

Chapter 25

Population Genetics

Population Genetics -- the discipline within evolutionary biology that studies changes in allele frequencies.

Population -- a group of individuals from the same species that lives in the same geographic area and that actually or potentially interbreeds.

Gene Pool -- all gametes made by all breeding individuals of a population in a single generation.

Populations are dynamic: they expand and contract through changes in birth & death rates, migration, or contact with other populations.

The dynamic nature of populations has important consequences and can, over time, lead to changes in a population's gene pool.

Hardy-Weinberg law illustrates what happens to alleles and genotypes in an “ideal” population (free of many of the complications that affect real populations) by using a simplified set of assumptions:

- 1. Population is infinitely large or large enough that sampling error is negligible.**
- 2. Panmixia**
- 3. No selection**
- 4. No mutation**
- 5. No emigration or immigration**
- 6. No genetic drift.**

The Hardy-Weinberg law demonstrates that an “ideal” population has these properties:

- 1. Frequency of alleles does not change from generation to generation.**
- 2. After one generation of random mating, offspring genotypic frequencies can be predicted from the parental allele freq., and would be expected to remain constant.**

$$\bullet p + q = 1$$

$$p = B = 0.8$$

$$1 = b = 0.2$$

$$1.0$$

-- Assuming Panmixia -- determine allele and genotypic frequencies in next generation.

$$\text{Probability of "B" gamete} = 0.8$$

$$\text{Probability of "b" gamete} = 0.2$$

Genotypic frequencies in NEXT generation assuming HWE:

$$BB = (0.8)(0.8) = 0.64$$

$$Bb = 2(0.8)(0.2) = 0.32$$

$$bb = (0.2)(0.2) = 0.04$$

Allele frequencies in NEXT generation:

$$B = 0.64 + (0.32/2) = 0.8$$

$$b = 0.04 + (0.32/2) = 0.2$$

Continued sexual reproduction with segregation, recombination, and panmixia *WILL NOT* alter these frequencies.

Thus, there is no evolution and the population *IS* in Hardy-Weinberg Equilibrium.

Hardy-Weinberg Equations:

$$p = B \qquad q = b$$

$$p + q = 1$$

$$p^2 + 2pq + q^2 = 1$$

(BB) (Bb) (bb)

Example:

-- phenylketonuria -- recessive trait

--U.S. -- 1/1000

$$--q^2 = 1/10,000 \text{ or } 0.0001$$

$$--q = q^{(0.5)} = 0.01$$

$$-- p = 1 - q = 1 - 0.01 = 0.99$$

Should be able to do the same is given:

-- frequency of a dominant phenotype:

$$[p^2 + 2pq + q^2 = 1]$$

-- sex-linked traits

$$\text{females } q^2 \quad \text{males } q$$

-- more than two alleles.

$$p + q + r = 1$$

genotypic frequencies = $(p + q + r)^2$

$$p^2 + q^2 + r^2 + 2pq + 2pr + 2qr = 1$$

Example:

$$A = 0.53, B = 0.13, AB = 0.08, O = 0.26$$

Because O is due to recessive allele,

$$O = 0.26 = r^2 \text{ and therefore, } r = 0.51$$

We can use the value for “r” to determine the allele frequencies of “A” and “B” as:

Combined frequency of type “A” an “O” blood is: $p^2 + 2pr + r^2 = 0.53 + 0.26 = 0.79$

$$(p + r)^2 = 0.79$$

$$p + r = (0.79)^{0.5} = 0.89$$

$$p = 0.89 - 0.51 = 0.38$$

Knowing p and r, we can now calculate q as:

$$p + q + r = 1$$

$$q = 1 - p - r$$

$$q = 1 - 0.38 - 0.51 = 0.11$$

Important Consequences of Hardy-Weinberg law:

1. shows that dominant traits do not necessarily increase from one generation to the next.

2. demonstrates that genetic variability can be maintained in a population since, once established in an ideal population, allele frequencies remain unchanged.

