

Variation in Populations



Phenotypic variation is common in natural populations

Some phenotypic variation is due to genetic differences and some is due to environmental differences





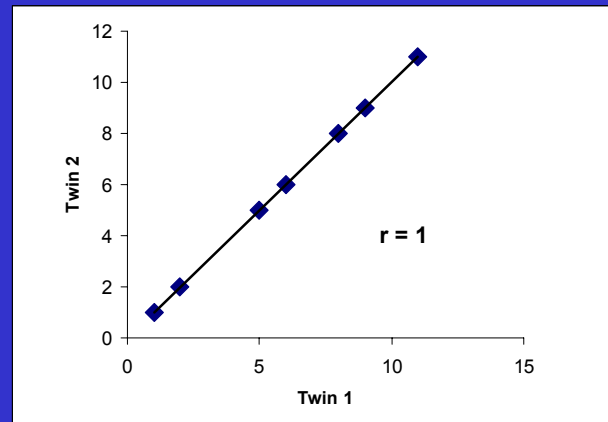
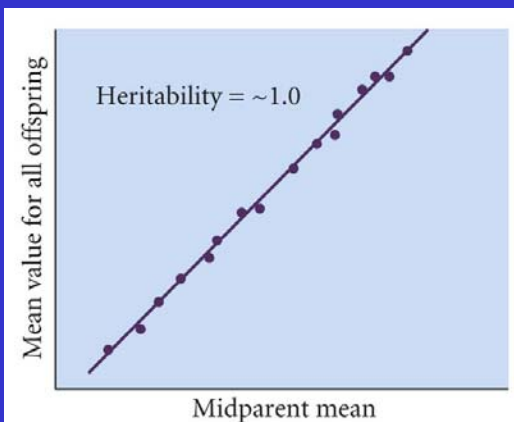
Commonly, phenotypic variation has an underlying genetic basis but is polygenic with many genes influencing the phenotype.

Variation that has an underlying genetic basis is important evolutionarily. The frequency of different forms of a gene can change.

Distinguishing between genetic and environmental sources of variation can be accomplished in several ways:

1. **Controlled crosses** - crossing individuals of different phenotypes to produce an F1 and F2 generation can show that variation segregates in a Mendelian fashion (3:1, 1:2:1, 9:3:3:1, etc.)

2. **Correlation between relatives.** On average parents and children share half their genes. Genetically identical twins should be identical for genetically determined traits.



Ideally, relatives should be reared in different environments to distinguish genetic and environmental sources of variation..

3. **Common garden study** - variation seen between two different natural populations could be due to genetic differences or due to the differences in the environments that influence the phenotype.

To distinguish between them, individuals from both populations can be reared in a common environment.

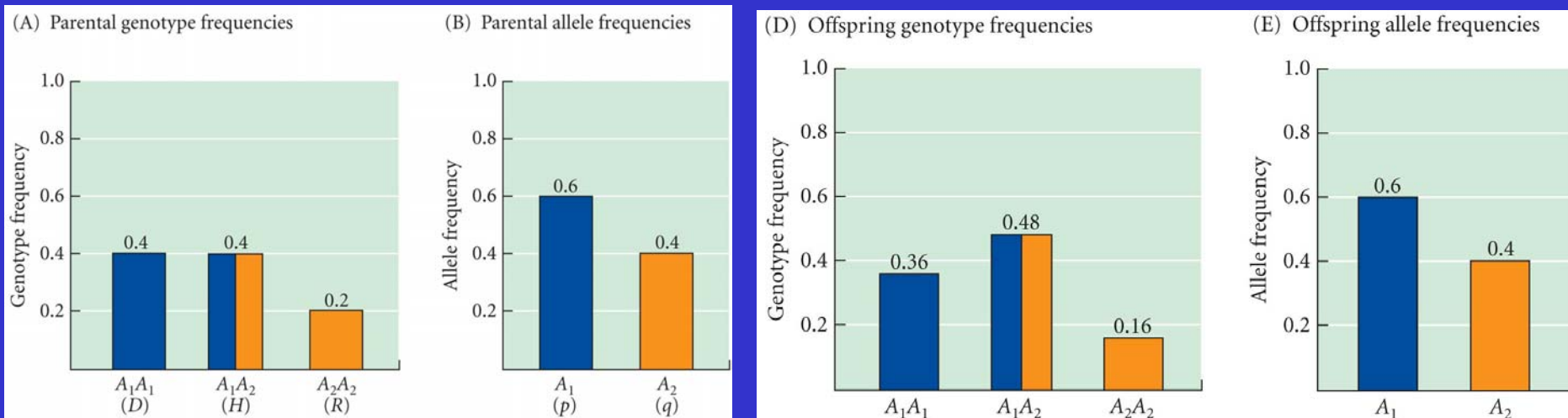
	<u>Lake Verret</u>	<u>Reelfoot Lake</u>
Wild Offspring Mass	0.81 mg	0.93 mg
Common Garden Mass	0.66 mg	0.79 mg



The difference in offspring size remains (there is a genetic basis for the difference) but the offspring were not as large as found in natural populations (there is also an environmental effect on offspring size).

How Genetic Variation Behaves in Natural Populations - the Hardy Weinberg Principle.

Regardless of the initial distribution of genotypes, one generation of random mating will produce a binomial distribution of alleles among genotypes - and this distribution will persist indefinitely ...



Offspring	Probability of a given mating producing the genotype
A_1A_1	$\Pr[A_1 \text{ egg}] \times \Pr[A_1 \text{ sperm}] = p \times p = p^2$ 0.6^2 = 0.36
A_1A_2	$\left\{ \begin{array}{l} \Pr[A_1 \text{ egg}] \times \Pr[A_2 \text{ sperm}] = p \times q = pq \\ \Pr[A_2 \text{ egg}] \times \Pr[A_1 \text{ sperm}] = q \times p = pq \end{array} \right\} = 0.48$
A_2A_2	$\Pr[A_2 \text{ egg}] \times \Pr[A_2 \text{ sperm}] = q \times q = q^2$ 0.4^2 = 0.16

... so long as mating is random, the population is very large, there is no input of alleles from other populations, there is no mutational change in alleles, and there is no differential survival or reproduction of different genotypes.

Violation of any of these requirements can result in a distribution of genotypes other than binomial or can result in a change in the frequencies of alleles from one generation to the next, i.e. evolution.

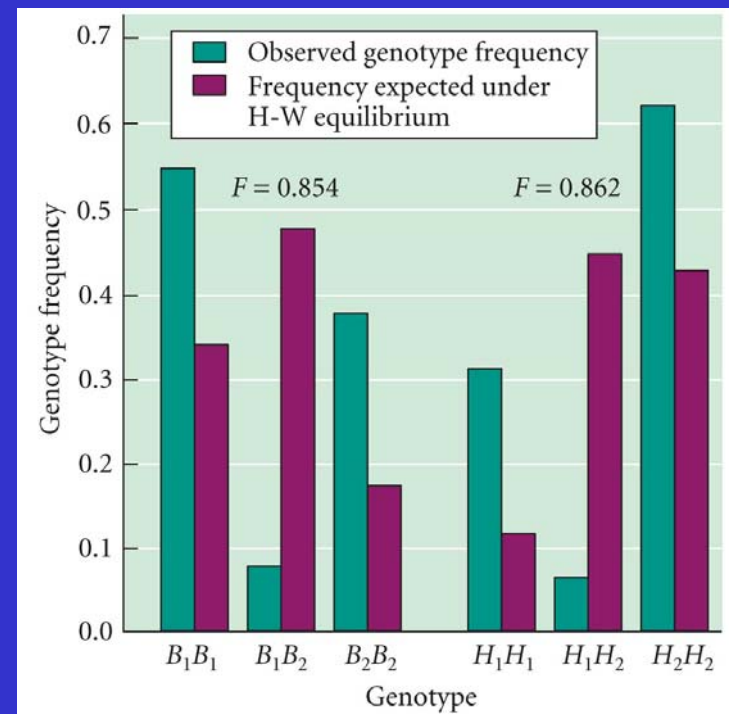
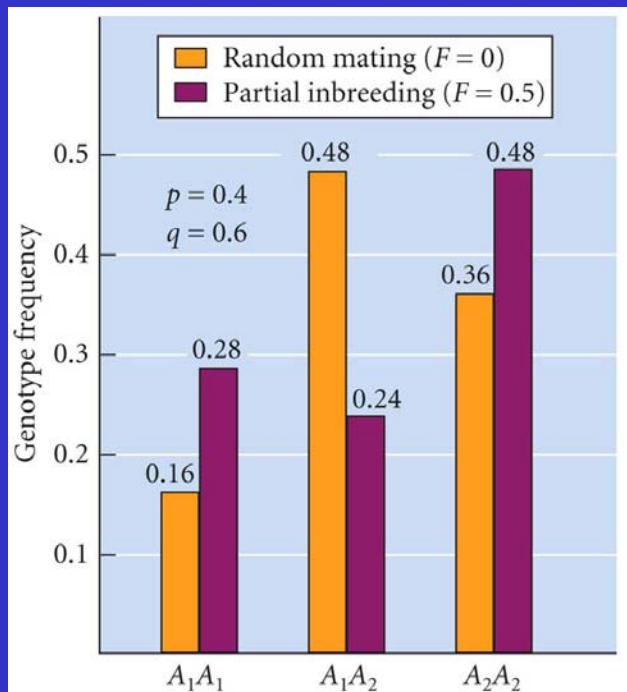
Non-random mating can take several forms - like mating with like, like mating with non-like, or any active preference of certain phenotypes for mates.

