

ASSESSING DISEASE-RELATED POLYMORPHISM VIA CORRELATION BETWEEN SNP FREQUENCIES AND EPIDEMIOLOGICAL DATA

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RESUMO

The genetic differences between human individuals do not go beyond 0.5%. This variation can be evaluated by Genome-Wide Association studies (GWAS) that analyze a huge number of DNA variants in a case-control strategy. However, sometimes is difficult to find biological significance to the GWAS results and complementary approach may be used. Here, we present an alternative approach to identify disease-causing variants focused on the simultaneous evaluation of genome variant data, obtained from databases such as the 1000 Genome Project, and epidemiological data of a given pathology. This approach is based in the premise that: if a population presents a high frequency of a given genetically determined pathology, variants that confer susceptibility to this disease also should be more frequent, regarding highly penetrant genetic variants. To test this premise three conjuncts of genes were analyzed: variants related with depression in GWAS (Overrepresented group), housekeeping genes no related with depression in the literature (*ACT*, *B2M*, *EEF2*, *GAPDH*, *PLEK2* and *PPIA*) (Underrepresented group); and four genes already tested by genotyping in case-control studies involving depression (*TPH2*, *NR3C1*, *SLC6A2*, and *SLCA3*). Nine of 82 SNPs, analyzed by GWAS, showed positive correlation between depression rate and genotype frequencies in the 1000 genomes populations, but no SNP showed correlation in the underrepresented group (286 SNPs); on the other hand, 2 SNPs of the *TPH2* (550 SNPs); 26 SNPs of the *NR3C1* (562 SNPs) and 4 SNPs of the *SLC6A3* (377 SNPs) showed correlation with depression rate. These data were compared with data obtained from the literature involving these SNPs in case-control studies of depressive patients and most of the results were similar with ours. Also, some SNPs did not studied in depressive patient were related to other psychiatric disorders. The results obtained here support this approach as a complementary method to classical association studies, aiming the identification of potential disease-causing variants.

PALAVRAS-CHAVE: Depression, variants, polymorphisms, genome projects, biobank, databases

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