3. if we invoke the assumptions of HWE, then knowing the frequency of just one genotype enables us to calculate the frequency of all other genotypes.

4. the most important role of Hardy-Weinberg law is that it is the foundation upon which population genetics is built.

Factors that disrupt HWE and cause evolution:

Natural Selection:

One assumption of HWE is that individuals of all genotypes have equal rates of survival and equal reproductive success.

**Example: a population of 100 individuals,
 $p = A = 0.5$; $q = a = 0.5$.**

Assuming the previous generation mated randomly, genotypic frequencies and number of individuals of each genotype in this generation are:

$$\mathbf{AA = (0.5)^2 = 0.25 * 100 = 25}$$

$$\mathbf{Aa = 2 * 0.5 * 0.5 = 0.5 * 100 = 50}$$

$$\mathbf{aa = (0.5)^2 = 0.25 * 100 = 25}$$

Now suppose that individuals with different genotypes have different rates of survival:

$$\mathbf{AA = 100\% \text{ survival} = 25 \text{ individuals}}$$

$$\mathbf{Aa = 90\% \text{ survival} = 45 \text{ individuals}}$$

$$\mathbf{aa = 80\% \text{ survival} = 20 \text{ individuals}}$$

When the survivors reproduce, each contributes 2 gametes so the total number of gametes is:

$$\mathbf{50 + 90 + 40 = 180.}$$

What are the allele frequencies in the population now?

$$p = A = (50 + 45)/180 = 0.53$$

$$q = a = 1 - 0.53 = 0.47$$

These differ from the frequencies we began with.

Allele A has increased and allele a has decreased.

Natural Selection is a difference among individuals in survival or reproduction rate (or both).

Natural Selection is the principal force that shifts allele frequencies in LARGE populations and is one of the most important factors in evolutionary change.

Selection occurs whenever individuals with a particular genotype have an advantage in survival or reproduction over other genotypes.

Selection can vary from < 1% to 100% for lethal genes.

Advantages in survival and reproduction ultimately translate into increased genetic contributions to future generations.

An individual's genetic contribution to future generations is called its FITNESS.

Hardy-Weinberg analysis allows us to examine fitness.

w = fitness thus, w_{AA} represents the relative fitness of genotype AA, w_{Aa} the relative fitness of genotype Aa, and w_{aa} the relative fitness of genotype aa.

Assigning the values of $w_{AA} = 1.0$, $w_{Aa} = 0.90$, and w_{aa} of 0.80 would mean that all individuals of AA survive, 90% of Aa survive, and 80% of aa individuals survive.

Example -- selection against a deleterious allele.

fitness values $w_{AA} = 1$, $w_{Aa} = 1$, $w_{aa} = 0$

This describes the situation in which allele “a” is a lethal recessive.

As homozygous individuals die without leaving offspring, the frequency of “a” will decline.

The decline in frequency of “a” is described by the equation:

