

# The Ghost of Hubby & Lewontin: Genetic Variation in the Uncharted Wilds of the Genome



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**Time: 2025. 07. 16 Wed. 15:30**

**Venue: Auditorium, 1<sup>st</sup> Floor**

**Interdisciplinary Research Building**

**跨領域科技研究大樓1樓演講廳**

**Host: Dr. Wen-Hsiung Li 李文雄特聘研究員**





## Abstract

Nearly sixty years after Hubby and Lewontin's seminal work on genetic variation, we revisit fundamental questions in recalcitrant regions of the genome using modern genomic techniques. Despite decades of improvement in our ability to survey genetic variation, in many ways, we still haven't satisfied the fundamental requirements for surveying genetic variation identified by Hubby and Lewontin almost 60 years ago. Focusing on highly repetitive genomic regions that encode essential functions but have remained challenging to study, we present significant improvements to the *Drosophila melanogaster* reference genome assembly. Our work successfully reconstructs previously elusive segments, including the complete histone locus and the pericentric heterochromatin of the X Chromosome, spanning from the Stellate locus to the distal flank of the rDNA cluster. We introduce a novel method using landmark anchors based on unique variants to infer structural changes in regions where traditional alignments are impractical. This approach reveals considerable structural variation between *D. melanogaster* strains, including differences in copy number and organization of homologous repeat units. In the histone cluster, we observe minimal genetic exchange indicative of meiotic crossing over, but variation patterns suggest mechanisms such as unequal sister chromatid exchange. We also examine the prevalence and scale of concerted evolution in the histone and Stellate clusters. This work demonstrates how advanced genomic analyses are providing new insights into the nature and extent of genetic variation in previously uncharted genomic territories. While addressing long-standing questions in evolutionary genetics, it also reveals the persistent challenges in fully meeting Hubby and Lewontin's original criteria. Our findings highlight both the progress made and the considerable work still needed to comprehensively characterize genetic variation, illustrating why the "Ghost of Hubby and Lewontin" continues to influence modern genomic research.