



The University of Chicago  
Department of Statistics  
Seminar Series

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**“Studying Recombination Rate as a Trait”**

**MONDAY May 9, 2005 at 4:00 PM**  
**133 Eckhart Hall, 5734 S. University Avenue**  
*Refreshments following the seminar in Eckhart 110.*

**ABSTRACT**

Recombination is crucial to human genetics and evolution by generating new combinations of variants for selection to take place. A recombination occurs at a location where a chromosome that is passed on from a parent to an offspring changes from what the parent inherited from one grandparent to what was inherited from the other grandparent. Mathematically the recombination locations and counts can be thought of as realizations of an inhomogeneous and non-Markov point process. While recombination rates were well known to differ between males (as fathers) and females (as mothers), both locally and globally, it is only recently that it becomes clear that individual women have different recombination rates. Furthermore there are systematic differences among the maternal recombination counts of children of the same mother — maternal recombination counts are highly correlated among chromosomes even after the adjustment for the mother effect. Part of this difference between siblings is due to the recombination count of a child is correlated with the age of the mother at birth. It is hypothesized that the latter is a result of a selection effect in that a higher recombination count for an oocyte/egg increases the chances of a lifeborn and this effect gets stronger as the mother ages. This hypothesis is supported by the observation that women who have a higher recombination rate tend to have more children, i.e. fertility is associated with recombination rate. Based on the correlation of estimated recombination rates of sisters, heritability of recombination rate is estimated to be about 30%. We recently identified a common inversion on chromosome 17 that is associated with both fertility and recombination, but its effect only explains 1% of the heritability. It is noted that recombination rate is a very difficult trait to study. Unlike other phenotypes, the recombination rate is not directly observable. Indeed, because it is a rate, mathematically it has to be interpreted as the average recombination counts averaging over an infinite number of children of a mother. Since a woman only has a limited number of children, the best we can get is an estimate of the rate based on a modest sample size. What makes it even more difficult is that recombination counts of children cannot be directly observed, and can only be inferred/estimated from genome-wide marker data on the mother, preferably also the father, and at least two children. We are fortunate that because of the Icelandic genealogy and genotype database that have been built up, we are able to obtain estimated recombination rates for over 5000 women. There are many methodological and computational challenges and so far our analyses have focused more on avoiding biases instead of optimizing efficiency. One can consider the data as resulting from a hierarchical model of point processes, with complications that the observed data are incomplete and the differences among individuals could be region specific. The genetic component of the trait and that the data could be observed with selection introduce further complexities.