Species Conservation: A Population-Biological Approach

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Population Extinction by Mutational Load and Demographic Stochasticity

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Abstract

Genetic aspects are important in the evaluation of the risk of extinction for small populations. Using estimates of rates and effects of slightly deleterious mutations, we calculate the mean time to extinction under the joint action of mutation load and density-dependent stochastic population regulation. Accumulation of mutations diminishes the individual survival probability, which leads to a reduction in population size. This, in turn, progressively facilitates the fixation of future deleterious mutations by random genetic drift. This synergistic interaction has been called the mutational melt-down.

In asexual populations, the probability of extinction increases as the mutational effect increases and as actual population size decreases. As reference points for sexual populations, we present the expected extinction times without mutational load but with stochastic fecundity and sex-ratio under a logistic population regulation. Selection and recombination does not prevent mutational melt-down in small sexual populations; slightly deleterious mutations reduce the mean time to extinction by several orders of magnitude. Stochastic fecundity is a minor direct source of extinction in sexual populations, but it leads to temporary reductions in effective population size, which increases the risk of extinction due to stochastic variations of the sex-ratio.

Introduction

Several well known risks for population extinction increase drastically with decreasing population size. Biotic and abiotic fluctuations of the environment or purely stochastic variation of demographic parameters (like birth and death rates, carrying capacities, and sex-ratio) can reduce a population to a level at which the probability of extinction is high. The smaller the number of individuals is, the more severe are genetic problems such as inbreeding depression, loss of adaptive variation by random drift, and reduction of fitness due to fixation of deleterious mutations. Theoretical aspects of the dynamics of populations

in variable environments have received considerable attention (RICHTER-DYN & GOEL 1972; FELDMAN & ROUGHGARDEN 1975; HANSON & TUCKWELL 1978; MAY 1981; HOPPENSTEADt 1982; NISBET & GURNEY 1982; WRIGHT & HUBBELl 1983, MODE 1985; EWENS *et al.* 1987, GOODMAN 1987, SOULÉ 1987). However, there have been few attempts to incorporate explicit genetic details into extinction models. In the following, we give a review of recent work on the interaction of deleterious mutations, population dynamics and extinction.

In principle, any deleterious mutation can reduce the size of a population. The probability of fixation of such mutations by random genetic drift increases with declining population size, leading to a synergistic interaction which LYNCH & GABRIEL (1990) called a "mutational melt-down". This process is especially relevant for slightly deleterious mutations in small asexual populations. However, because recombination facilitates selection against bad mutations, it is believed that genetic risks are small compared to risks by demographic and environmental influences (LANDE 1988). This opinion will be questioned later on in this paper when we demonstrate the possibility of mutational melt-down in small sexually reproducing species.

Estimates for Mutation Rates and Selection Coeffiecients

The effectiveness of the mutational melt-down depends critically on the rate and effects of mutations. We do not consider lethal recessive mutations since we know from earlier simulations that their influence is small compared with mildly deleterious mutations. Many estimates of mutation rates and mutational effects are available for *Drosophila melanogaster*. According to the review of CROW & SIMMONS (1983) each animal incurs an average of 0.6 new non-lethal mutations, each of which reduces the viability by ≈2.5%. Such mutations appear to be approximately additive within loci (MUKAI 1979). If mutations on different loci act independently then the fitness reductions from each locus can be treated as multiplicative. We will adopt this assumption, although data are not precise enough to rule out other interpretations.

