POPULATION GENETICS

Evolution Lectures 4

POPULATION GENETICS

The study of the rules governing the maintenance and transmission of genetic variation in natural populations.

Some Definitions

Population: A freely interbreeding group of individuals.

Gene Pool: The sum total of genetic information present in a population at any given point in time.

Phenotype: A morphological, physiological, biochemical, or behavioral characteristic of an individual organism.

Genotype: The genetic constitution of an individual organism.

Locus: A site on a chromosome, or the gene that occupies the site.

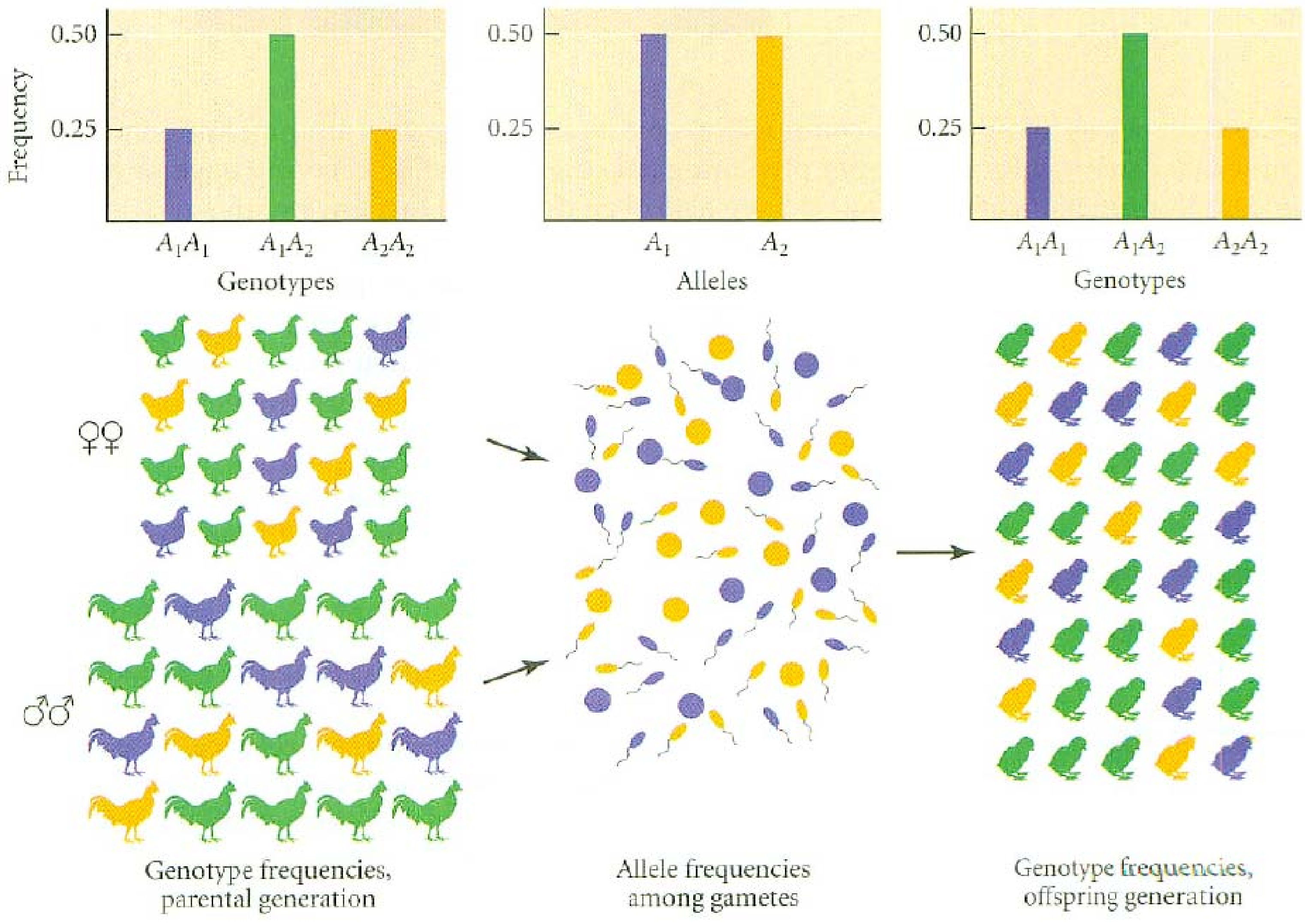
Gene: A nucleic acid sequence that encodes a product with a distinct function in the organism.