$$q_g = q_0 / (1 + q_{g0})$$

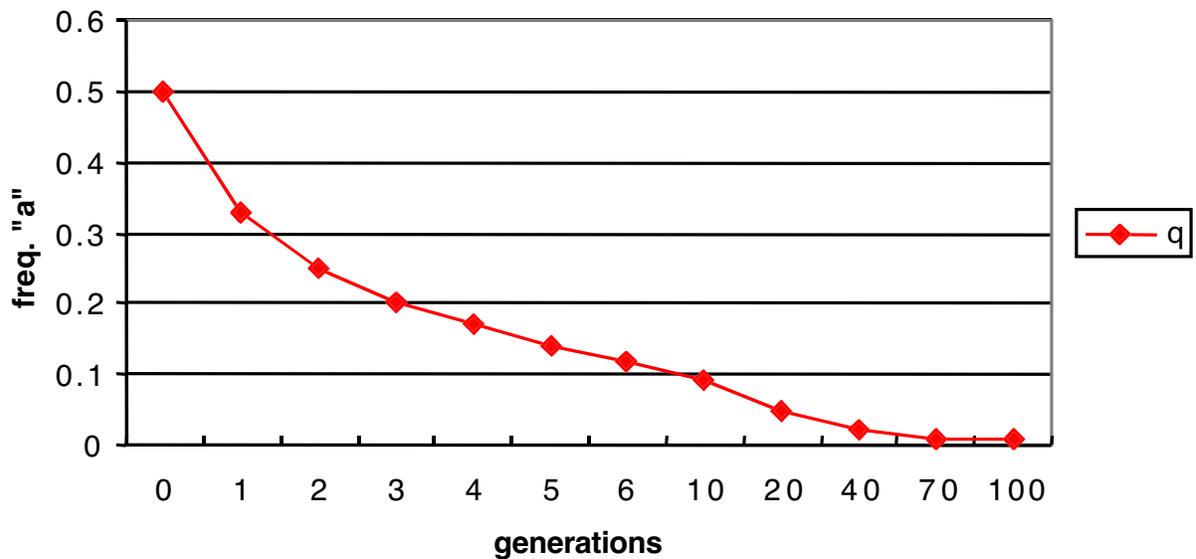
where:

q_g = frequency of “a” in generation g

q_{g0} = starting frequency of “a”

g = number of generations.

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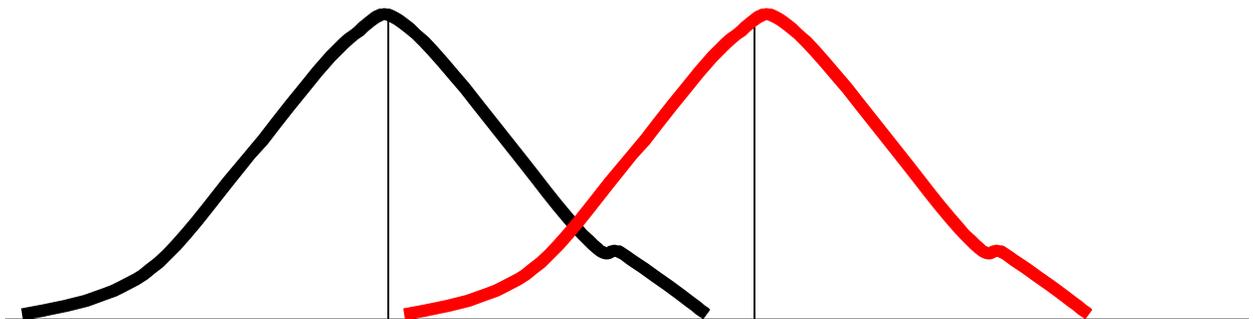
As long as heterozygotes continue to mate, it is difficult for selection to completely eliminate a recessive allele.

A deleterious allele need not be recessive, many other scenarios exist.

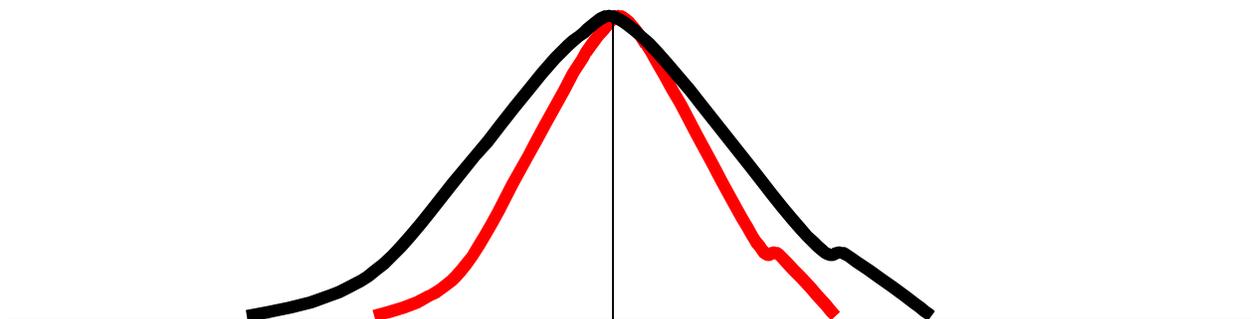
A deleterious allele may be codominant, so that the heterozygotes have intermediate fitness or an allele may be deleterious in the homozygous state but beneficial in the heterozygous state such as with sickle-cell anemia.

