Abstract/Session Information for Program Number 1521

Session Information

Session Title: . Evolutionary and Population Genetics **Session Type:** Poster

Session Location: Exhibit Hall A, Lower Concourse Level, Convention Center **Session Time:** Thu 7:00AM-7:00PM

Abstract Information

Program Number: 1521/T **Presentation Time:** Thu, Nov 4, 2010, 5:00PM-6:00PM

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Abstract Content

Human genetic differentiation from HapMap data. *E. Elhaik, A. Chakravarti* McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, Baltimore, MD.

Apportionment of human genetic variation has long established that most human variation is within groups and that the additional variation between groups is small but greatest when comparing continental populations. These studies have used Wright 's F_{ST} that apportions the standardized variance in allele frequencies within and between groups in a hierarchical manner. High values of F_{ST} are unlikely in humans due to genetic drift and migration and are consequently used to identify genes undergoing directional or heterotic selection. The availability of the HapMap data from phases I - III now allows us to reexamine these questions. We analyzed data on ~3 million autosomal, X-linked, Y-linked, and mitochondrial SNPs from the HapMap database on 602 samples from 8 populations and a common subset of ~1 million autosomal and X-linked SNPs that have been genotyped in all populations. We identified two major features of the data. First, only a paucity (12%) of the total genetic variation is among populations of different continents and even a lesser (1%) amount among populations of the same continent. These data are remarkably consistent with the early observations of Lewontin in 1972. Second, we demonstrate that, although the overall distribution is similarly shaped (inverse J), the distribution of F_{ST} varies significantly by mean allele frequency. Since the mean allele frequency is a crude indicator of allele age, these distributions mark the time-dependent change in genetic differentiation. The change in mean F_{ST} of these distributions is linear in mean allele frequency suggesting the nature of allele frequency dynamics. These observations are true for autosomal, X-linked, and mitochondrial SNPs, but not Y-linked SNPs. These results suggest that investigating the extremes of the F_{ST} distribution for each allele frequency class may be more efficient for detection of selection. Consequently, we demonstrate that such extreme SNPs are more clustered that that expected from linkage disequilibrium for each allele frequency class. These genomic regions are likely candidates for natural selection.

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