Allele: A particular form of a gene.

Gene (Allele) Frequency: The relative proportion of a particular allele at a single locus in a population (a number between 0 and 1).

Genotype Frequency: The relative proportion of a particular genotype in a population (a number between 0 and 1).

Genotype vs. Allele frequency



Calculation of allele frequency

- Lets assume:
  - In a population 400 people have the genotype TT, 400 have Tt and 200 have tt
  - So they have 2000 allele total
  - Of these 1200 are T and 800 are t
  - If frequency of T is p, then p=0.60
  - If frequency of t is q, then q=0.40

Calculation of allele frequency

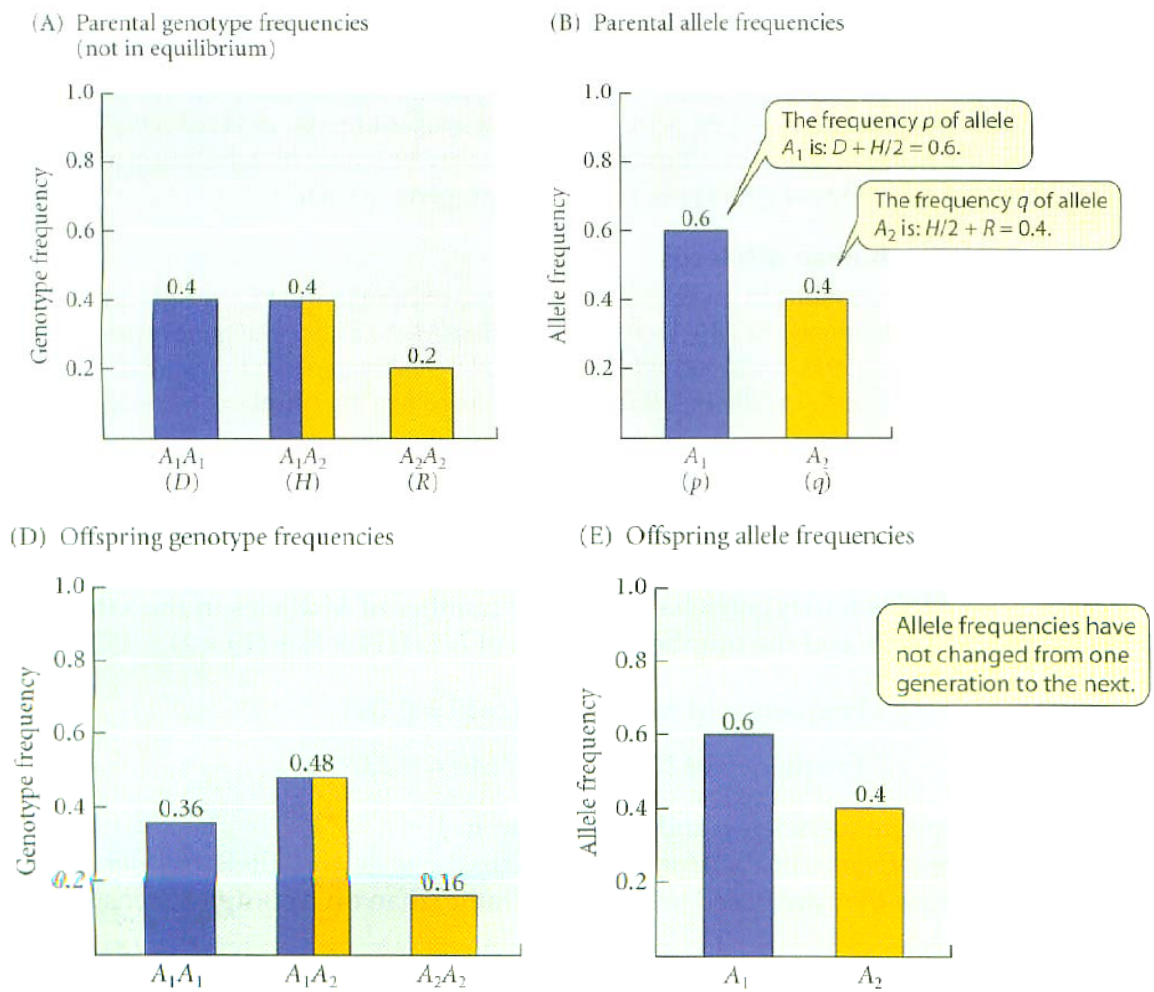
- By counting
  - T = 800 in TT + 400 in Tt = 1200/2000 = 0.60
  - t = 400 in Tt + 400 in tt = 800/2000 = 0.40
  - Or frequency of an allele=frequency of homozygote for that allele + ½ frequency of heterozygote
- Using genotype frequency
  - T= 0.40 TT + ½ (0.40 Tt) = 0.40 + 0.20 = 0.60
  - t= 0.20 tt + ½ (0.40 Tt) = 0.20 + 0.20 = 0.40