Natural Selection and Quantitative Traits

Directional Selection -- Important to plant and animal breeders. Usually selecting for extreme traits.



Stabilizing Selection -- Favors intermediate phenotypes with extreme phenotypes at both ends being selected against.

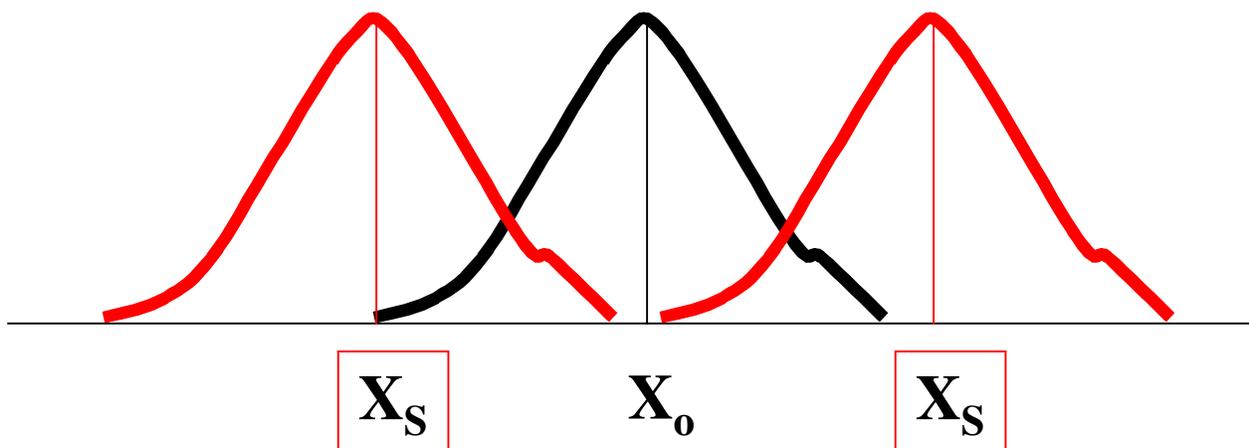


X_0
 X_S

Example of Stabilizing Selection -- Human birth weights

- 11 year study examined 13,730 births.
- Infant mortality increased on both sides of a mean birth weight of 7.5 pounds.

Disruptive Selection -- Selection AGAINST intermediates.



***Mutation* -- Only source of NEW genetic variation for a species.**

Mutation by itself has little effect on large populations.

***Migration of Gene Flow* -- movement of fertile individuals or the transfer of gametes between populations.**

Natural populations will gain or lose alleles by gene flow since they are not closed systems.

$$p = B; q = b$$

Change in frequency of “B” in one generation is:

$$\Delta p = m(p_m - p)$$

p = freq. of “B” in existing population

p_m = freq. of “B” in immigrants

m = coefficient of migration, proportion of migrant genes entering the population each generation

Example: $p = 0.4$; $p_m = 0.6$

10% of adults giving rise to the next generation are immigrants ($m = 0.1$)

$$\Delta p = m(p_m - p)$$
$$0.1(0.6 - 0.4) = 0.2$$

In the next generation:

