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"A TALE OF GENETIC VARIATION IN THE HUMAN SLC22A1 GENE ENCODING OCT1 AMONG TYPE 2 DIABETES MELLITUS POPULATION GROUPS OF WEST BENGAL, INDIA"

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ABSTRACT

The organic cation transporter 1, OCT 1 (also called SLC22A1-Solute Carrier Family 22 member 1), appears to play a role in the efficacy and disposition of variety of organic cation including drugs. Genetic polymorphisms in the drug transporter have been increasingly recognized as a possible source of variation in drug disposition and response. Genetic variants in OCT1 have been identified largely in European, Asian (Japanese, Chinese and Korean) populations. Interestingly, eight genetic variations were found in the human *SLC22A1* gene, which encodes OCT 1, from 50 type 2 diabetes mellitus individuals (T2DM), in West Bengal population. The purpose of this study was to investigate genetic variants of OCT1 in West Bengal populations. We detected the three previously reported non-synonymous variations, 480 G>C (L160F); 1022 C>T (P341L); 1222 A>G (M408V) and one synonymous variations 156 T>C (S52S) at a minor allele frequencies (MAF) of 0.63, 0.20, 0.43 and 0.27 respectively. We also found four previously reported intronic variations: IVS1-43(T>G), IVS2 -99(C>T), IVS5 -61(G>A), IVS9 +43(C>T) with minor allele frequencies of 0.20, 0.17, 0.18, and 0.37 respectively.

KEYWORDS: SLC22A1, OCT1, Non-Synonymous Single Nucleotide Polymorphisms, Type 2 Diabetes Mellitus, West Bengal Population