Conservation of allele frequency

♂ / ♀	TT=0.40	Tt=0.40	tt=0.20
TT=0.40	0.160	0.160	0.08
Tt=0.40	0.160	0.160	0.08
tt=0.20	0.08	0.08	0.04

In F1; TT=0.36, Tt=0.48; tt=0.16
But; T=0.60 and t=0.40

Conservation of allele frequency



Calculation of allele frequency

- In F1; TT=0.36, Tt=0.48; tt=0.16
- And T=0.60 and t=0.40
- Also if p=T and q=t
- Then p^2=0.36, q^2=0.16 and 2pq=0.48

The Hardy-Weinberg Equilibrium

A single generation of random mating establishes H-W equilibrium genotype frequencies, and neither these frequencies nor the gene frequencies will change in subsequent generations.

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

Hardy-Weinberg assumptions

- Mating is random (with respect to the locus).
- The population is infinitely large.
- Genes are not added from outside the population (no gene flow or migration).
- Genes do not change from one allelic state to another (no mutation).
- All individuals have equal probabilities of survival and reproduction (no selection).

Implications of the Hardy-Weinberg equilibrium

- A random mating population with no external forces acting on it will reach the equilibrium H-W frequencies in a single generation, and these frequencies remain constant there after.
- Any perturbation of the gene frequencies leads to a new equilibrium after random mating.
- The amount of **heterozygosity** is maximized when the gene frequencies are intermediate.
  - **2pq has a maximum value of 0.5 when**
    - **p = q = 0.5**