Inbreeding - the mating of close relatives - is a type of nonrandom mating that can be detected by examining genotypic frequencies - the frequency of heterozygotes is less than expected for a randomly mating population.

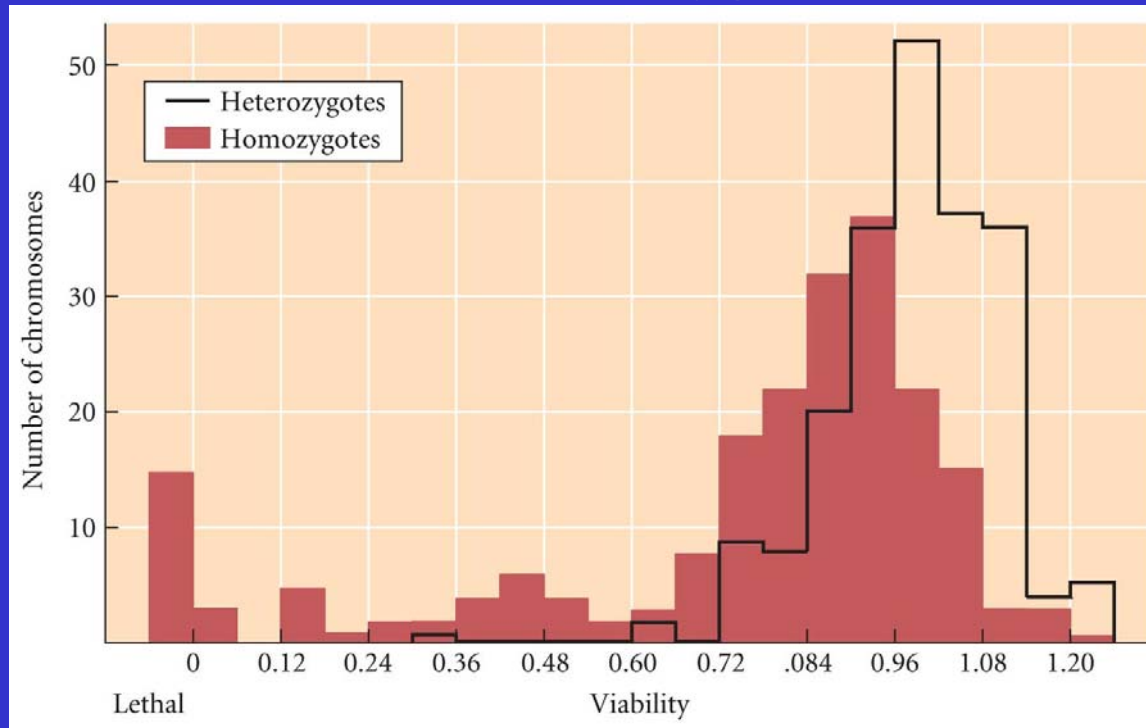
The difference between the observed frequency of heterozygotes (H) and the expected frequency of heterozygotes (H_0 - if mating was random) relative to the expected frequency (H_0) is a measure of inbreeding - the inbreeding coefficient (F).

$$F = \frac{H_0 - H}{H_0}$$

If inbreeding is the cause of the heterozygote deficiency, then F should be nearly the same for all genes.

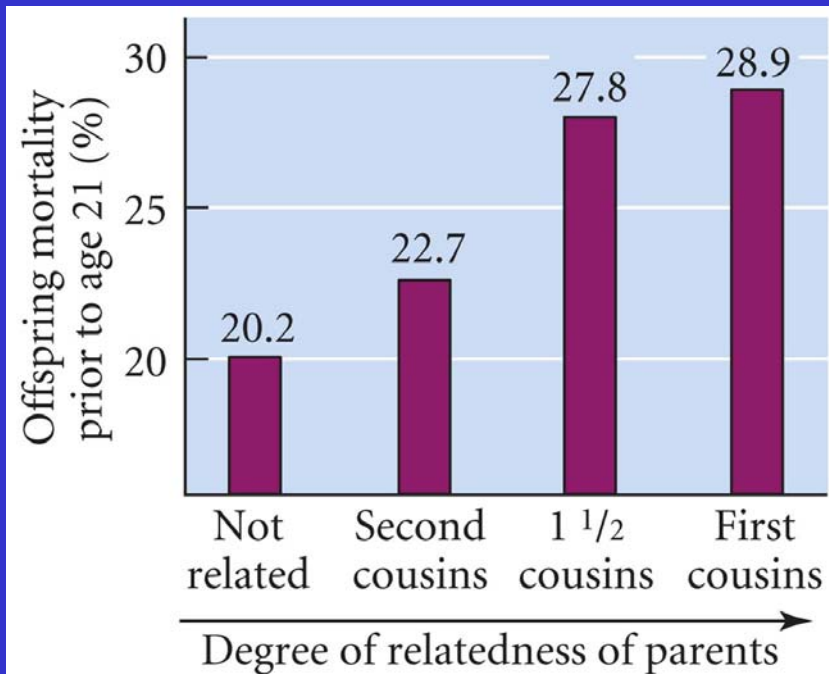


Studies of inbred lines of *Drosophila* revealed that heterozygotes often have higher viability than homozygotes. There is a large amount of deleterious genetic variation in natural populations that is not expressed in heterozygotes.

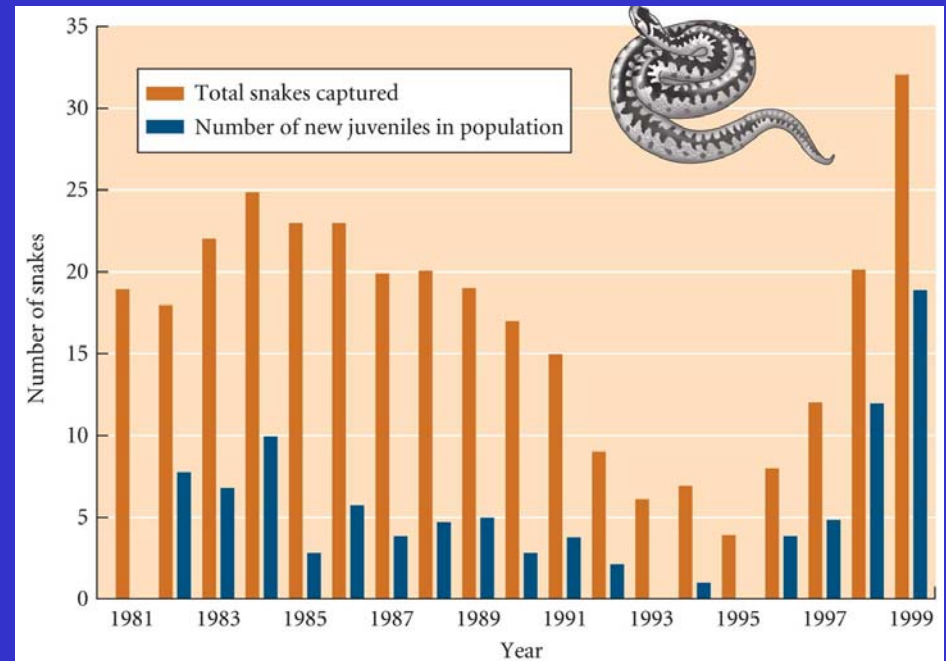


Other studies have confirmed the high degree of deleterious recessive genetic variation in wild populations of other organisms.