Therefore, letting W' be the fitness of an organism that carries slightly deleterious mutations at n loci and W_0 be the fitness of an organism without these mutations, we have

$$W' = W_0 (1-s_1)(1-s_2)\ldots(1-s_n) , \quad (1)$$

where s_i is the selection coefficient at locus i. Assuming that each new mutation occurs at a different locus and that the selection coefficient is the same for all these loci, one gets

$$W'/W_0 = (1-s)^n \approx (1-s)^{\mu t} \quad (2)$$
$$\approx 1-\mu s t \text{ for small } \mu s t$$

with t as time and with μ as the zygotic mutation rate per time unit and with μt as the expected number of mutations. With these assumptions, LYNCH & GABRIEL (1990) estimated the mutation load for organisms other than *Drosophila* using data from

experiments designed for other purposes. For eukaryotes their estimated boundaries for the mutation load μs are

$$0.0002 < \mu s < 0.02. \quad (3)$$

Within the range of realistic parameters, the melt-down process is determined mainly by the product μs so μ and s need not be known separately.

Deleterious Mutations in Asexual Populations

Although asexual species are often considered to be evolutionary dead-ends, polygenic mutation provides an evolutionary potential which is sufficient for considerable phenotypic evolution (LYNCH & GABRIEL, 1983). Nevertheless, a severe handicap for asexual species is the accumulation of unconditionally deleterious mutations. If, by chance, the genotype with the fewest deleterious mutations does not contribute to the successful offspring in the next generation, then this genotype is removed from the population forever. Eventually, the second best genotype will have the same fate of being lost from the population - and so on. MULLER (1964) first noticed this process, which has become known as "MULLER's ratchet". The velocity at which the ratchet turns, depends on the population size, mutation rate, and selection coefficient.

Earlier theory and simulations of the ratchet (e.g. MAYNARD SMITH 1978, BELL 1988) kept the (effective) population size constant. This led to the prediction that the ratchet is less effective for higher selection coefficients; i.e., the average time for the ratchet to make one turn increases with increasing s. However, by making the population size dependent on mutation load, LYNCH & GABRIEL (1990) found that the mean extinction time declines as mutations become more deleterious. This means that the lower speed of the ratchet under stronger selection (higher s values) is more than compensated for by the greater reduction in survivorship per turn of the ratchet. A synergistic interaction between mutation load and random genetic drift is responsible for this process. As deleterious mutations reduce the number of surviving offspring, random genetic drift becomes of greater importance and facilitates the fixation of further deleterious mutations. This "mutational melt-down" eventually leads to population extinction.

Figure 1 gives estimates of extinction time for asexual populations under a low mutational load of $\mu s = 0.0002$ for various intrinsic growth rates r. These results are obtained by a model with very simple population density regulation and non-overlapping generations. Each member of the population can produce $R = e^r$ offspring on average but offspring numbers are Poisson distributed around this expectation. The population size is restricted to K individuals (= carrying capacity). If the total number of offspring exceeds the carrying capacity, K individuals are drawn randomly to start the next generation. The accumulated mutations in each offspring determine its probability of survival until reproduction. For small μs, the combined effect of these random processes can be

approximated by a deterministic solution which can easily be calculated by iterations (for details, see LYNCH & GABRIEL 1990).

The results of this conservatively simple model, which ignores all environmental sources of mortality, suggest that the survival times of clonal lineages of a carrying capacity $K < 10^7$ are unlikely to exceed 10^4 generations. This view is consistent with molecular data that suggest that most parthenogenetic animals are phylogenetically young (BELL 1982, LYNCH 1984). It is in contradiction to the existence of a few very old obligate parthenogenetic groups (e.g. bdelloid rotifers). Assuming errors have not been made in the identification of the breeding system of such groups, their escape from mutational melt-down may be a consequence of a high incidence of compensatory mutation. LYNCH & GABRIEL (1990) modelled the melt-down process using a distribution of mutational effects with a constant mean. They found that the longevity of asexual lineages can be enhanced dramatically if the variance in s becomes large enough so that some mutations are beneficial.

