

Large Scale SNP Validation in Koreans

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Since genetic variation plays an important role in many diseases, a major focus of the human genome project has been to identify a large number of uniquely mapped single nucleotide polymorphisms (SNPs) to serve as tools in genetic studies of complex traits. The SNP Consortium (TSC) initiated the Allele Frequency Project to determine the allelic frequencies for single nucleotide polymorphisms (SNPs) in African Americans, Asians (Japanese-Chinese), and Europeans. The Korean government launched a similar project to compare genetic make up of Korean population with those of the other populations. Here we report the largest and most extensively characterized set of SNPs covering the human genome to date. Very few SNPs are highly divergent between any two populations. Our results further document that much but not all genetic variation is shared among populations. For autosomes, some 44% of these SNPs have a minor allele frequency $\geq 10\%$ in each population, and the average allele frequency differences between populations with different continental origin were less than 19%. The differences among the Asian populations were small but significant. The Japanese-Korean comparison was the smallest. For each of the three comparisons, at least 99.0% of SNPs have a divergence of less than 35%. Allele frequency differences among Korean, Japanese, and Chinese populations are a few percent, suggesting caution in using mixtures of well-established populations for case-control studies to find complex disease variants.