Genetic Load in *Drosophila*

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ABSTRACT

What is the biological significance of sexual reproduction and recombination? And why are polymorphisms maintained in natural populations? To address these questions, I have focused on the concept of "genetic load." It is thought that one reason why organisms have sex is to avoid the effects of Muller's ratchet. Deleterious mutations are likely to accumulate on balancer chromosomes (balancers) of *Drosophila melanogaster*, in which recombination is suppressed. I counted the recessive lethal mutations on two balancers from the *In(2LR)SM1/In(2LR)Pm* strain by using second-chromosome deficiencies. Ten and three recessive lethal mutations were detected on *In(2LR)Pm* and *In(2LR)SM1*, respectively. Thus, I experimentally proved the concept of Muller's ratchet in the genomic region in which recombination is suppressed. Next, I measured genetic loads in two wild-type laboratory strains of *D. melanogaster*, Oregon-R and Canton-S, in which inbreeding depression is expected because the strains have been maintained as small populations. I divided the genetic load of each into two categories: that caused by polymorphic deleterious mutations (polymorphic genetic load, PGL) and that caused by fixed deleterious mutations (fixed genetic load, FGL), where the FGL is considered to be indicative of Muller's ratchet. By using balancers, I extracted second chromosomes from each of the laboratory strains and determined the relative viability of each second chromosome in homozygotes and heterozygotes. The frequency of lethal chromosomes was smaller, the allelic ratio of lethal chromosomes was larger, and population size was smaller in the laboratory populations when compared with the natural population that was previously analyzed. Recessive lethal mutations are purged from the natural populations via heterozygotes, while they are purged from the laboratory populations via both homozygotes and heterozygotes. Because the values of random load [#1] were similar to those of mutation load, a mutation-selection balance was met in the laboratory

populations. This supports the classical hypothesis [#2] as the mechanism which maintains polymorphism of deleterious mutations. The distributions of the second-chromosome viability indicated that the laboratory populations have a smaller PGL than the natural population and the laboratory populations have an FGL. In conclusion, the present study showed experimentally that part of the biological significance of sexual reproduction and recombination is their role as a countermeasure against genetic load. Because the composition of genetic loads differs depending on the population size, some advantages of sexual reproduction and recombination must be dependent on population size.

[#1] Random load is the genetic load that is expressed in a random mating population; see Materials and Methods of Part II.

[#2] Classical hypothesis is a hypothesis to explain genetic polymorphisms; see Introduction of Part II.