It should be mentioned here that the above statements on MULLER's ratchet for the case of constant mutational effect rely on the assumption of unconditionally deleterious mutations. This means that mutational effects are independent of the actual genetic background. An alternative treatment involves the influence of mutations on quantitative traits (GABRIEL & WAGNER 1988; WAGNER & GABRIEL 1990). In this case, fitness is determined by several quantitative traits, each trait of which is controlled by many loci. A single pleiotropic mutation can affect each trait simultaneously. Since polygenic mutations can increase or decrease the value of a trait, deleterious mutations for one trait or locus can be compensated for by advantageous mutations for others, even when the average effect of

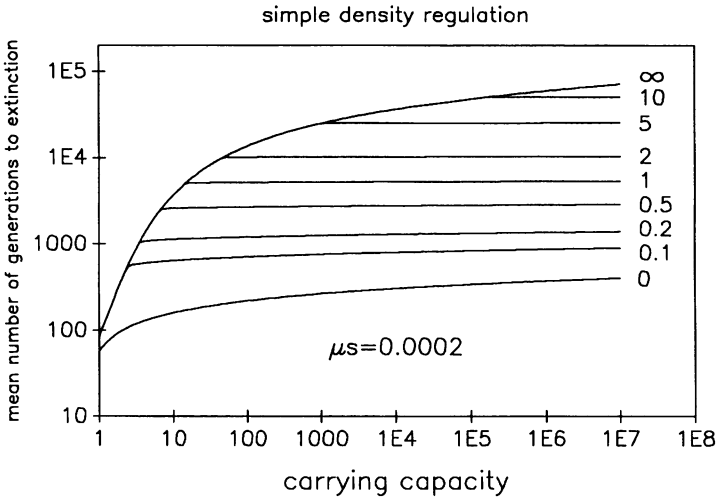

Fig. 1: Survival of parthenogenetic populations under mutational load (μs = 0.0002) and simple density- dependent population regulation (see text). The mean number of generations to extinction is calculated as a function of carrying capacity K for various intrinsic growth rates r (in units per generation as indicated by the numbers in the graph).

each mutation is a reduction of fitness. This model is a quantitative genetic analog to MULLER's ratchet. Simulations and mathematical analyses show that the compensatory mutations inherent under this scenario are as effective as recombination in halting the decline of fitness caused by MULLER's ratchet.

A Simple Logistic Model with Stochasticity in Sex-Ratio and Fecundity

The simple population regulation described in the previous section is not very realistic but sufficient for initial insight into the interplay between genetics and population dynamics. In the following we will use a model with a logistic type of population growth. This also can be parameterized by the carrying capacity K and the growth rate r. (K is the population size, to which an undisturbed population would converge during time; r is equivalent to the intrinsic rate of exponential growth at small population size.) These parameters are usually used in differential-equation models where time is a continuous variable. An analogous difference-equation description for discrete generations is

$$N_{(t+1)} = N_{(t)}\, e^r\, [1 + N_{(t)}(e^r - 1)/K]^{-1}. \qquad (4)$$

With this equation there are no periodic orbits or chaotic behavior (for details see MAY 1981). Instead, convergence to K occurs in a monotonic way.

We use equation (4) to calculate the expected number of offspring per female. For asexual populations mean family size would be just $N_{(t+1)}/N_{(t)}$. For sexual populations we assume an expected ratio of females to males of 1:1. In order to achieve the same population regulation as in the asexual case, each female has to produce $2N_{(t+1)}/N_{(t)}$ offspring on average. The actual number of newborns is drawn from a Poisson distribution. Each female randomly chooses a mate, and the offspring's genome is constructed by free recombination. For this purpose, each mutation and its locus is stored. Every new mutation is assumed to occur at a new locus.

Thus, the model we now consider involves logistic growth but with stochasticity in fecundity and in the sex-ratio. There are two potential causes of extinction: either the number of surviving offspring is zero by chance, or mating is impossible because there are only males or only females left.

The Risk of Extinction from Random Variation in Fecundity and Sex-Ratio without Mutational Load

Before we study the combined effect of demographic stochasticity and mutational load, we analyze the risk of extinction without mutations. To get reference points for evaluating the relative importance of the underlying processes, we first look at the consequences of sex-ratio fluctuation when the population size is kept at the carrying capacity. Then,

neglecting the sex-ratio fluctuations, we study the effect of stochastic fecundity in a monoecious population with density- dependent offspring number. Finally, we calculate the expected extinction times under the simultaneous operation of both stochastic processes for logistic population regulation.

