Recombination Suppression Drives Expansion of the Drosophila Dot Chromosome: methods and sensitivity checks

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Genome size varies widely, even among closely related species, yet much less is known about chromosome size variation. Here we use the fourth chromosome of Drosophila , also known as the "Muller F element" or "dot chromosome", as a model to investigate chromosome-specific size expansion. The F element of most Drosophila species is small (?1.3 Mb) and almost entirely heterochromatic, yet harbors approximately 80 protein-coding genes. Here, we study D. kikkawai , D. takahashii , D. ananassae , and D. bipectinata , whose F elements are 2- to 15-fold larger in size compared to D. melanogaster . Through manual gene curation and comparative genomic analysis, we find that their F elements have expanded primarily via accumulation of transposable elements (TEs) in introns and intergenic regions. Natural selection appears less efficient on these expanded F elements: they have smaller effective population sizes and their genes exhibit reduced usage of optimal codons, compared to D. melanogaster . We propose that F element size variation is driven by differences in F element recombination rates. The ultra-long (?20 Mb) F elements of D. ananassae and D. bipectinata display high rates of rearrangement and sequence evolution and exhibit independent TE-driven expansions. Our results suggest that F elements of most Drosophila species likely recombine enough to prevent size expansion, while F element recombination in D. ananassae and D. bipectinata is either absent or rare enough to allow TEs and other deleterious mutations to accumulate via Muller's ratchet; thus, these chromosomes evolve more like a Y chromosome than a typical Drosophila F element.

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