

Mutation and Migration

- HWE requires that there be no mutation and no migration.

I) Sources of Genetic Variation – there are several way to “cast” mutations role

- Basically mutation is the primary source of new genetic variation.
- μ is the rate of mutation measured in the number of mutations per gene per generation.
 - ❖ Rate varies considerable between nucleotides within a gene, between genes within an organism, between organisms.
 - ❖ Typically rates are 1×10^{-3} to 1×10^{-9} mutations per gene per generation.
- μ is a stochastic process similar to radioactive decay not a metronomic.

A) Irreversible Mutation – assume no selection and infinite population size.

- $A \Rightarrow a$ but $a \neq X \Rightarrow A$

$$p_t = p_{t-1}(1 - \mu)$$

Basic expansion:

$$p_t = p_0(1 - \mu)^t$$

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B) Reversible Mutation – assume no selection and infinite population size.

➤ **A => a at a rate of μ**

➤ **a => A at a rate of ν**

$$p_t = p_{t-1}(1-\mu) + (1-p_{t-1})\nu$$

didn't change a => A

$$p_t = p_{t-1} - \frac{\nu}{\mu + \nu} (1 - \mu - \nu) + \frac{\nu}{\mu + \nu}$$

$$p_t = p_0 - \frac{\nu}{\mu + \nu} (1 - \mu - \nu)^t + \frac{\nu}{\mu + \nu}$$

$$\text{as } t \Rightarrow \infty (1 - \mu - \nu)^t \Rightarrow 0.0$$

$$p_e = \frac{\nu}{\mu + \nu}$$

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C) Fixation of Mutations – we will revisit this more completely when we formally treat drift, but for now...

➤ **Frequency of any new mutation is α**

$$F(\alpha) = \frac{1}{2N}$$

$$F(\text{not } \alpha) = 1 - \frac{1}{2N}$$

for a gamete population size of $2N$

When considering the gametes that contribute to the next generation they can be α or not α with the probability of $\frac{1}{2N}$ and $1 - \frac{1}{2N}$, respectively.

Combinations of allele frequencies in the next generation (i.e., expansion of the binomial $\frac{1}{2N} + 1 - \frac{1}{2N}$)^{2N}

$$\frac{n!}{r!(n-r)!} \alpha^r (1-\alpha)^{n-r}$$

where $n = 2N$ and $r = \alpha = \frac{1}{2N}$

probability that there are no α alleles in the next generation:

$$\frac{2N!}{0!(2N-0)!} \frac{1}{2N}^0 \left(1 - \frac{1}{2N}\right)^{2N-0}$$

Note: $\frac{2N!}{2N!} = 1.0$, $0.0! = 1.0$, and $\frac{1}{2N}^0 = 1.0$

$$1 - \frac{1}{2N}^{2N} \approx \frac{1}{e} \approx 0.367879$$

- There is about a 37% chance that a new allele goes to extinction in the first generation after creation (drift).

II) Mutational Models – there are a variety of ways to model the relationship between mutation and alleles.

A) Infinite Allele Model – every mutation creates an allele not already in the population. Most commonly invoked model.

- **therefore alleles IBS are also IBD.**
- **all homozygotes are autozygous.**
- **F = probability of autozygosity.**
- **assume equal gamete production for all adults.**

Frequency of an allele in adults = frequency in gametes = $\frac{1}{2N}$

Two types of homozygotes (probability of autozygosity):

- 1) If I have an allele α , the probability of drawing that allele again (i.e., another copy from the same adult) is $\frac{1}{2N}$. The probability that neither of these mutates is $(1 - \mu)^2$.**
- 2) If I have an allele α' , the probability of drawing different allele from a different individual is $1 - \frac{1}{2N}$. The probability that these allele are IBD = F_{t-1} = autozygosity of the adult generation. The probability that neither of these mutates is $(1 - \mu)^2$.**

Taken together:

$$F_t = \frac{1}{2N}(1 - \mu)^2 + \left(1 - \frac{1}{2N}\right)(1 - \mu)^2 F_{t-1}$$

= autozygous because they are unmutated gametes from the same individual autozygous because they are unmutated identical gametes from different individuals.

if we ignore μ^2 and $\frac{\mu}{N}$ because they are quite small

$$\hat{F} = \frac{1}{4N\mu + 1}$$

Since this is an infinite allele model, \hat{F} = equilibrium frequency of homozygosity and $1 - \hat{F}$ = equilibrium frequency of heterozygotes.