Inbreeding results in greater expression of deleterious recessive alleles than would be seen in randomly mating populations. The overall reduction of the average fitness of individuals in the population is called “**inbreeding depression.**”



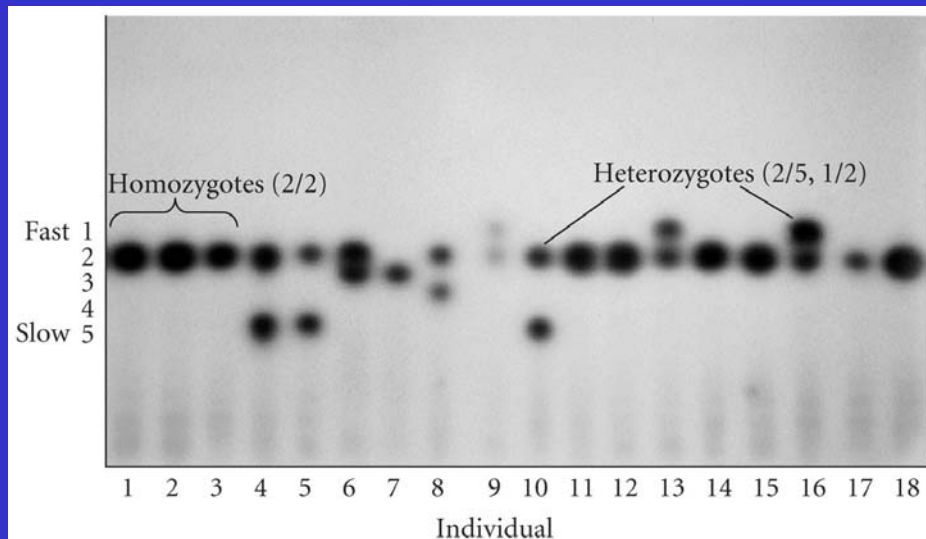
From a study of humans in Italy 1903-1907.



Viability was increased in a small population of adders in Sweden when individuals from outside were introduced in 1992.

Although genetically based phenotypic variation is common, it only accounts for a small fraction of the total genome. Protein variation - the product of genes - can be assessed using gel electrophoresis.

Lewontin & Hubby (1966) first assessed protein variation in *Drosophila* using gel electrophoresis. They assayed enzymatic variation. Different forms of an enzyme are called allozymes. Allozymes differ by one or more amino acids and thus migrate at different rates when placed in an electric field.



Their study and similar studies in other organisms have shown that on average individuals are heterozygous for 10% or more of their genes. There is a lot of genetic variation in populations.

Although random mating among any distribution of genotypes produces a binomial distribution of genotypes in just one generation, the variation at two different genes does not mix as quickly.

For two genes, A and B, each with two alleles (A_1, A_2, B_1, B_2) and with each allele in equal frequency [$f(A_1)=p, f(A_2)=q, f(B_1)=r, f(B_2)=s$] we should expect each to have a binomial distribution and be present in all possible combinations in proportion to their frequencies.

Substituting a for A_2 and b for B_2 :

For the A gene:

$$f(AA) = p^2, f(Aa) = 2pq, f(aa) = q^2$$

For the B gene:

$$f(BB) = r^2, f(Bb) = 2rs, f(bb) = s^2$$

For both genes together

$$f(AABB) = p^2 * r^2$$

$$f(AABb) = p^2 * 2rs$$

$$f(AAbb) = p^2 * s^2$$

$$f(AaBB) = 2pq * r^2$$

$$f(AaBb) = 2pq * 2rs$$

$$f(Aabb) = 2pq * s^2$$

$$f(aaBB) = q^2 * r^2$$

$$f(aaBb) = q^2 * 2rs$$

$$f(aabb) = q^2 * s^2$$

Equilibrium at 2 genes can take several generations to achieve.

When a population is not at equilibrium for multiple genes it is in **“linkage disequilibrium.”**

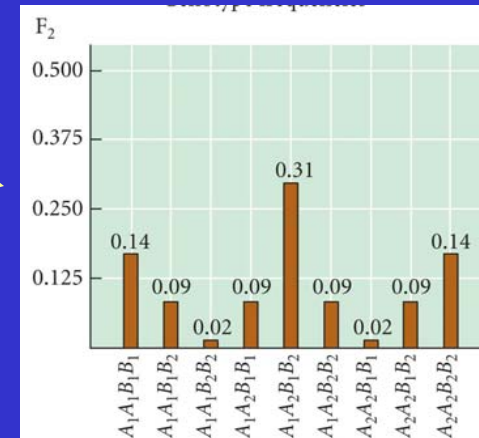
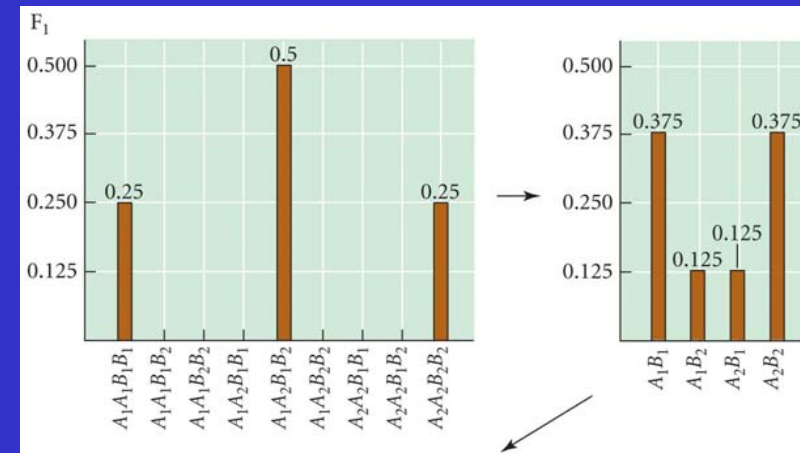
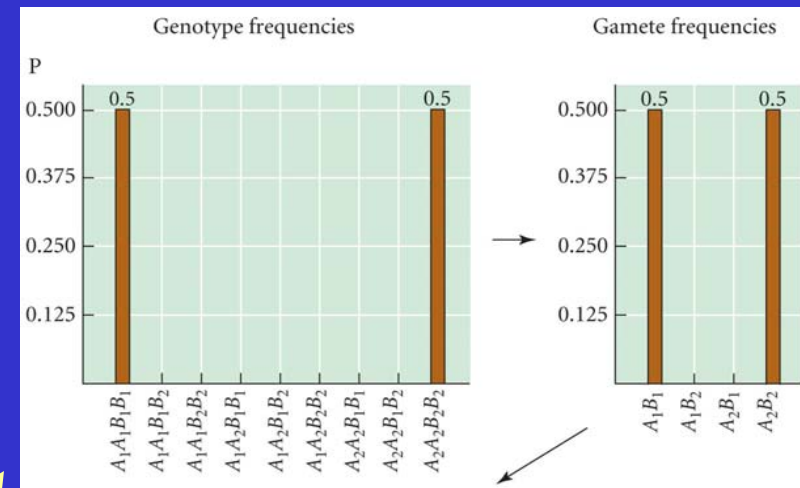
Disequilibrium can be measured as $d = f(AB) * f(ab) - f(Ab) * f(aB)$

$$d_0 = 0.5 * 0.5 - 0 * 0 = 0.25$$

$$d_1 = 0.375 * 0.375 - 0.125 * 0.125 = 0.125$$

$$d_2 = 0.3075 * 0.3075 - 0.1875 * 0.1875 = 0.0594$$

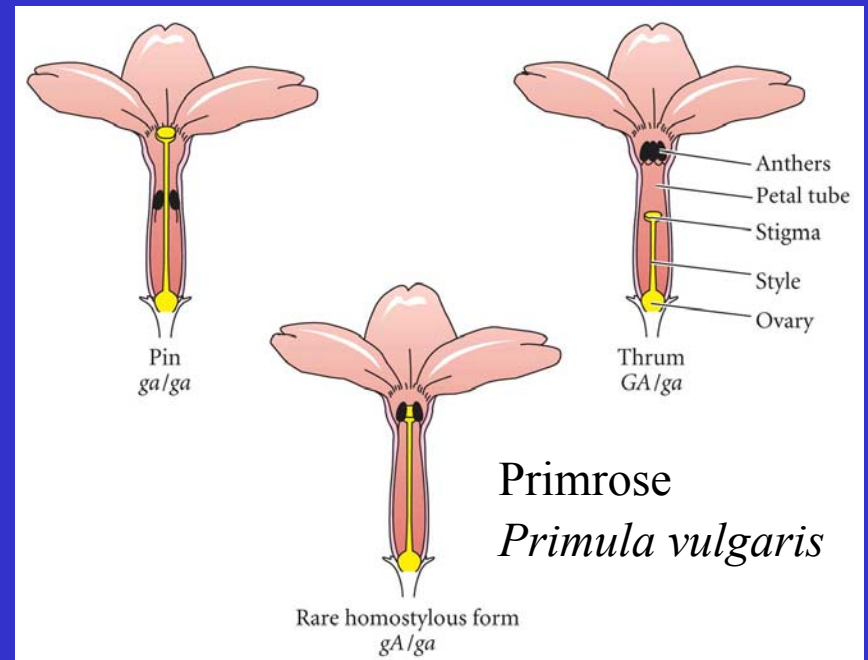
with continued random mating disequilibrium decreases to 0.



