Chapter 18
Population Genetics

Genetic variation, detection, concepts, sources, and forces

What is population genetics?

- Analyzes the amount and distribution of genetic variation in populations and the forces that control this variation

- mathematically based principles for changes in genotypes through time—individuals, populations, etc.

- examine mutation, migration, breeding system, among-population interactions, stochastic forces and selection on allele frequencies

- developed to bridge gap between "genes" and "species evolution"- microevolution

- "alleles" may be any kind of heritable mutation

Detecting Genetic Variation

- Single nucleotide polymorphisms (SNPs)
- Microsatellites (repeating sequences)
- Haplotypes (and the Human Hap Map)
- Other detection methods possible

The Gene Pool Concept and the Hardy-Weinberg Law

- Gene pool- the sum total of all alleles in the population

- Genotype frequencies and allele frequencies

- The Hardy-Weinberg Law provides an equation to relate the genotype frequencies and allele frequencies in a randomly mating population: $p^2 + 2pq + q^2 = 1$ for 2 alleles such as A and a

- If no forces act on a population in Hardy-Weinberg equilibrium, proportion of genotypes will stay the same

Hardy-Weinberg equilibrium
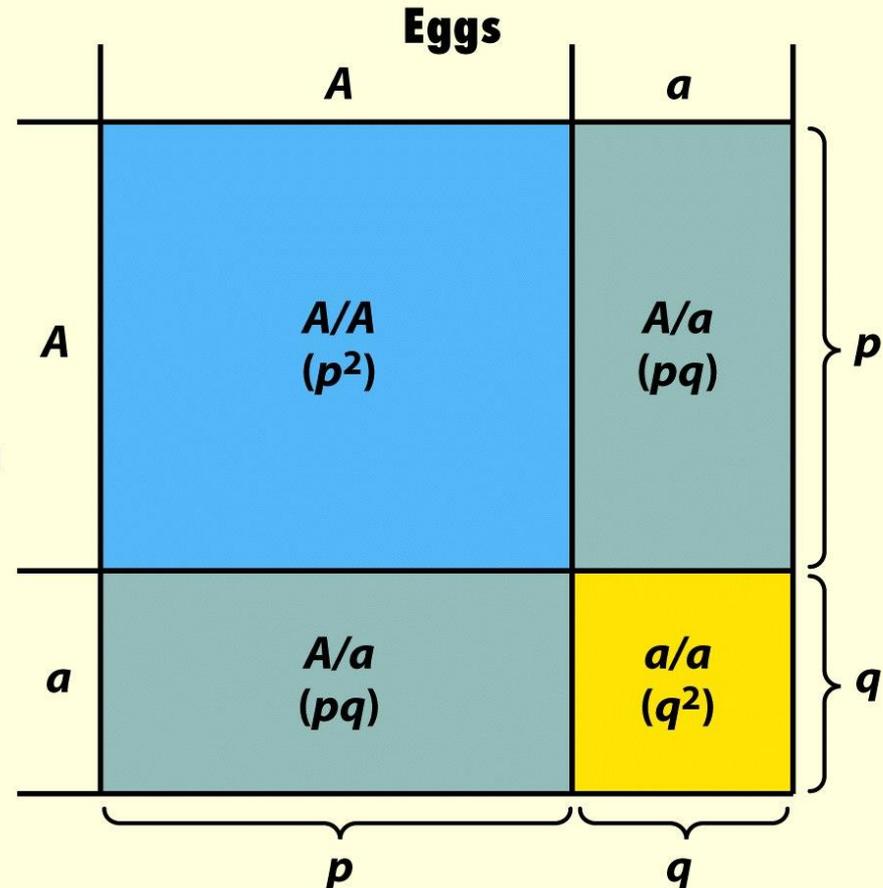
$$p2 + 2pq + q2 = 1$$

where p, q are
probabilities of
allele frequencies

The Hardy-Weinberg Equilibrium

Eggs

	A	a
A	A/A (p²)	A/a (pq)
a	A/a (pq)	a/a (q²)

Sperm

Hardy-Weinberg Sample Problem p.679

About 70% of all Caucasians can taste the chemical phenylthiocarbamide, and the remainder cannot. The ability to taste this chemical is determined by the dominant allele T, and the inability to taste is determined by the recessive allele t. If the population is assumed to be in H-W equilibrium, what are the genotype and allele frequenceis in this population?

