Single Nucleotide Polymorphism

A Single Nucleotide Polymorphism, (SNP, pronounced snip; plural snips) is a DNA sequence variation occurring commonly within a population (e.g. 1%) in which a single nucleotide — A, T, C or G — in the genome (or other shared sequence) differs between members of a biological species or paired chromosomes. For example, two sequenced DNA fragments from different individuals, AAGCCTA to AAGCTTA, contain a difference in a single nucleotide. In this case we say that there are two alleles. Almost all common SNPs have only two alleles. The genomic distribution of SNPs is not homogenous; SNPs occur in non-coding regions more frequently than in coding regions or, in general, where natural selection is acting and 'fixing' the allele (eliminating other variants) of the SNP that constitutes the most favorable genetic adaptation.

Other factors, like genetic recombination and mutation rate, can also determine SNP density.

The purpose of a study of Zholdybayeva et al. was to explore the relationship between single nucleotide polymorphisms (SNPs) and intracranial aneurysm (IA) in Kazakhstan population. The patients were genotyped for 60 single nucleotide polymorphisms. Genotyping was performed on the QuantStudio 12K Flex (Life Technologies). A linear regression analysis found 13 SNPs' significant association with development and rupture of IA: the rs1800956 polymorphism of the ENG gene, rs1756 46 polymorphism of the JDP2 gene, variant rs1800255 of the COL3A1, rs4667622 of the UBR3, rs2374513 of the c12orf75, rs3742321 polymorphism of the StAR, the rs3782356 polymorphism of MLL2 gene, rs3932338 to 214 kilobases downstream of PRDM9, rs7550260 polymorphism of the ARHGEF, rs1504749 polymorphism of the SOX17, the rs173686 polymorphism of CSPG2 gene, rs6460071 located on LIMK1 gene, and the rs4934 polymorphism of SERPINA3. A total of 13 SNPs were identified as potential genetic markers for the development and risk of rupture of intracranial aneurysms in the Kazakh population. Similar results were obtained after adjusting for the confounding factors of arterial hypertension and age [1)].

Zhang et al. assessed three SNPs (rs161827, rs161818, and rs161810) of the CD137 gene and their association with ischemic stroke in a northern Chinese Han population. A total of 496 ischemic stroke patients and 486 gender-matched control subjects were genotyped.

They classified these patients according to complications with diabetes and hypertension and also by ischemic stroke subtypes. Allele, genotype, and haplotype association studies were tested in all patients and subgroups.

They used multivariable logistic regression analysis combined with 10,000 permutations to analyze the association of CD137 polymorphisms with ischemic stroke. After adjusting for relevant factors, rs161827 was significantly different between patients with and without diabetes and the control group ($p = 0.0001$, $p = 0.014$, and $p = 0.0001$, respectively). In addition, rs161818 and rs161810 differed significantly between patients without diabetes and the control subjects ($p = 0.0001$ and $p = 0.004$, respectively). rs161827, rs161818, and rs161810 were all statistically significant among the combination stroke subgroup compared with the controls. These results indicate that the CD137 gene is associated with risk of ischemic stroke in the northern Han Chinese. Moreover, CD137 gene polymorphism may be one mediating factor between diabetes and ischemic stroke [2)].

References

1)

Zholdybayeva EV, Medetov YZ, Aitkulova AM, Makhambetov YT, Akshulakov SK, Kaliyev AB, Talzhanov YA, Kulmambetova GN, Iskakova AN, Ramankulov YM. Genetic Risk Factors for Intracranial Aneurysm in the Kazakh Population. J Mol Neurosci. 2018 Aug 18. doi: 10.1007/s12031-018-1134-y. [Epub ahead of print] PubMed PMID: 30121816.

2)

Zhang S, Li Z, Zhang R, Li X, Zheng H, Ma Q, Zhang H, Hou W, Zhang F, Wu Y, Sun L, Tian J. Novel CD137 Gene Polymorphisms and Susceptibility to Ischemic Stroke in the Northern Chinese Han Population. Neuromolecular Med. 2017 Jul 28. doi: 10.1007/s12017-017-8457-7. [Epub ahead of print] PubMed PMID: 28755037.