neuropathy/genetics', and 'genome-wide association study'. The identified variants were annotated using the SnpEff software to filter the exonic and splice-site variants. The MAF of each of the selected variants in the Sri Lankan population were calculated using the de-identified exome sequencing data of 47 Sinhalese individuals available in the genomic database maintained at the 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 database namely: 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).

Results: The MAF of exonic and splice-site variants of *BDKRB2, ADIPOQ, VEGFA* and *HSPA5* genes in the Sri Lankan population showed statistically significant ($p<0.05$) differences in comparison to all the other globally represented populations. The *MAF* of *MTHFR* variant in *CEU, CHB* and *GBR, CYBA* variant in *CHB* and *PPARG* in *ASW* populations, respectively showed statistically significant differences ($p<0.05$). The MAF of *KCNJ11* and *APOE* gene variants in the Sri Lankan population showed no statistically significant differences compared with all the other globally represented populations.

Conclusion: This study shows that the MAF of important exonic and splice-site variants of key genes associated with DPN in Sinhalese in Sri Lanka had statistically significant differences when compared with other global populations. It would be necessary to undertake studies to understand the prognostic and therapeutic implications of these variants in a large cohort study.

Biography

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