The extinction probabilities due to sex-ratio fluctuations can easily be calculated for constant population sizes. (Further details are given elsewhere; see GABRIEL and BÜRGER, submitted). For a population of size K the mean time to extinction due to sex-ratio variation is

$$t_E = 2^{K-1} + 1, \qquad (5)$$

given an expected sex-ratio of 1:1. The extinction times are geometrically distributed so that the standard deviation is equal to the expectation.

Without sex but under stochastic fecundity and logistic population regulation, there exists no simple analytic expression for the distribution of extinction times. For the combined process, it is even more hopeless to find analytical solutions other than approximations for special cases. Therefore, to get reliable results, we applied two

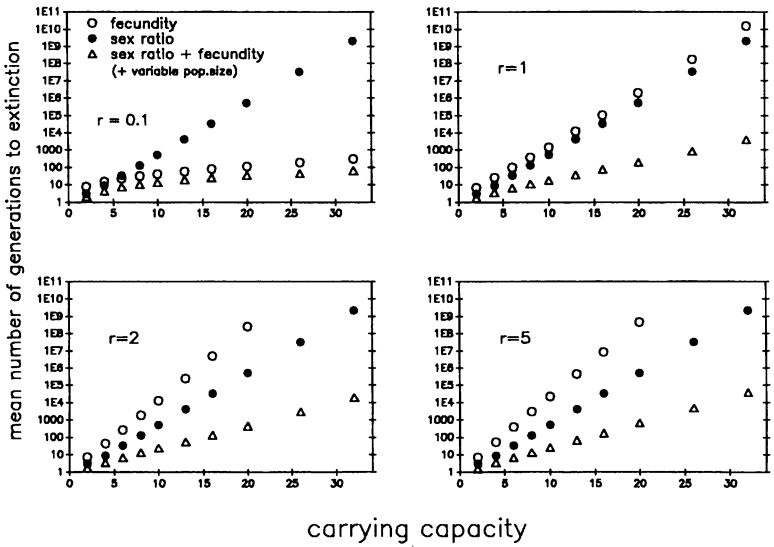

Fig. 2: Extinction times due to stochastic fecundity and/or stochastic sex-ratio depending on carrying capacity K and intrinsic growth rate r (which is measured in units of inverse generation time; therefore, r = 1 implies 2.72 offspring and r = 5 leads to 148 offspring on average.). The values for stochastic sex-ratio (closed circles) are calculated for constant population size (= K). For stochastic fecundity (open circles), the expected number of offspring is regulated by logistic growth but the number of offspring produced by the individual females fluctuates around this expectation value according to a Poisson distribution. The populations start in the first generations with K individuals. The results of the combined processes of stochastic sex-ratio and stochastic fecundity are given by the triangles.

independent methods: a) straight-forward Monte Carlo simulations, and b) description of the processes by Markov chains and numerical solution of the corresponding (quite large) systems of equations. For details see GABRIEL and BÜRGER (submitted). We obtained identical results from both methods so that we can be sure that there are no errors in the program for the Monte Carlo simulation and that we did not run into numerical problems in the solution of the equations of the Markov chain model.

Figure 2 compares the extinction times under the influence of stochastic sex-ratio alone, stochastic fecundity alone, and under the combined action of both for various growth rates and carrying capacities, with the populations always starting at the carrying capacity (K). Only for small r is the probability of extinction due to sex-ratio variation larger than that due to stochastic fecundity. For increasing r the risk of extinction due to stochastic fecundity converges rapidly to an analytically calculable limit, e.g. mean time to extinction converges to e^K if only stochastic fecundity is considered (see GABRIEL and BÜRGER, submitted).