Genotype frequency distribution

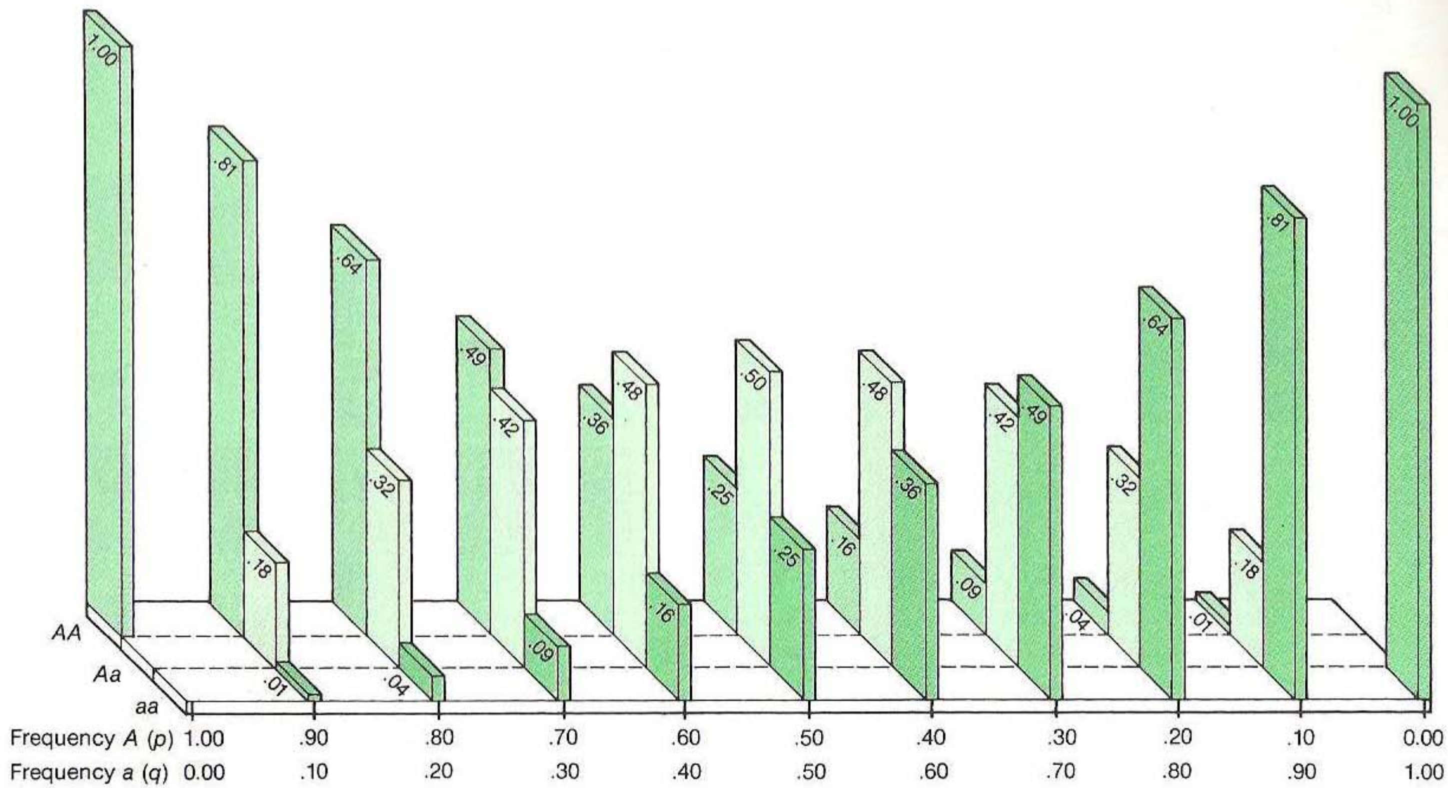
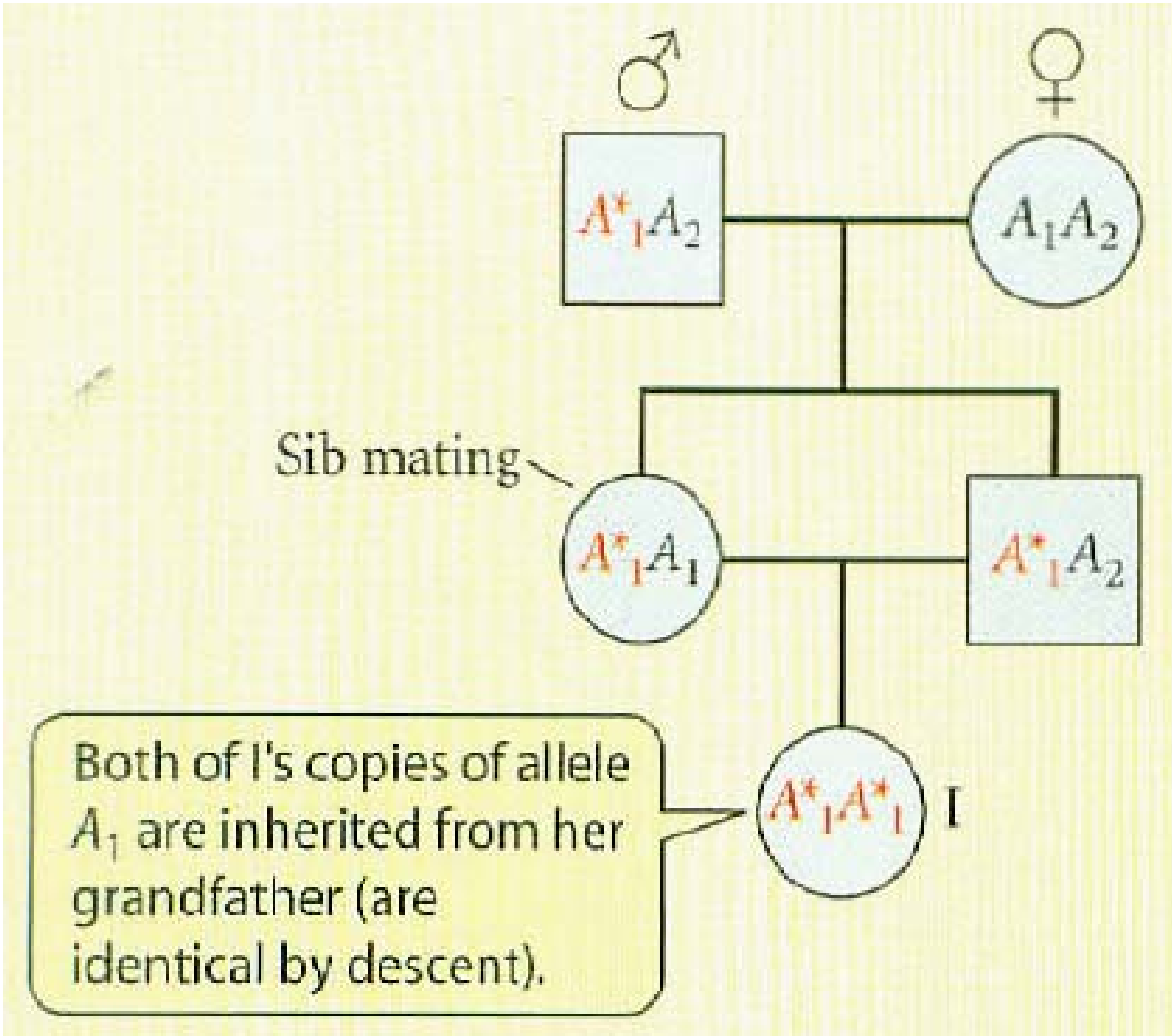