$$1 - \hat{F} = \frac{4N\mu}{4N\mu + 1}$$

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B) Other Models – there are specialized models for different situations. Most are too complicated to derive similar equations for, but generally the relationship between autozygosity, population size, and mutation rate holds

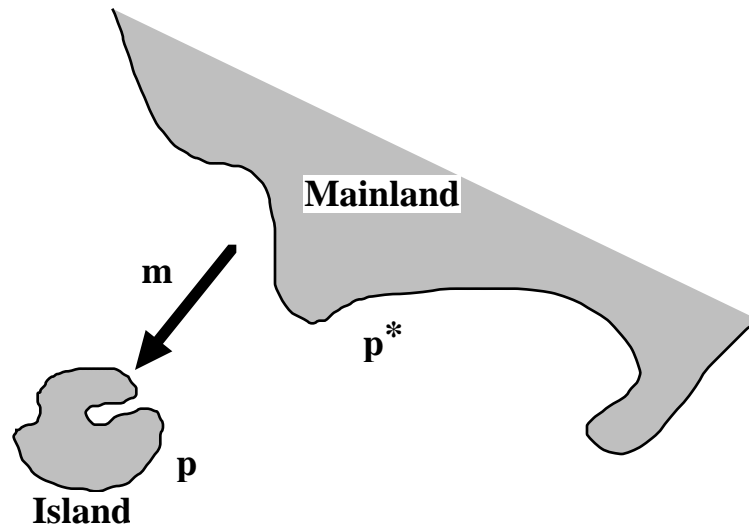
- 1) Stepwise Mutational model – developed for simple sequence repeat loci that increase or decrease by one repeat unit.**
- 2) Two phase mutational model – combination of the stepwise and the infinite allele.**

III) Mutation and Migration – mutation in two populations will cause evolutionary divergence. Migration between them will retard divergence.

A) Migration Models – the form of migration among subpopulations can determine the outcome.

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B) Mutation and Migration in the Island-Continent Model (one way or two way where $m_i \lll m_c$)



$$p' = (1 - m)p + mp^*$$

$$p_t = p^* + (1 - m)^t (p_0 - p^*)$$

$$t = 0, \quad p_t = p_0$$

$$t \Rightarrow \infty, \quad p_t = p^*$$

if $p^* = 0.0$ (i.e., allele is only in island), then:

$$p' = (1 - m)p \quad (\text{assume } p = \frac{1}{2N}, \text{ i.e., new mutation on island})$$

if $m = 0.0$ then proportion of next generation on island that are migrants is 0.0 and if equal contribution from all individuals on island

$$p' = p = \frac{1}{2N}$$

if $m > 0.0$ and $p^* = 0.0$, then individuals on island are a mix of migrants and nonmigrants and:

$$p' = (1 - m)p \quad (\text{frequency of new allele and it is not a migrant})$$

and

$$p_t = p_0(1 - m)^t$$

as $t \Rightarrow \infty$, $(1 - m)^t \Rightarrow 0.0$ and therefore $p_t \Rightarrow 0.0$ (note similarity with irreversible mutation model)

In words, a new allele will be “diluted” each generation by migration and ultimately be lost from the island population.

C) Mutation and Migration and the Propagule Pool Model (AKA Island model).

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- equal migration between all populations
- individuals mate in the home population and cast propagules (or gametes) into common pool in equal proportions. Propagules set in populations with equal probability (i.e., without respect to parental population).
- modification is stepping stone, note edge effects.

$$\bar{p} = \frac{1}{n} \sum_{i=1}^n p_i \text{ where } n = \text{number of populations}$$

$$p_{t,i} = p_{t-1,i}(1 - m) + \bar{p} m \quad (\text{as for island-continent except } \bar{p} \text{ replaces } p^*)$$

$$p_{t,i} = \bar{p} + (1 - m)^t(p_0 - \bar{p})$$

as $t \Rightarrow \infty$, $p_t \Rightarrow \bar{p}$ therefore, all populations converge on the average population allele frequency. This happens very quickly since m is usually relatively large.

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Note: remember $\hat{F} = \frac{1}{4N\mu + 1}$ and it can be shown that $\hat{F} = F_{ST}$ so,

$$F_{ST} = \frac{1}{4Nm + 1} \text{ and } Nm = \frac{1}{4}(F_{ST} - 1)$$

IV) Recombination and Mutation – recombination can also be a strong evolutionary force in that it can create and break up coadapted gene complexes.

➤ assume mutation are favorable and confer increased fitness

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