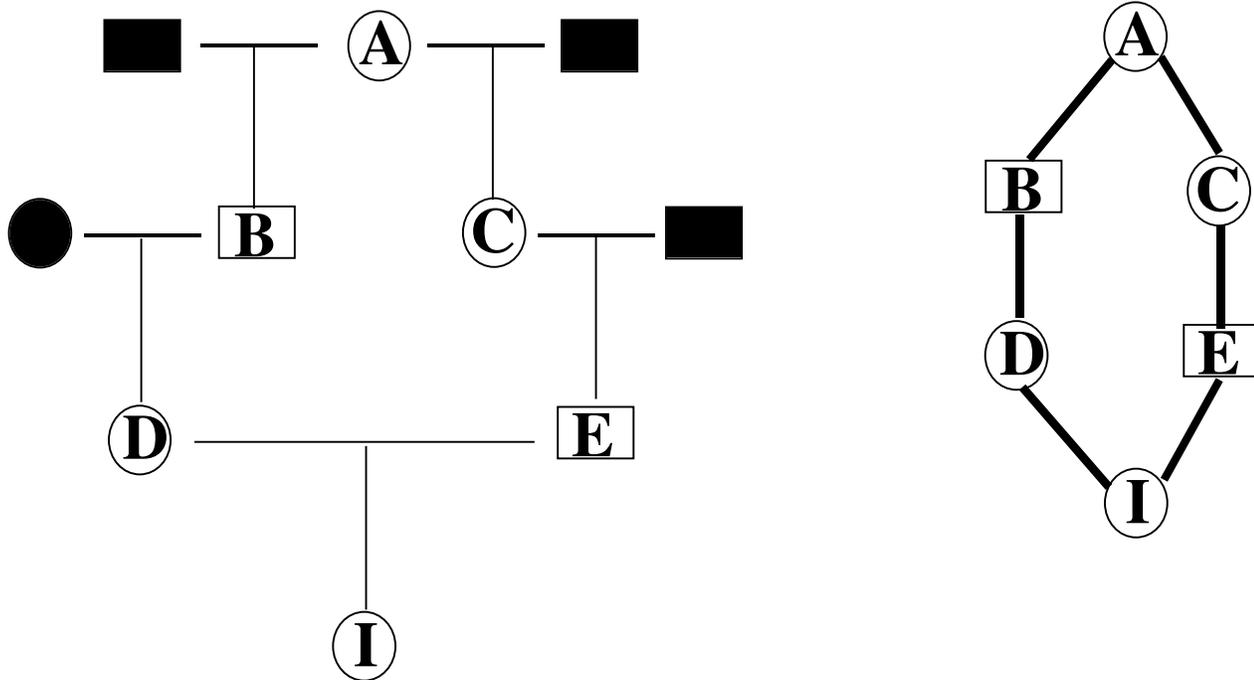
$$p_1 = p + \Delta p$$
$$= 0.4 + 0.2 = 0.42$$

Inbreeding -- when mating occurs between close relatives.

-- Self-fertilization is the most extreme example of inbreeding.

--Inbreeding will result in genotypic ratios ($p^2 + 2pq + q^2$) that deviate from HWE but does NOT alter frequency of p or q .