FIGURE 21-4 Genotypic frequencies at Hardy–Weinberg equilibrium for a variety of allele frequencies of *A* (*p*) and *a* (*q*). (Adapted from Wallace, B., 1970. *Genetic Load: Its Biological and Conceptual Aspects.* Prentice Hall, Englewood Cliffs, NJ.)

Factors changing equilibrium

Inbreeding

Example


♂
♀
$A^*_1A_2$
A_1A_2
Sib mating
$A^*_1A_1$
$A^*_1A_2$
Both of I's copies of allele A_1 are inherited from her grandfather (are identical by descent).
$A^*_1A^*_1$
I

Calculations

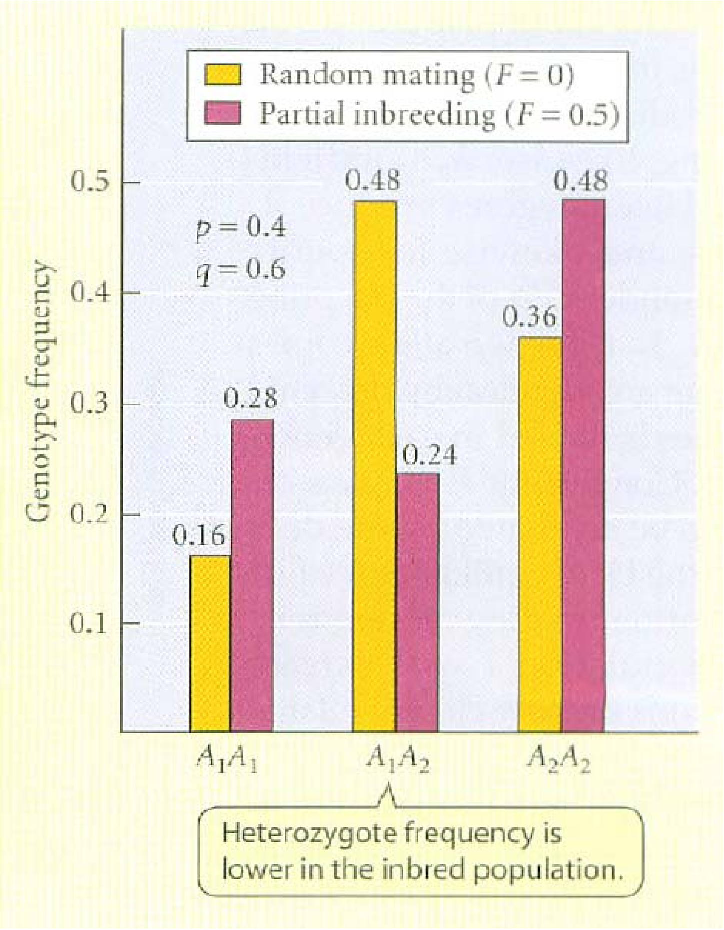
- Definition first
  - Autozygous: individuals homozygous with the identical allele by descent
  - Allozygous: either heterozygous or homozygous individuals with non-identical allele
  - Inbreeding coefficient (F) is the probability of an individual taken at random from a population will be autozygous
  - In a randomly mating population F=0 and in an all inbreed population F=1

Calculations

In a population with some inbreeding 1-F is the allozygous frequency if F is autozygous frequency

	Allozygous	Autozygous	Genotype frequency
A_1A_1	$p^2(1\text{-}F)$	$+pF$	$=p^2+Fpq$
A_1A_2	$2pq(1\text{-}F)$		$2pq(1\text{-}F)$
A_2A_2	$q^2(1\text{-}F)$	$+qF$	$=q^2+Fpq$

In a graph



Find: How to calculate inbreeding frequency in a population?

