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The genetic structure of the Greek population in relation to the European HapMap reference samples

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The HapMap project has provided a unique tool for the analysis of human genetic variation, providing reference information for allele frequency and genotype distributions as well as linkage disequilibrium patterns of Single Nucleotide Polymorphisms (SNPs) across the entire genome. The latest release of HapMap phase 3 data provides genotypes for millions of SNPs in 11 populations from around the world, with Europe being represented by the CEU (originating from Northwestern Europe) and the TSI populations (Tuscan Italians from Southern Europe). Although initial studies support the fact that the CEU can be used as reference for the selection of tagging SNPs in other European populations, a critical step in the design of genetic association studies, this hypothesis has not been extensively studied across Europe and in particular in Southern Europe. We set out to explore the extent to which the HapMap populations can be used as reference for a previously unstudied population of South-Eastern Europe, the Greek population. To do so we studied genomic variation in more than 1,500 SNPs, genotyped by our group in 56 individuals of Greek origin, and compared them to the CEU and TSI genotypes. Genotype, allele frequency, and pairwise LD measures were compared across all three populations. Tagging SNPs were selected in the CEU and TSI samples and their transferability to the Greek population was tested, using the r^2 metric. Our results are applicable in medical genetics, in order to inform the design of genetic association studies, as well as in studies of evolutionary relationships of Southern European populations.