

Workshop: Computational Genomics

Detecting global evidences of recent positive selection in human

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Genome-wide scanning for signals of recent positive selection is essential for a comprehensive and systematic understanding of human adaptation. Here we present a genomic survey of recent local selective sweeps, especially aimed at those nearly or recently completed. A novel approach was developed for such signals, based on contrasting the extended haplotype homozygosity (EHH) profiles between populations. We applied this method to the genome SNP data of both the International HapMap Project and Perlegen Sciences, and detected widespread signals of recent local selection across the genome, consisting of both complete and partial sweeps. A challenging problem of genomic scans of recent positive selection is to clearly distinguish selection from neutral effects, given the high sensitivity of the test statistics to departures from neutral demographic assumptions and the lack of a single accurate neutral model of human history. We therefore developed a new procedure that is robust across a wide range of demographic and ascertainment models, which indicates that certain portions of the genome clearly depart from neutrality. Simulations of positive selection showed that our tests have high power towards strong selection sweeps that have undergone fixation. Gene ontology analysis of the candidate regions revealed several new functional groups that might help explain some important inter-population differences in phenotypic traits.