

HL106

## **Detecting Evolutionary Strata on the Human X Chromosome in the Absence of Gametologous Y-Linked Sequences**

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### **Abstract**

Mammalian sex chromosomes arose from a pair of homologous autosomes that differentiated into the X and Y chromosomes following a series of recombination suppression events between the X and Y. The stepwise recombination suppressions from the distal long arm to the distal short arm of the chromosomes are reflected as regions with distinct X-Y divergence, referred to as evolutionary strata on the X. All current methods for stratum detection depend on X-Y comparisons but are severely limited by the paucity of X-Y gametologs. We have developed an integrative method that combines a top-down, recursive segmentation algorithm with a bottom-up, agglomerative clustering algorithm to decipher compositionally distinct regions on the X, which reflect regions of unique X-Y divergence. In application to human X chromosome, our method correctly classified a concatenated set of 35 previously assayed X-linked gene sequences by evolutionary strata. We then extended our analysis, applying this method to the entire sequence of the human X chromosome, in an effort to define stratum boundaries. The boundaries of more recently formed strata on X-added region, namely the fourth and fifth strata, have been defined by previous studies and are recapitulated with our method. The older strata, from the first up to the third stratum, have remained poorly resolved due to paucity of X-Y gametologs. By analyzing the entire X sequence, our method identified seven evolutionary strata in these ancient regions, where only three could previously be assayed, thus demonstrating the robustness of our method in detecting the evolutionary strata.