

A NOTE ON GENETIC LOAD

Genetic load is a term deriving from Muller's (1950) discussion of the human population's "load" of mutations. Muller meant to refer to alleles clearly regarded as deleterious, in essentially any environment, to the individuals possessing them, or to their offspring; but the term was later extended in population genetics models to refer to the less reproductive alleles whenever there is more than a single allele at a given locus, in any part of the species' environment; in other words, the term was extended to refer to alleles deleterious only in relation to other alleles, or when the part of the environment rendering them deleterious is solely the presence of a competing allele.

Population geneticists speak of (1) mutational load (that owing to loss of individuals with unfavorable mutations; (2) segregational load (that owing to loss of homozygotes in cases of balanced polymorphism or when the heterozygote is the best genotype); and (3) substitutional or evolutionary load (that owing to the loss of individuals during the substitution of one allele for another as a result of selection). These terms could be used to refer to offspring or other descendants of some individual whose fitness is under consideration, but in population genetics they have always involved losses to the population as a whole.

When only one allele is present at any particular locus under consideration, the average fitness of genotypes in the population is usually described by geneticists as 1.0. When any other allele is also present, the more favorable of the two is given a fitness value of 1.0, the less favorable a value lower than 1.0, depending on the selection coefficient used in the model. This artifact of numerical calculation together with the tendency to look at the "average" fitness within the population, or the "fitness of the population," explains how misconceptions about genetic load arose. If one gave a new beneficial allele a fitness value higher than 1.0, "fitness" would climb, and numerical treatment would be difficult. But by assigning the value of 1.0 always to the better allele, one implies that populations with two alleles are always less fit than populations with single alleles; and one's attention is diverted to something called the average fitness of the population, and away from the question of the relative fitnesses of the individuals within the population. There is no reason to suppose that the original population would die out if a new allele did not appear, or that the process of substitution of one allele for another during selection is in any meaningful way expensive to the population.

"If a population has only one genotype, that genotype will be classed as 'optimum' and there can be no inferior genotypes. Thus, the mathematical model will show no mortality, at all, even though, as in the case of some egg-laying organisms, the actual average of individual survival is somewhere in the vicinity of one per million. Even worse, a one-genotype population may appear to be doing very well in the mathematical model, even though in reality it is rapidly becoming extinct because its one genotype is inadequate under current conditions. On the other hand, a population with a variety of genotypes will always show some selection losses in the model because some of its genotypes have to be rated less than 1, although it may, in fact, be increasing in numbers quite rapidly. The paradox becomes clearer if we imagine a situation in which there occurs within a population a new mutation with favorable effects on viability. In order to incorporate this into the scheme, it is necessary to demote the previously optimum genotype to a selection value less than 1. This creates a fictitious genetic load, since the old genotype is just as good as it ever was, and the gradual introduction of a superior one can have no effect but to continually enhance the viability of the population and presumably increase its numbers." (Alice Brues, 1963).

"When the heterozygote is the most fit genotype, it has seemed logical to some people to rate it fully fit and assign a load to both homozygotes. . . . To

others this does not seem logical at all, since the heterozygote cannot be established as the sole genotype [in ordinary sexual organisms] Some dissenters have suggested that the most fit homozygote be assigned the rating of 1.0. This necessitates a 'negative load' for the heterozygote, for which fitness then becomes greater than 1.0 Actually one may seriously question whether the segregational load is a load at all, in the sense that mutational load is Obviously the maximum load will be obtained when the optimum genotype is used as a base and all others are declared deficient. The fact that the optimum genotype, if a heterozygote, cannot be established in any entire population of a higher animal is perhaps not the most important point. If we are interested in relative load as a means of comparing the success of two populations with different genetic equipment, the comparison cannot be fairly made unless the standards of fitness applied to the two are the same. If we use a selection coefficient of less than 1 for a homozygote in a population in which a superior heterozygote is present, we should use the same coefficient for the same homozygote in a population in which the heterozygote is not present, if we wish to compare the fitness of the two populations. Only if our interests are limited to determining gene-frequency change or equilibrium point within a single population are we justified in establishing our selection coefficients on a basis unique to that population." (Brues, 1969).

Population genetics rightly focuses on the population as the unit of evolutionary change. It appears that in the process, as indicated in some other essays to which you have been exposed, it has also tended to view the population as the unit of selection, not after a careful review of the likelihood of this being true, but as a more or less accidental outcome of the following factors:

1. A focus on the average fitness of the population rather than on the relative fitness of the individuals within populations.
2. The supposition that if there are two alleles one must be better than the other -- in other words, a focus on the average fitness of each allele rather than on the question of how selection may vary across the range of the species, and how polymorphism may be affected by variations in selection and varying amounts of interbreeding between segments of the same species.
3. Use of 1.0 as maximum fitness
4. A failure to distinguish between the effects of what Wallace (1968) called "hard" and "soft" selection -- in other words, failure to note that during the process of substitution of one allele for another, the individuals in the population may be faring better and better; and the addition of a new allele in a monomorphic population does not necessarily mean that an increased number of individuals must die. Wallace discusses hard and soft selection in a recent paper in Evolution (Vol. 29:465-473), "Hard and soft selection revisited." Hard selection has meant generally that a certain proportion of individuals must die; soft selection means simply that there is a genetic change occurring, but the population numbers are not decreased in the process, and even may be increasing -- it is not how many genotypes are disappearing but which ones.

All of the references above for which only authors and dates are given are listed in the paper: "The search for a general theory of behavior."