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NEWS AND COMMENTARY

Evolutionary genetics

Autosomes behaving badly

RC Moore

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At the heart of the male/female dichotomy in human beings are two morphologically and functionally distinct chromosomes: the X and Y sex chromosomes. Understanding the origin and evolution of these chromosomes is fundamental to understanding what makes us human. Indeed, one of our most detailed models of sex chromosome evolution comes from genomic analysis of the human sex chromosomes. However, human sex chromosomes are evolutionarily old, having arisen 240–320 million years ago (Lahn and Page, 1999), obscuring the early events in sex chromosome evolution. Would not it be nice to step back in time and observe the initial events leading to the differentiation of the X and Y chromosomes?

This is what Zhiyong Liu *et al* (2004) have accomplished – minus the time travel – by analyzing the genomic organization of the incipient sex chromosomes in the flowering plant, papaya (*Carica papaya*). Although plants and animals have evolved fundamentally different molecular machinery to control the expression of sexual phenotypes, the genetic architecture of their sex chromosomes may have been shaped by common evolutionary forces (Negrutiu *et al*, 2001; Charlesworth, 2002).

Papaya is polygamous; it has male, female, and hermaphrodite individuals. Males and hermaphrodites are considered to be heterogametic, as their sex is determined by the presence of a dominant allele on the papaya Y chromosome. However, the X and Y chromosomes of papaya are morphologically indistinct, unlike the human sex chromosomes.

While the papaya sex chromosomes may 'look' like autosomes, they behave as *bona fide* sex chromosomes: in particular, there is reduced recombination on part of the Y chromosome. Liu *et al* used a combination of genetic and genomic mapping techniques to determine the underlying genetic mechanism responsible for the inhibition of recombination in this 4–5 Mbp region of the Y chromosome. This region, which represents ~10% of the Y chromosome, contains 342 genetic markers that cosegregate with the sex determination locus.

These data directly support the hypothesis that sex chromosomes evolve from a single pair of autosomes, initially via the inhibition of recombination in the region containing the sex determination loci. Similar conclusions have been inferred from studying other sex chromosome systems. However, by studying the genomic architecture of the incipient sex chromosomes of papaya, it is evident that the extent of recombination suppression can be relatively small, of the order of a few Mbp. This is in direct contrast to the sex chromosomes of humans where recombination is suppressed across the majority of the Y chromosome.

It is unclear as to whether the size of the nonrecombining region of the papaya Y chromosome represents the exception or the rule. Does recombination inhibition start small and expand across the entire chromosome with time? In humans, there is evidence that multiple chromosomal inversions followed by autosomal translocations on the Y resulted in genetic isolation of the Y from the X (Lahn and Page, 1999). It will be necessary to date the age of the nonrecombining region of the papaya sex chromosome, via molecular phylogenic analyses of homologous regions in sister species, to determine whether or not the cessation of recombination occurred relatively recently. Has the nonrecombining region had time to expand in size or not?

Perhaps the initial size of this region is determined more by the chance positioning of sex determining loci. Recombination suppression is predicted to arise via selective pressure to link at least two sex determination genes, one masculinizing and one feminizing (Charlesworth, 2002). If these genes are initially located relatively close to each other, possibly as a result of transposition, the nonrecombining region can be relatively compact. If they are farther apart, recombination must necessarily be repressed over much larger chromosomal regions, possibly due to large-scale chromosomal rearrangements.

Liu *et al* also found evidence for significant genetic degradation within the nonrecombining region of the Y chromosome. A 513 kbp region of the nonrecombining region contained a higher density of retroelements and inverted repeats, and lower gene density than a genome-wide survey of a similarly sized region. Moreover, ~65% of 'test sequences' are divergent between the X and Y, being present only in males and hermaphrodites. If the nonrecombining region of the papaya Y chromosome is relatively young, then it is clear that chromosomal degradation occurs rather quickly. Future studies to analyze the molecular population genetics of genes in this region will determine if these genes exhibit characteristically low nucleotide variation, as has been seen for the nonrecombining regions of other Y chromosomes (McAllister and Charlesworth, 1999; Filatov *et al*, 2000; Bachtrog and Charlesworth, 2002). Such analyses will help determine the evolutionary forces, such as background selection or sexual selection, which have contributed to degradation of the nonrecombining region of the Y (Charlesworth and Charlesworth, 2000).

It will also be of interest to assay the types of genes found in the nonrecombining region. In one of the success stories of the human genome project, sequencing of the Y has found that the majority of Y-linked genes are involved in spermatogenesis (Skaletsky *et al*, 2003). These are genes that have either escaped degradation or have translocated to the Y. This observation has debunked the idea that the Y chromosome is a genetic wasteland; rather, it is functionally coherent. The accumulation of male developmental genes on the Y agrees with sexual conflict theories, which predict the migration of male genes from the X to either the Y or the autosomes (Wu and Xu, 2003). Has such a process already occurred in the papaya nonrecombining chromosomal region, or is this a process that occurs rather late in the evolution of sex chromosomes?

This study of sex chromosome structure and function in the incipient sex chromosomes of papaya has led to greater understanding of the evolutionary mechanisms common to all sex chromosomes. Liu *et al* (2004) have painted a clear picture of the early events in sex chromosome evolution: origination from a common pair

of autosomes, localized inhibition of recombination in the region containing sex determination loci, and rapid degeneration of the nonrecombining region. By analyzing the genetic architecture of papaya sex chromosomes, the authors have provided a glimpse of our own evolutionary history. Future investigations will determine how far the similarities between different sex chromosome systems extend.

RC Moore is at the Department of Genetics, North Carolina State University, USA.

e-mail: rcmoore@unity.ncsu.edu

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