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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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The organic cation transporter-1, OCT1 (also called SLC22A1-Solute Carrier Family 22 member 1), appears to play a role in the efficacy and disposition of variety of organic cations 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 OCT1, 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.

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Construction of molecular breeding platform using population sequencing information in rice

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We re-sequenced a biparent genetic population including 132 core Recombinant Inbred Lines (RILs) of Liang-You-Pei-Jiu (LYP9), the widely cultivated super hybrid rice and constructed a high-resolution linkage map with 171,847 Single Nucleotide Polymorphism (SNP) markers. This is the first to have successfully assembled the genome sequence of the maternal cultivar PA64 of LYP9 and we have significantly improved the genome sequence of the paternal variety 93-11. Based on the high-density physical map, we detected 43 yield-associated quantitative trait loci, of which 20 are unique. Also these genetic materials were used in whole genome transcription analysis. Using the IlluminaHiSeq 2000 technique and the 132 RILs, the transcript expression of eQTL in different tissues of heading stage was analyzed. Compared with the yield QTLs, some stable or differential eQTLs were found. In the leaves and seeds, there were 270 and 2791 F1 differential expresses, respectively, relative to their parents mainly involved in carbon metabolism and plant hormone signal transduction pathway. We also analyzed specific expression of allele genes and more than 70% of the transcripts of allelic gene expression trends to the male parent. Multiple genome modules were also identified to control the same trait or a few characteristics. The result has provided an ideal platform for dissecting yield-associated loci, eQTL in super hybrid rice and molecular breeding.

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