Calculation of Inbreeding from Pedigrees

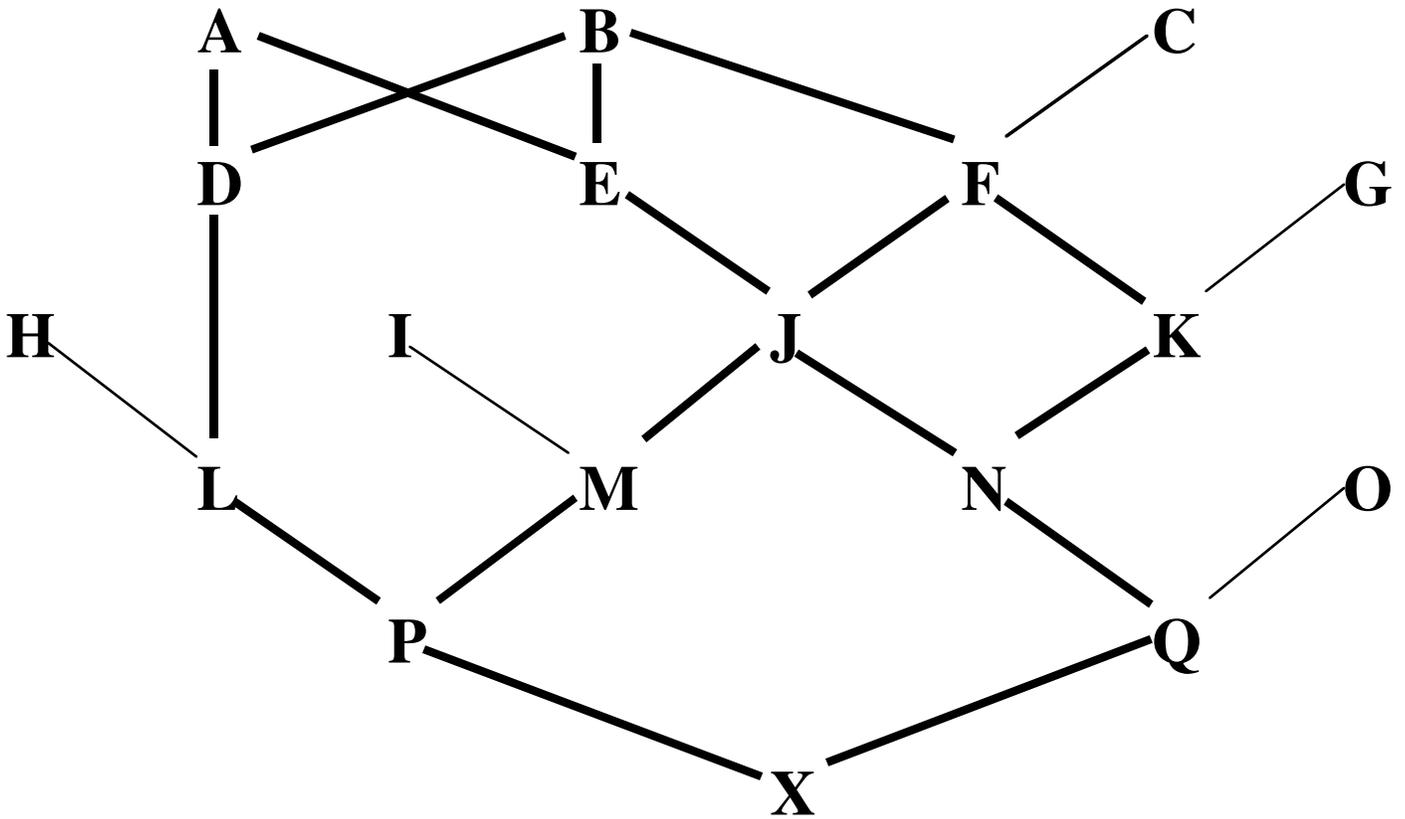


The inbreeding coefficient of I (F_I) can be calculated from path analysis.

The paths are: D-B-A-C-E

$$F_I = (1/2)^i(1 + F_A)$$

If we assume no previous inbreeding in A then this reduces to $F_I = (1/2)^5 = 1/32$



PATHS	N	F_A	Contribution to F_X
PLD<u>A</u>EJNQ	8	0	(1/2)⁸ = 0.0039
PLD<u>B</u>EJNQ	8	0	(1/2)⁸ = 0.0039
PLD<u>B</u>FJNQ	8	0	(1/2)⁸ = 0.0039
PLD<u>B</u>FKNQ	8	0	(1/2)⁸ = 0.0039
PMJ<u>E</u>BFKNQ	9	0	(1/2)⁹ = 0.0020
PMJ<u>F</u>KNQ	7	0	(1/2)⁷ = 0.0078
PM<u>J</u>NQ	5	1/8	(1/2)⁵ X 9/8 = 0.0352
			F_X = 0.0606 19