Even though it is called linkage disequilibrium it occurs with linked or nonlinked genes. The example above assumed the genes were on different chromosomes, assorting independently. If the genes are on the same chromosome, crossing-over is required to produce the recombinant gametes (Ab, and aB in this case). The more closely linked the genes are the less will be the frequency of crossing-over and it will take longer to achieve linkage equilibrium.

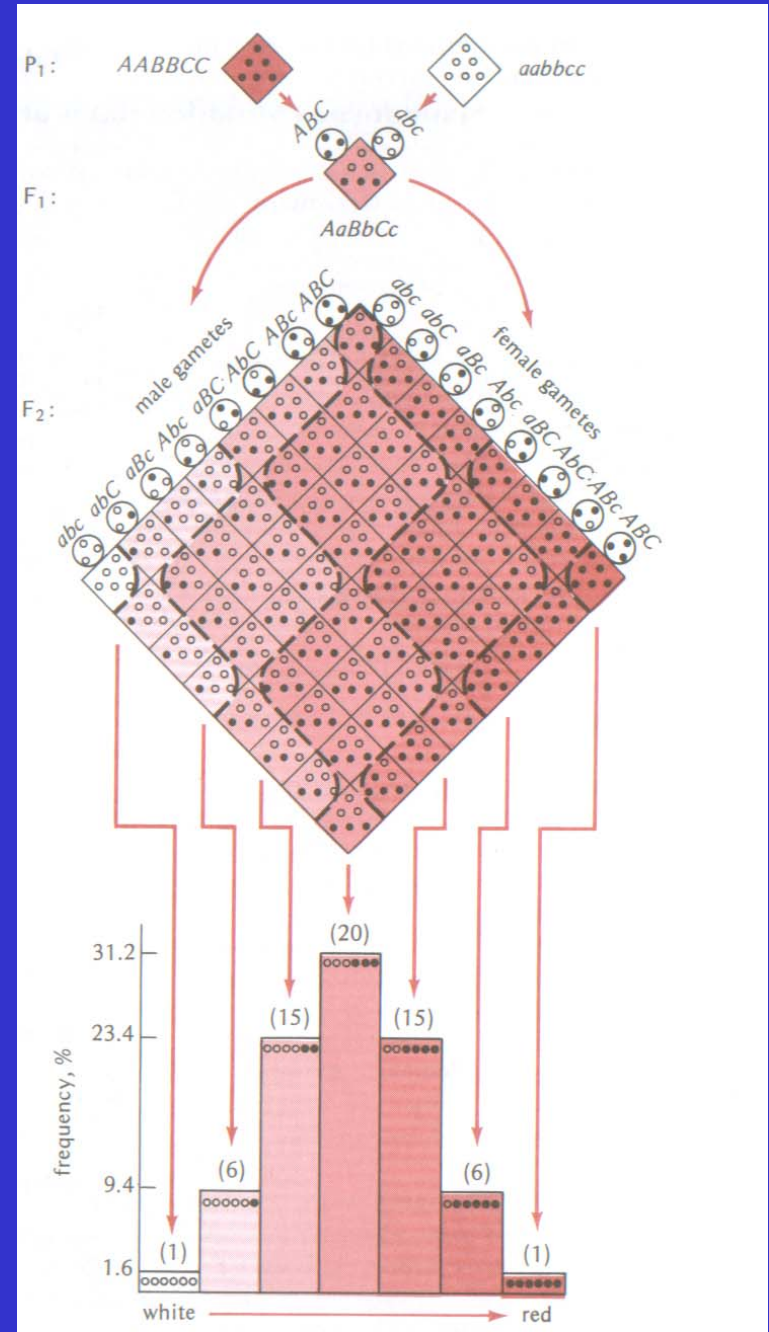
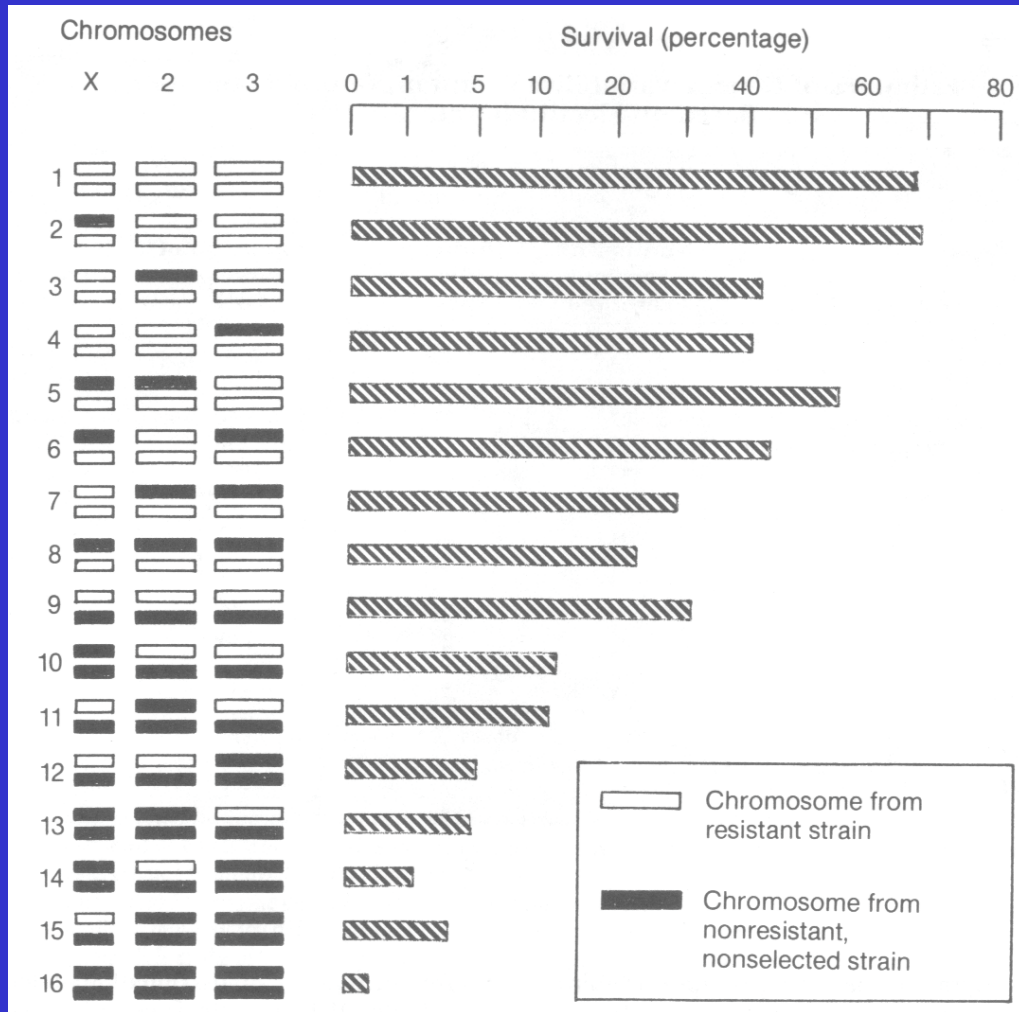
Most populations exhibit equilibrium for multiple genes, but there are some exceptions.

Inversion heterozygotes don't produce recombinant gametes - preserving the original gene combinations and linkage disequilibrium.