Factors changing equilibrium

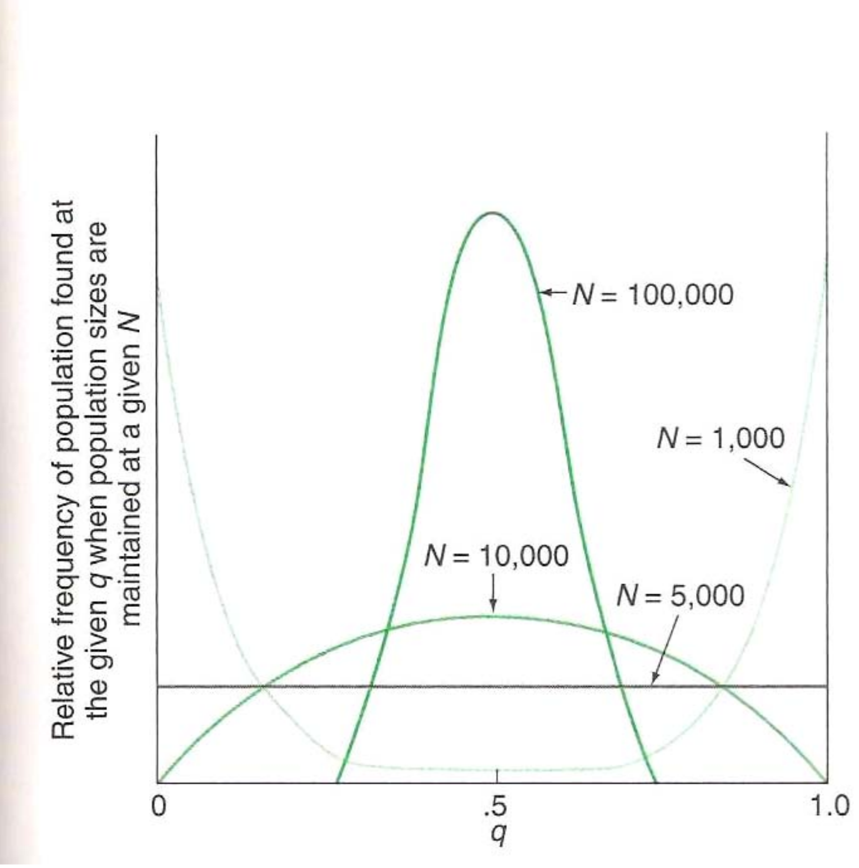
Genetic drift

GENETIC DRIFT

- Alteration of gene frequencies due to chance (stochastic) effects.
- Most important in *small* populations.
- Tends to reduce genetic variation as the result of extinction of alleles.
- Generally does not produce a fit between organism and environment; can, in fact, result in nonadaptive or maladaptive changes.

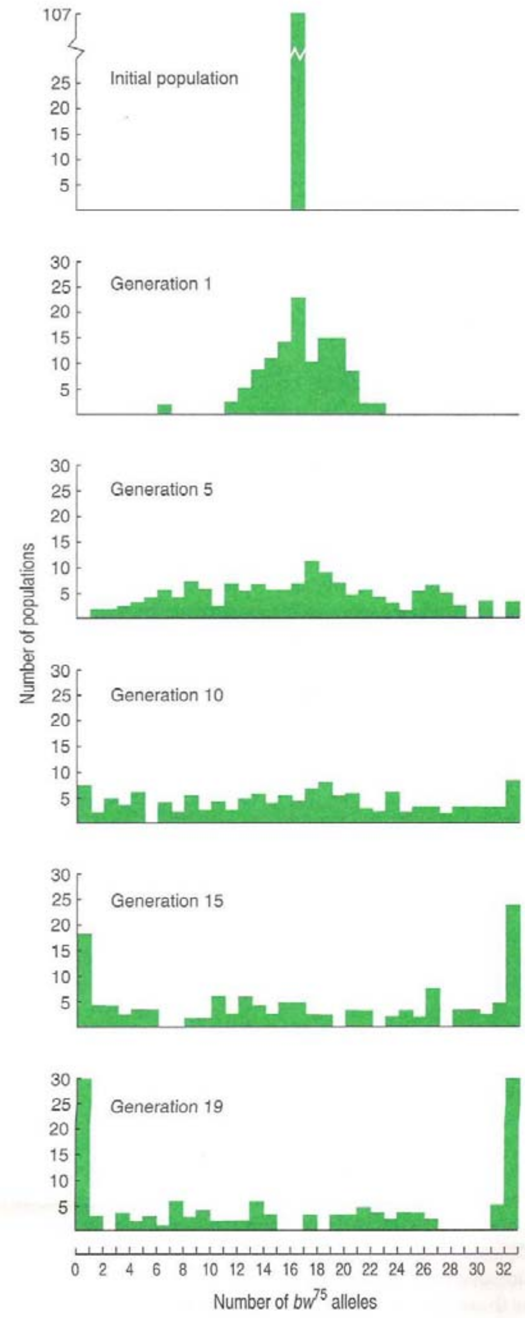
How does it work?

