



The University of Chicago
Department of Statistics

Seminar Series

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Rapid Evolution in the Human Genome

MONDAY, January 14, 2008 at 4:00 PM
133 Eckhart Hall, 5734 S. University Avenue

Refreshments following the seminar in Eckhart 110.

ABSTRACT

Comparative genomics is a powerful approach to investigating the genetic basis for what makes us human. I will describe two different methods we have developed for identifying lineage-specific evolution: a phylogenetic hidden Markov model (phylo-HMM) and a likelihood ratio test (LRT). The phylo-HMM works well for identifying relatively ancient events, while the LRT is much more powerful at the leaves of a phylogeny (e.g. the human lineage). Using this LRT, we identified 202 Human Accelerated Regions (HARs) that were extensively changed in the last 6 million years since divergence from our common ancestor with chimpanzee, but are highly conserved in other species and thus are likely to be functional. The HARs are mostly non-coding sequences, and the set of genes near HARs is enriched for transcription factors, suggesting a role for HARs in the evolution of human gene regulation. I will describe a few of the most intriguing HARs before turning to a curious observation: the most accelerated regions of our genome show a striking bias for AT to GC (“weak-to- strong”) nucleotide substitutions. This pattern is used to speculate about the cause of rapid, biased evolution in the primate genome and to date the chromosome fusion that formed human chr2.