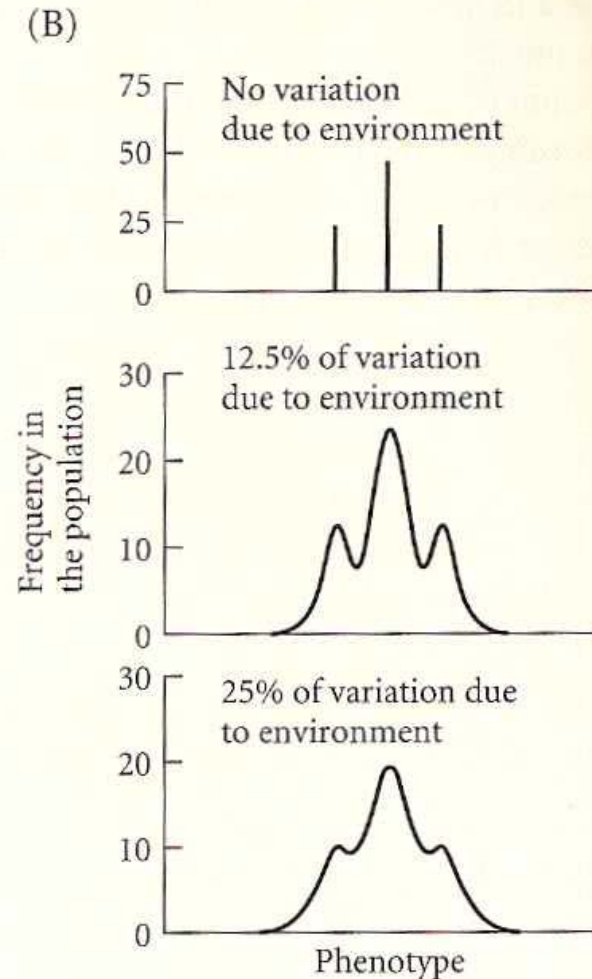
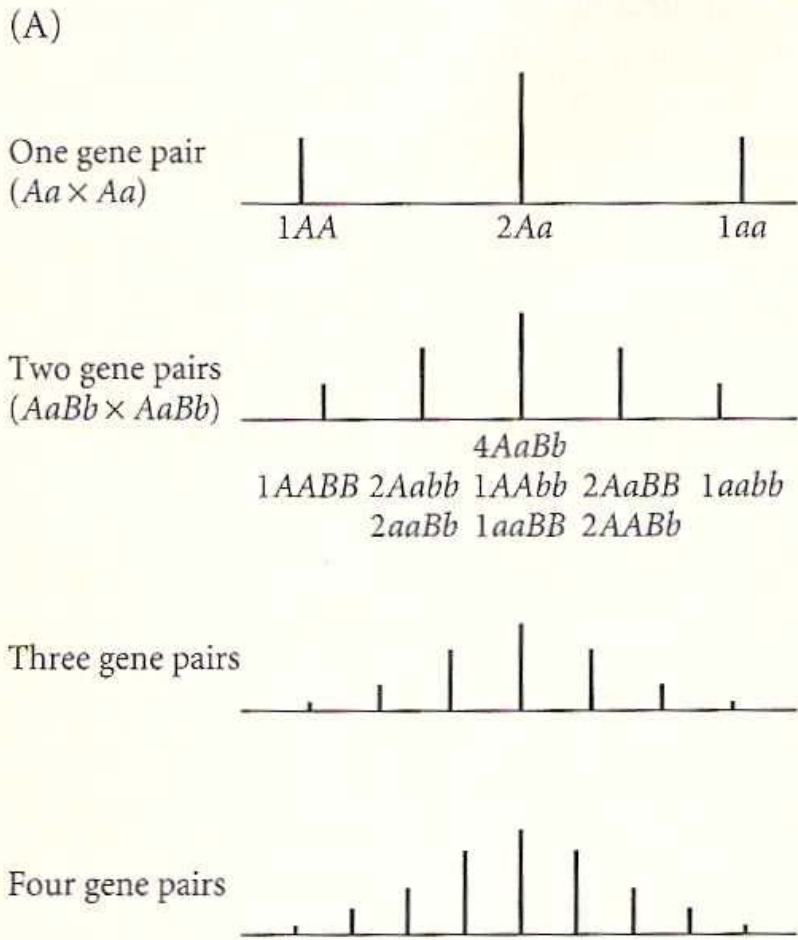


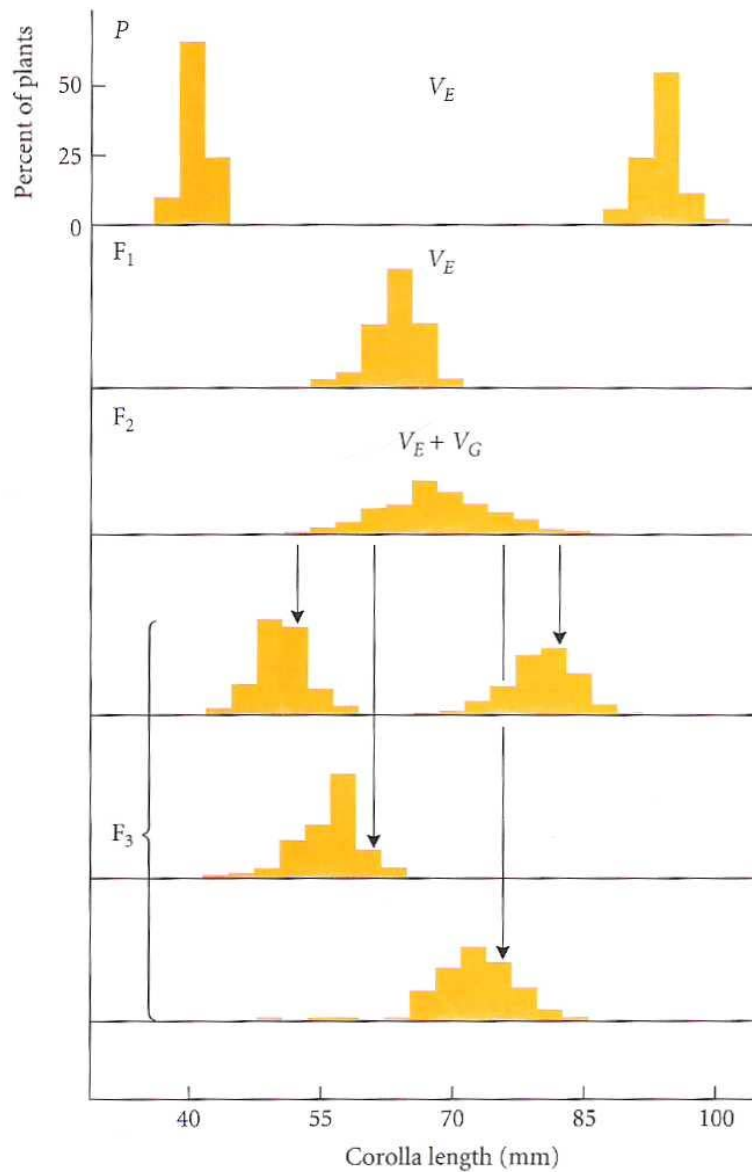
G and A are closely linked

Polygenic inheritance - for many traits, phenotypic variation is due to multiple genes that each have small effects on the phenotype - **quantitative variation**.



The environment can also influence the phenotype. Phenotypic variation that is due to different environmental conditions is called environmental variation (V_e)





Variation in phenotypes is due to the environment (V_e) and due to genetic differences (V_g). The contributions of each can be quantified.

Within genetically identical lines the only phenotypic variation is environmental variation.

Heritability (H^2)- the proportion of phenotypic variation (V_p) in the population that is due to underlying genetic variation (V_g)

$$V_p = V_e + V_g \quad H^2 = V_g/V_p$$

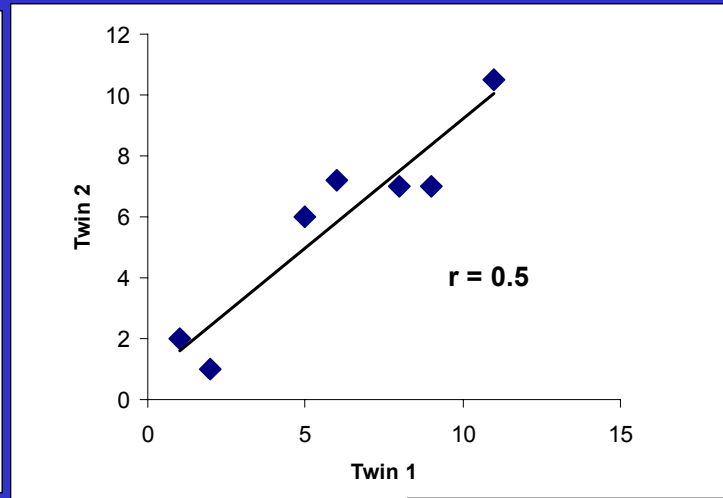
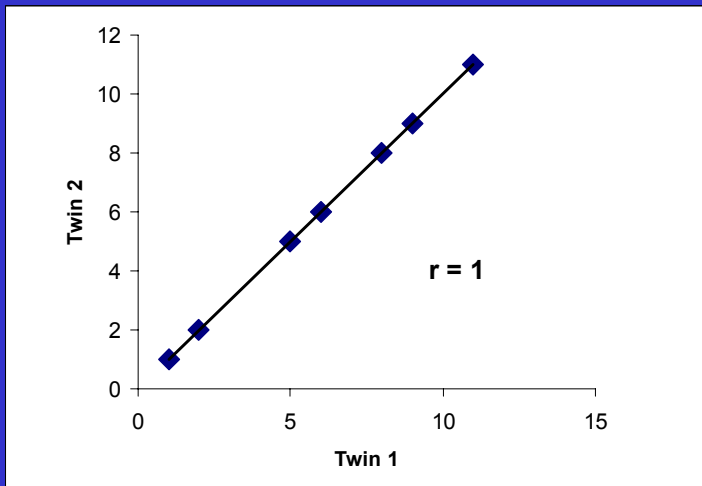
$H^2 = 1$ all phenotypic variation is due to genetic differences