- Calculated by standard deviation $\sigma=\sqrt{pq/N}$ (p is the frequency of one allele, q is another; N is the number of genes sampled)
- For diploid population $\sigma=\sqrt{pq/2N}$
- If we start p=q=0.5 and N=100000
- Then σ=.001
- That is the frequency of either p or q will vary 0.5±0.001
- This is genetic drift and is greater in smaller population



Example

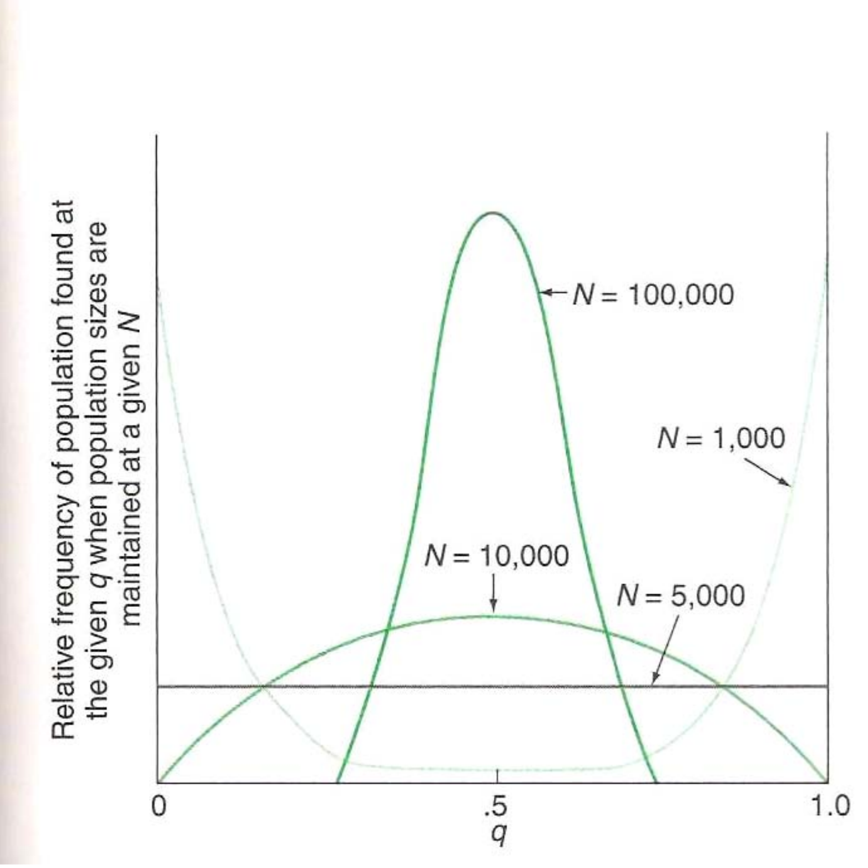
- Laboratory expt by Buri (1956)
- Set up 107 separate *D. melanogaster* lines, each with two brown alleles *bw* and bw^{75}
- Started each generation with 16 parents (8 males and 8 females, random selection)
- Therefore each generation started with 16X2=32 gene copies
- Continued for 19 generations
- See the changes in the allele frequency in the first generation
- At 19th 30 populations lost all bw^{75} allele and 28 have been fixed.
- Remember: No selection pressure



How does it work?

