Short Communication

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Bioinformatics investigation of SNP distributions among human populations

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Abstract

Next Generation Sequencing revealed an intricate distribution of millions of genetic variants among different human populations. Both rare and frequent genetic variants may bring to light important information about history, evolution and health status of humans. First, we present a novel computational method for detecting identical-by-descent (IBD) chromosomal segments between sequenced genomes. It utilizes the distribution patterns of very rare genetic variants (vrGVs), which have minor allele frequencies less than 0.2%. Contrary to the existing probabilistic approaches our method is rather deterministic, because it considers a group of very rare events which cannot happen together only by chance. This method has been applied for exhaustive computational search of shared IBD segments among 1092 sequenced individuals from 14 populations. It demonstrated that clusters of vrGVs are unique and powerful markers of genetic relatedness, that uncover IBD chromosomal segments between and within populations, irrespective of whether divergence was recent or occurred hundreds-to-thousands of years ago. We found that several IBD segments are shared by practically any possible pair of individuals belonging to the same population. Moreover, shared short IBD segments (median size 183 Kb) were found in 10% of inter-continental human pairs, each comprising of a person from Sub-Saharan Africa and a person from Southern Europe. The shortest shared IBD segments (median size 54 Kb) were found in 0.42% of inter-continental pairs composed of individuals from Chinese/Japanese populations and Africans from Kenya and Nigeria. Knowledge of inheritance of IBD segments is important in clinical casecontrol and cohort studies, since unknown distant familial relationships could compromise interpretation of collected data. Clusters of vrGVs should be useful markers for familial relationship and common multifactorial disorders. Lastly, we present whole-genome investigation of human most frequent genetic variants and their arrangement in common haplotypes in various geographical regions. Three prominent types of common haplotypes.

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Biography

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