$H^2 = 0$ all phenotypic variation is due to environmental differences

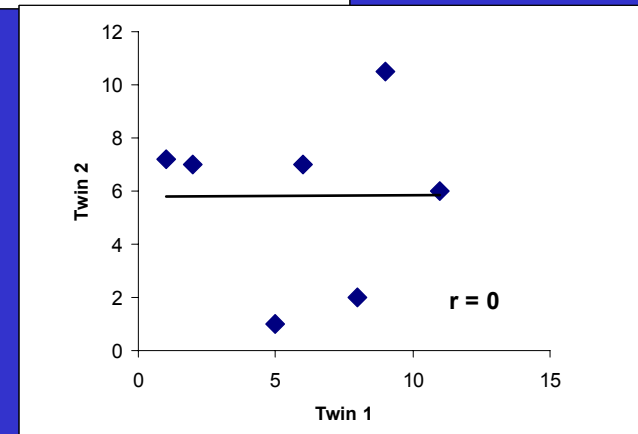
$H^2 = 0.6$ 60% of the phenotypic variation in the population is due to genetic variation, 40% is due to environmental variation

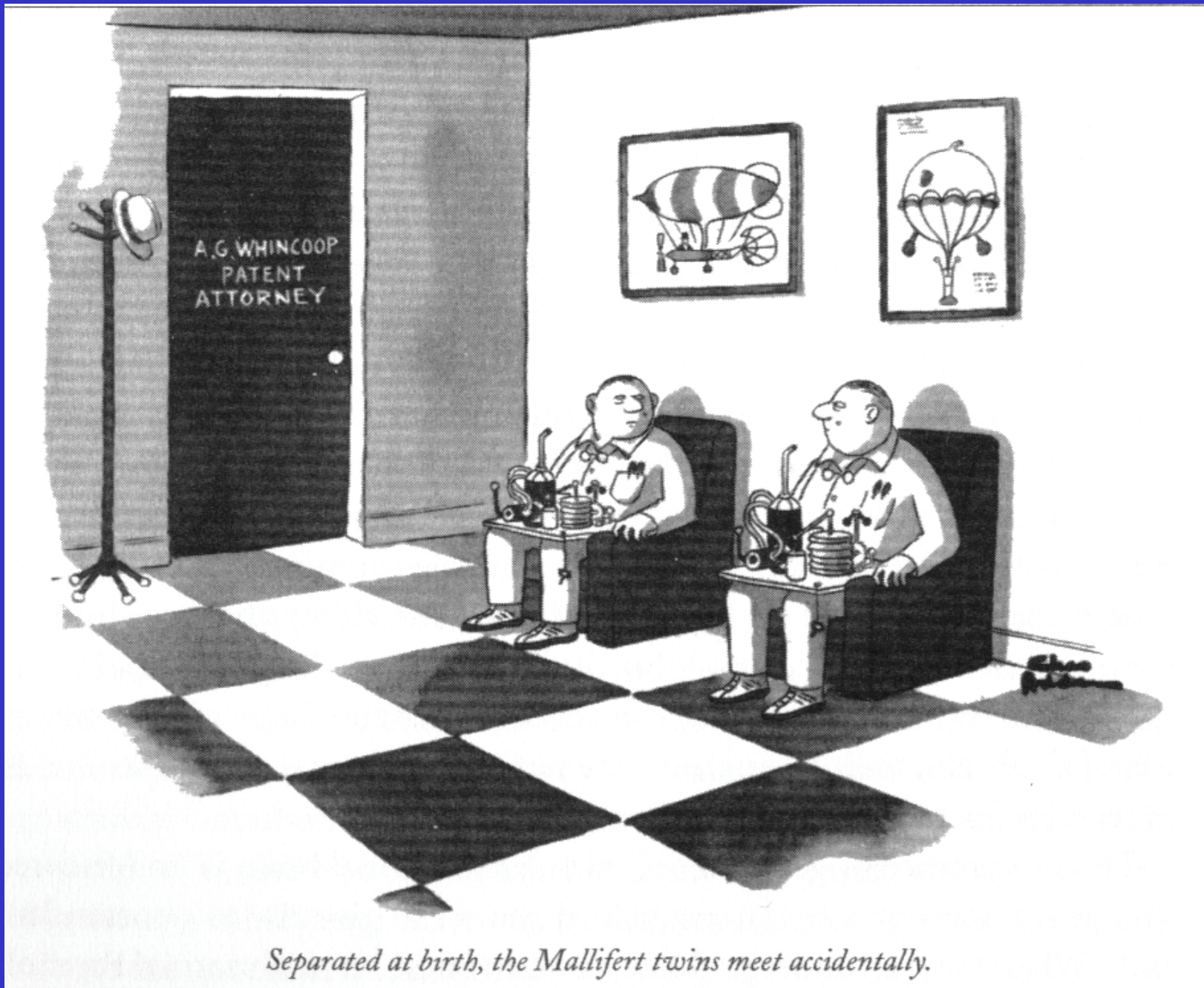
Measurement of Heritability

twin studies - a large set of identical twins reared apart can indicate the how much variation is due to environmental differences since there are no genetic differences between identical twins.



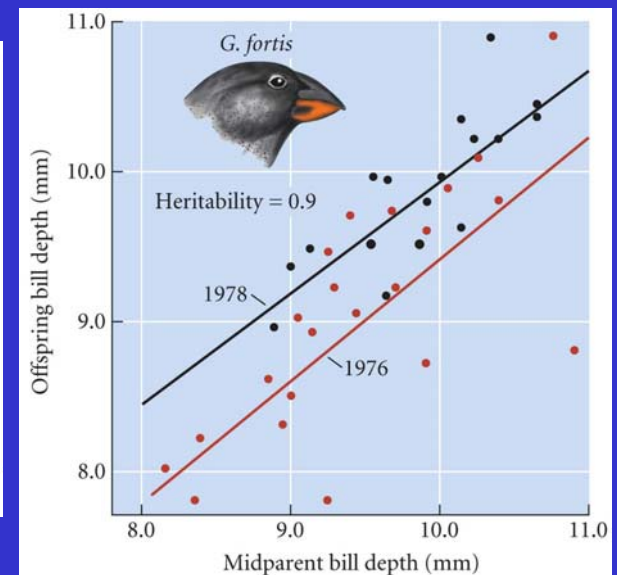
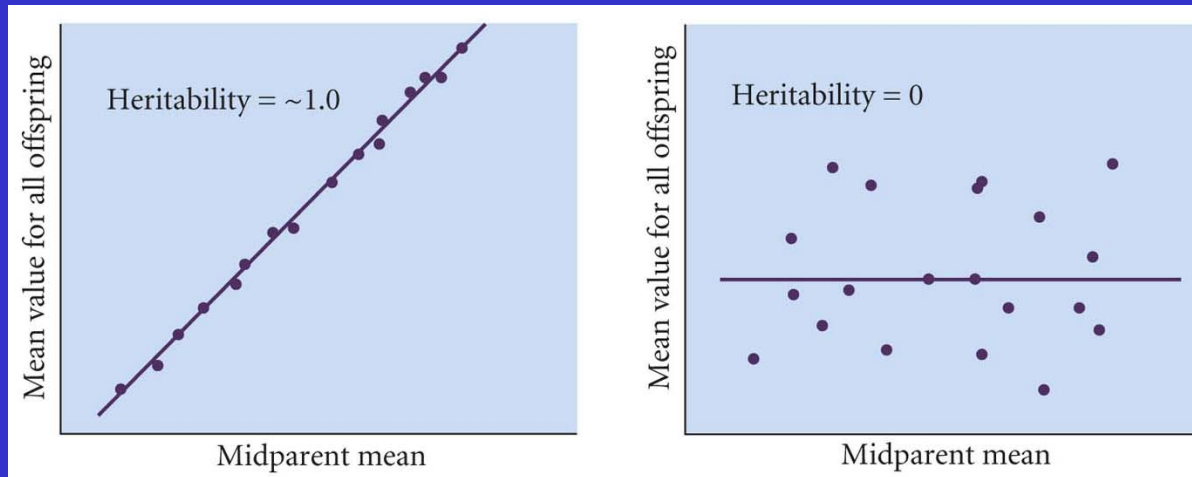
For identical twins reared apart, the correlation coefficient (r) is equal to the heritability of the phenotypic trait.