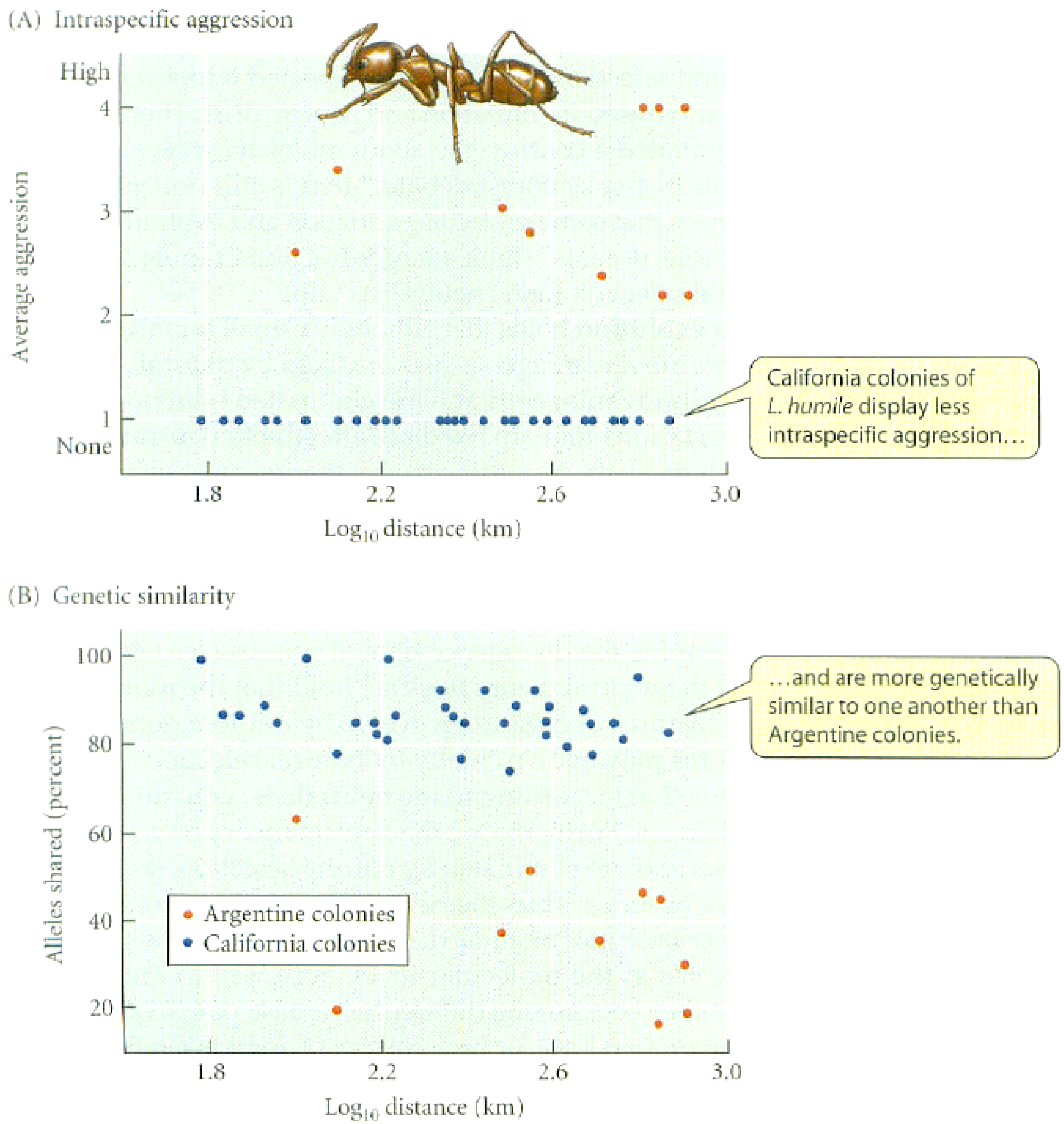
- If we start p=q=0.5 and N=214
- Then σ=.034
- That is the frequency of either p or q will vary 0.5±0.03
- This is just one generation
- In the next
  - at one end 0.47±0.03
  - and at the other 0.53±0.04
- Do the calculations for few more generations

- This is what is called Founder's Effect



Example

- Argentine ant (*Linepithema humile*)
- Introduced in California accidentally
- Highly aggressive in Argentina, between colonies
- Each colony has different odor
- But they form super colony in California
- Small group brought had same colony odor



Coalescence