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Are there rearrangement hot spots in the human genome?

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The debate over the validity of genomic rearrangement “hotspots” has its most recent addition in a new theory put forth by researchers at the University of California San Diego. The study, published on November 9 in *PLoS Computational Biology*, holds that there are indeed rearrangement hotspots in the human genome.

Doctors Max Alekseyev and Pavel Pevzner developed a theory for analyzing complex rearrangements (including transpositions) which demonstrates that even if transpositions were a dominant evolutionary force, there are still rearrangement hotspots in mammalian genomes.

In 1970 the random breakage model (RBM) was proposed by Susumo Ohno, and later formalized by Nadeau and Taylor in 1984. This model postulates that rearrangements are “random,” and thus there are no rearrangement hotspots in mammalian genomes. Biologists largely embraced the model as it held such predictive powers.

However, in 2003 the model was refuted by Pevzner and Tesler, who suggested an alternative fragile breakage model (FBM) of chromosome evolution. FBM implies that the human genome is a mosaic of solid regions with low propensity for rearrangements and fragile regions where rearrangement hotspots reside. The rebuttal of RBM resulted in a rebuttal of the rebuttal, and a scientific divide was begun.

Most recent studies support the existence of rearrangement hotspots, but some researchers still uphold the RBM model. This study represents a

major advance in the debate.

Source: Public Library of Science