There is of course no linear interaction between the pure sex-ratio and fecundity risks because in the combined process the risk due to sex-ratio is not determined by the carrying capacity K but by the actual population size in each generation. An analysis of causes for extinction shows that the probability of population extinction due to non-surviving offspring becomes very small as the carrying capacity increases. Figure 3 shows the extinction risks due to the combined stochasticity of fecundity and sex-ratio depending on K for various r values.

One should keep in mind that the plotted numbers are expected times to extinction. The corresponding distributions of extinction probabilities are very broad since they follow roughly a geometric distribution. It should also be kept in mind that the extinction time

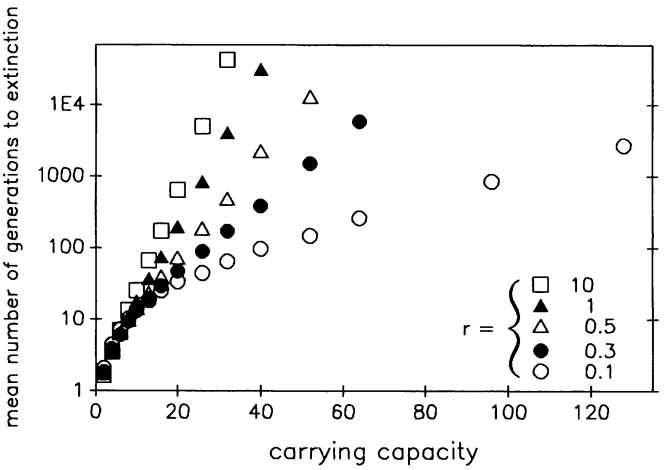

Fig. 3: Extinction risk due to stochastic sex-ratio and stochastic fecundity under logistic growth as in Figure 2 for various intrinsic growth rates.

depends on the initial population size N_0 which for all results presented here was assumed to be equal to K. Under the logistic population regulation used in this study, the mean extinction time increases monotonically as the initial population size increases and (if $r > 0.5$) rapidly reaches an upper limit, which is almost identical with the values for populations starting at carrying capacity as presented here. Only for very small r and very small initial population size ($N_0 < 4$) is the corresponding extinction time considerably smaller than for $N_0 = K$ (see GABRIEL & BÜRGER submitted).

Mutational Melt-down in Sexual Populations

One might expect that the process of mutational melt-down, as discussed in the previous section for asexual organisms, is not relevant under sexual reproduction because selection and recombination can eliminate bad mutations. But for stochastic logistic population regulation, it is critical to check first how large a carrying capacity has to be in order to produce a sufficiently large effective population size so that selection and recombination will be efficient enough to prevent the accumulation of deleterious mutations. In sexual populations the melt-down process can be effective if the population is temporarily reduced to a size at which the probability of extinction due to sex-ratio imbalance becomes important.

The risk of extinction for sexual populations due to mutational load is demonstrated in Figure 4. Except for very small carrying capacities, the mean times to extinction are reduced by several orders of magnitude relative to the extinction times without mutational load. Preliminary results indicate that this is true also for mutation loads at the lower end of the estimates given in equation (3).

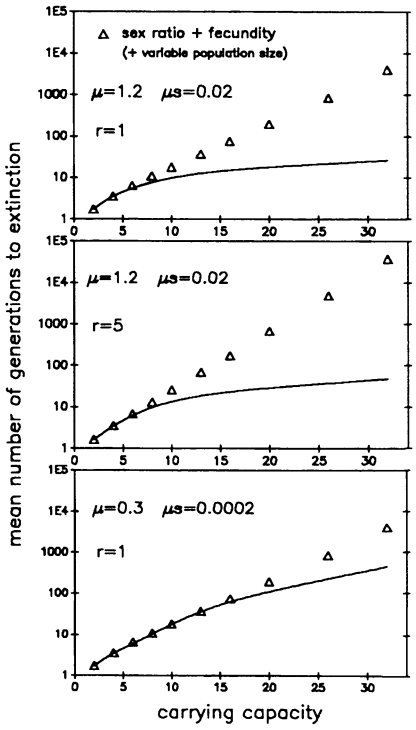

Fig. 4: Survival of sexual populations under mutational load and demographic stochasticity (=solid lines). Intrinsic growth rate r, mutational load (μs), and mutation rates μ are given in each panel. The triangles indicate the corresponding extinction risks without mutational load. (The results for μs = 0.0002 are preliminary.)