Separated at birth, the Mallifert twins meet accidentally.

Correlation between close relatives - the correlation statistic (r) between the phenotypes of close relatives should reflect their degree of genetic relatedness if the trait has heritability near 1. A correlation less than expected based on genetic relatedness indicates that some of the phenotypic difference between relatives is due to environmental differences. Most often parents and offspring are compared using the parental mean phenotype of the offspring mean phenotype.



Selection experiments - selection based on phenotypic differences should result in change in the average phenotype of the next generation if the phenotypic difference is heritable. The amount of change relative to the amount of selection reflects heritability.

Selection experiment:

Parental generation has an average phenotype of **P**

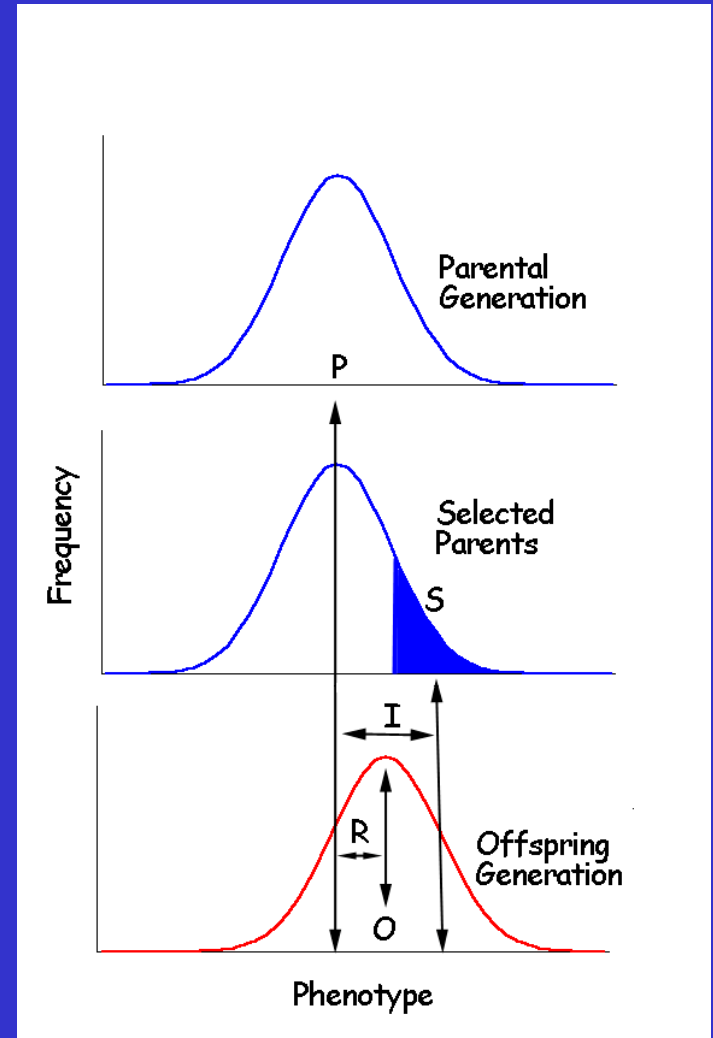
Some of those parents are selected to produce the next generation. They have an average phenotype of **S**

They produce offspring with an average phenotype of **O**.

The selection intensity: $I = S - P$

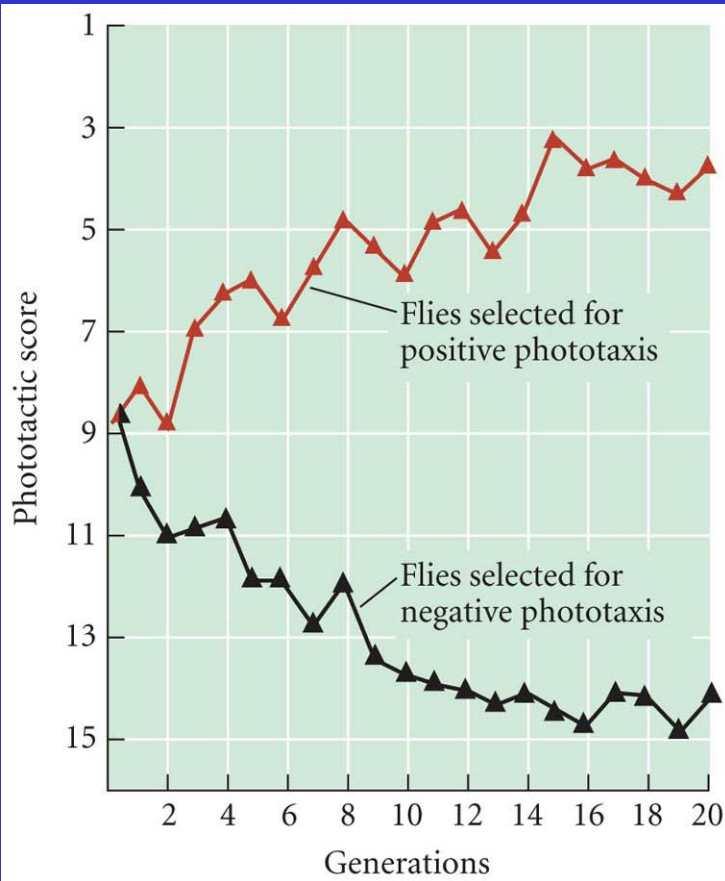
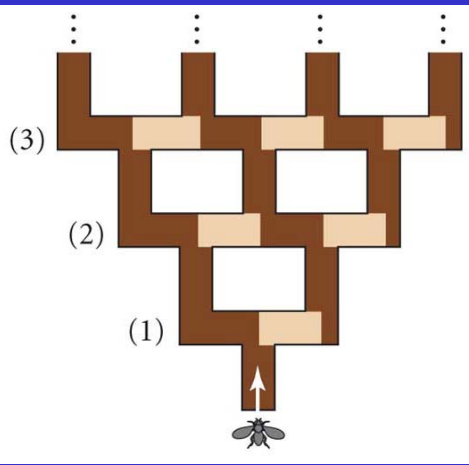
The response to selection: $R = O - P$

Heritability: $H^2 = R/I$



Flies with no overall preference for flying toward or away from the light can be selected for positive phototaxis or negative phototaxis.

$$H^2 = 0.09$$



After many generations of selection the response to selection decreases.

Pure-bred lines have little genetic variation for responding to selection.

Variation within pure-bred lines is mostly or entirely environmental - heritability is very low.

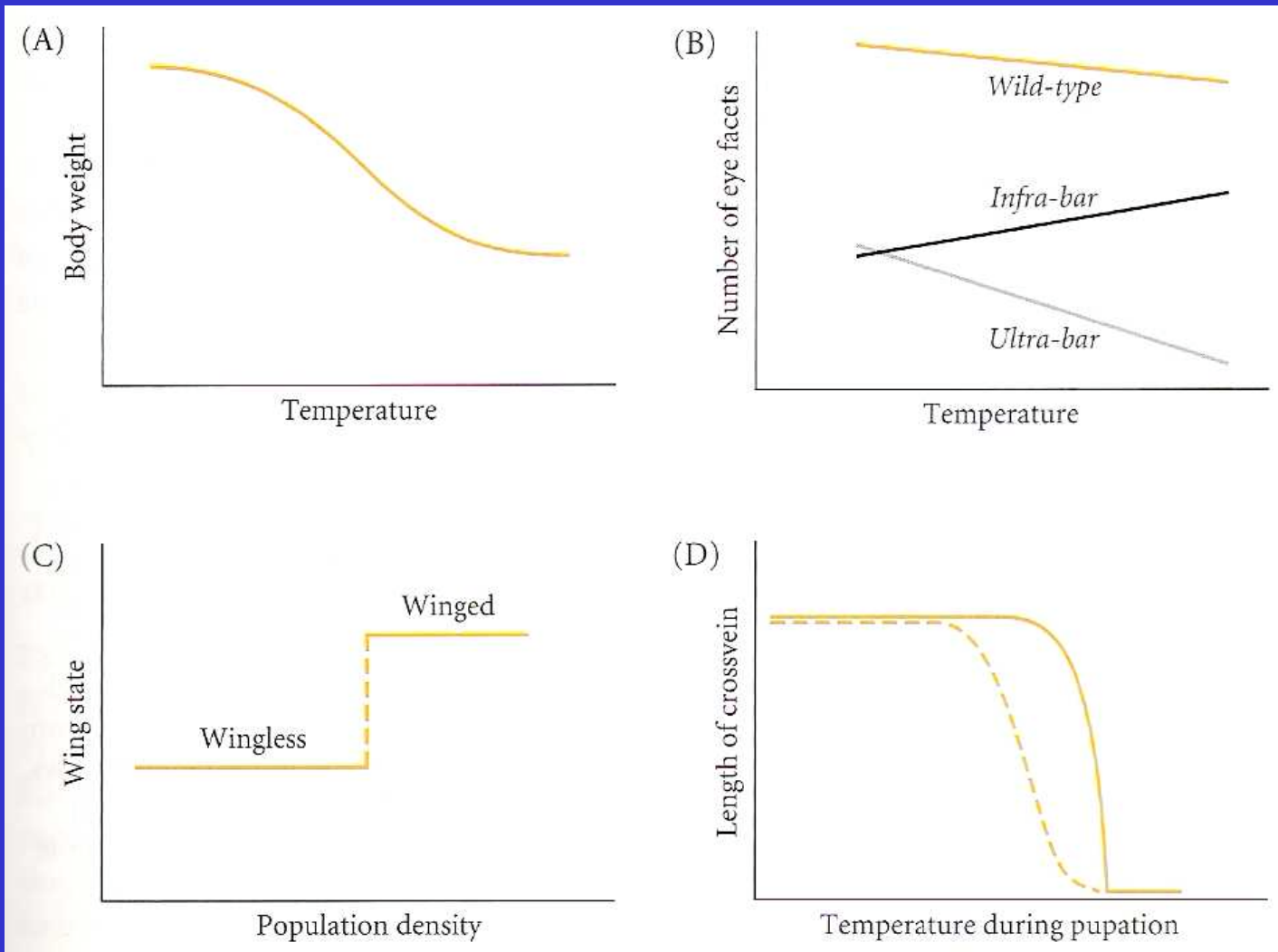
All estimates of heritability apply only to the environment in which they were measured.