Conclusions

The joint action of deleterious mutation and demographic stochasticity has a deleterious synergistic effect which can lead to a mutational melt-down and ultimately to extinction. This process cannot be analyzed in the usual tradition of population genetics which keeps the (effective) population size constant. Evaluation of the effect of deleterious mutation on population viability must be treated in the context of density-dependent population regulation and should allow for demographic variation in family size and sex-ratio.

Since we have neglected risks such as lethal mutation and environmental sources of mortality, times to extinction calculated in this paper are definitely underestimates. For real populations, growth rates and carrying capacities are not constant but more or less dependent on variable abiotic and biotic factors of the environment. Such additional risks are also expected to interact synergistically with unfavorable genetic processes.

Our quantitative results on the impact of mutational load on population extinction are based on estimates of the mutation load derived from experiments which were not primarily designed to measure the mutational melt-down. However, unless the existing estimates of mutational load are greatly exaggerated, the conclusion that the accumulation of deleterious mutations is an important determinant of population extinction seems inescapable for populations with upper size limits of several dozen or smaller. Theoretical studies which combine population genetics with population dynamics are fundamental to the field of conservation biology.

Zusammenfassung

Um die Aussterbezeiten kleiner Populationen abzuschätzen, ist es im Gegensatz zu einer weitverbreiteten Meinung notwendig, auch genetische Faktoren zu berücksichtigen. Dies wird am Beispiel von Mutationen mit nur geringer schädlicher Wirkung demonstriert. Mit Hilfe vorhandener Abschätzungen über Mutations- raten und Mutationseffekte wird untersucht, wie sich solche Mutationslast in Populationen mit dichteabhängigem Wachstum und unter demographischer Stochastizität auswirkt. Die Anhäufung von Mutationen setzt die Überlebenswahrscheinlichkeit der Individuen herab und kann so zu einer zeitweisen Verringerung der Populationsgröße führen, die ihrerseits die Fixierung weiterer schädlicher Mutationen durch genetische Zufallsdrift erleichtert. Diese synergistiche Interaktion wird "mutational melt-down" genannt.

In parthenogenetischen Populationen liegt die "proximate" Ursache für das Aussterben darin, daß auf Grund von Zufallsprozessen keine Nachkommen überleben beziehungsweise geboren werden. Die Wahrscheinlichkeit dafür steigt mit zunehmender Mutationslast und mit abnehmender aktueller Populationsgröße. Für sexuelle Populationen werden als

Referenzpunkte zunächst die Aussterbezeiten unter logistischer Populationsregulation ohne Mutationslast in Abhängigkeit von Wachstumsrate und Kapazität ("carrying capacity") bestimmt. Dabei wirken als stochastische Größen nur Zahl und Geschlecht der Nachkommen. In kleinen sexuellen Populationen können Rekombination und Selektion einen "mutational melt-down" nicht verhindern: unter der Wirkung schwach schädlicher Mutationen verkürzt sich die Aussterbezeit um mehrere Größenordnungen. Entscheidend ist dabei nicht das Ausbleiben von überlebenden Nachkommen sondern die durch temporäres Absinken der Populationsgröße erhöhte Wahrscheinlichkeit, daß nur männliche oder nur weibliche Nachkommen erzeugt werden.

Zur genaueren Abschätzung der Aussterbewahrscheinlichkeit kleiner Populationen sind weitere experimentelle Untersuchungen zur Mutationslast und theoretische Studien zur Interaktion von Populationsgenetik und Populationsdynamik dringend erforderlich.

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