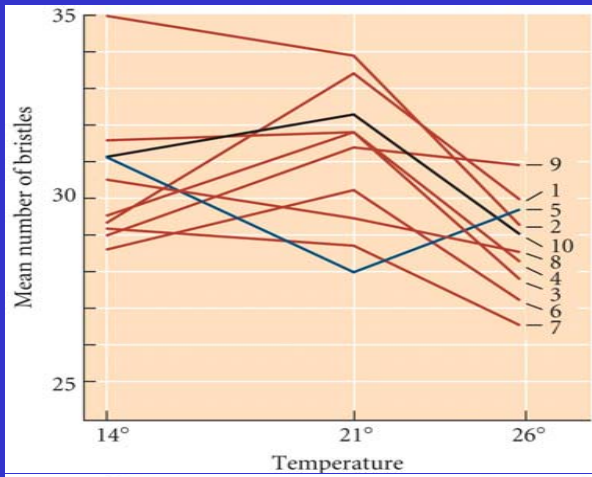
$$V_p = V_e + V_g \quad H^2 = V_g/V_p$$

If the study is done in an environment that varies little, then little phenotypic variation will result from environmental variation and the estimate of heritability will be high.

If the study is done in an environment that varies greatly, then there may be a large amount of phenotypic variation that is due to environmental variation and the estimate of heritability will be low.

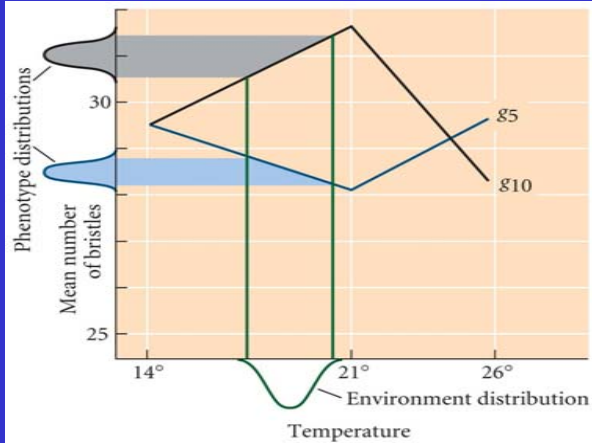
The expression of a single genotype may be different in different environments. The pattern of change in expression with change in environment is called the “**norm of reaction.**”



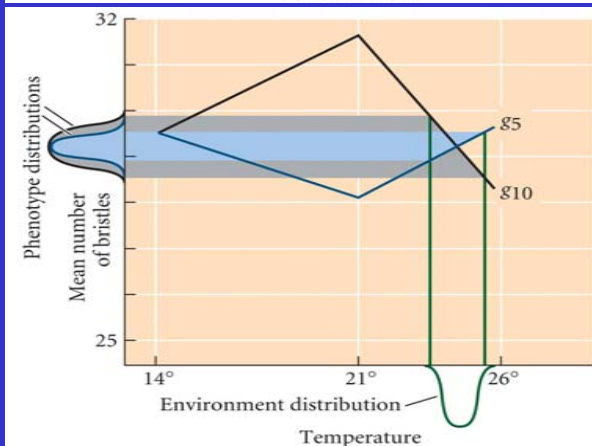


When the norm of reaction differs between different genotypes there is a **genotype x environment interaction**

The measurement of heritability often depends on the environment in which the measurement is made.



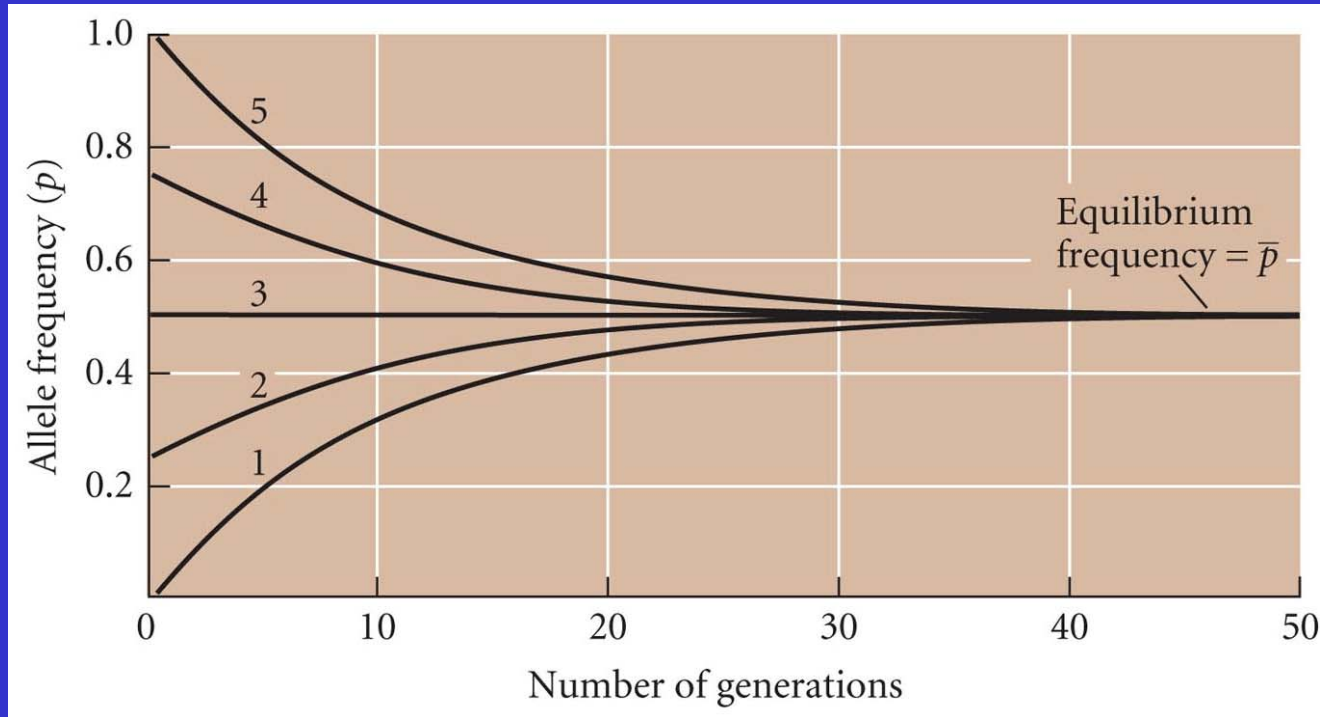
V_p large and V_e small $\rightarrow H^2$ high



V_p small and V_e larger $\rightarrow H^2$ low

Migration (gene flow) and genetic variation.

Gene flow has a homogenizing effect on populations



The amount of gene flow depends on species mobility and barriers to dispersal.

Read: Variation among Populations (p 212-216)