Population Genetics

- Species-a group of organisms of the same kind that have the ability to interbreed to produce fertile offspring

- Population-a group of organisms of the same species located in the same place and time

- A population can be defined by its gene pool (i.e., every allele of every gene of every organism in the population)

- Alleles occur at certain frequencies (e.g., for a gene with 2 alleles, p and q, p + q = 1.0)

- Variation exists in populations (e.g., height, weight, eye color, etc., and this variation is governed by particular alleles-genetic variation)

The Hardy-Weinberg principle relates genotypes and allelic frequencies

- For 2 alleles, p and q, $p + q = 1$ and
- following mating, $p^2 + 2pq + q^2 = 1.0$
- Populations in which allele frequencies do not change are in genetic equilibrium (zero evolution)
- Assumes 5 conditions
1. Population is large
2. Mating is random
3. No immigration or emigration
4. Natural selection is not occurring
5. No mutations

Microevolution-changes in allele frequency over time; caused by:

- Populations can be small and experience genetic drift (chance events)/founder effect/population bottleneck
- Non-random mating
- Gene flow (individuals do leave or enter populations)
- Natural selection-nature selects individuals in a population that have "favorable" alleles which allow for survival in a given environment (e.g., Peppered moth)
- Mutations do occur

Figure 8.9 The Hardy-Weinberg proportions of genotypes in a population can be illustrated using a square. The lengths across the top of the square are divided into proportions p and q, representing the frequencies of alleles M and m in populations of peppered moth. The areas represent the proportions of the different genotypes in the population. In (a), the proportions represent the situation before industrial melanism. In (b), the proportions represent the situation after natural selection has increased the frequency of the M allele. Note that the areas of both the p^2 and p x q boxes are larger in (b), and the area of the q^2 box is smaller.
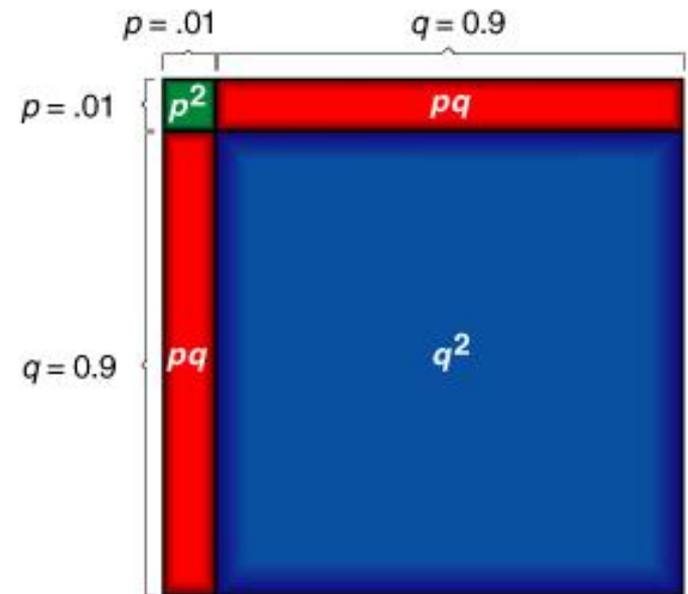
(a) Before industrial melanism
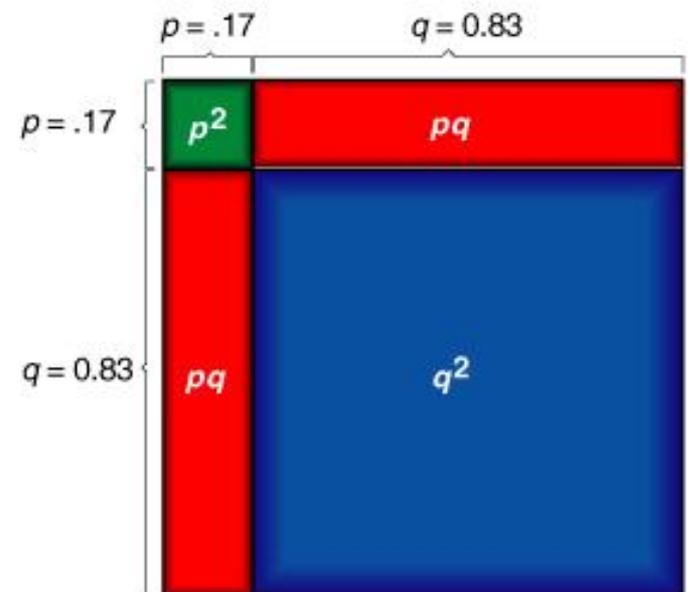
(b) After industrial melanism

Hardy-Weinberg principles

- populations, lacking external forces acting on them, will achieve H-W equilibrium if:
  - truly random mating
  - infinitely large population size
- many wild species probably do NOT meet either criterion, despite theoreticians' claims

Mating is often not random; three common mating systems

- Assortive mating (positive and negative)

- Isolation by distance (leds to subpopulations and perhaps new species)

- Inbreeding (increases homozygousity and can result in a reduction in vigor and reproductive success, i.e., inbreeding depression)

Sources of Genetic Variation in Populations

- Mutations

- Migration (gene flow)

- Recombination (linkage disequilibrium)

- Genetic drift (chance)-founder effect & bottleneck

Variation from recombination

- linkage equilibrium—repeated recombination between genes, randomizing combinations of alleles of different genes

  Example: if frequency of allele a is 0.2 and frequency of b is 0.4, under linkage equilibrium:

  frequency of $ab = 0.2*0.4 = 0.08$

Variation from recombination

- linkage disequilibrium - original <u>nonrandom</u> association between alleles of different genes on same chromosome

- linkage disequilibrium changes slowly through time, at rate proportional to amount of recombination between genes

- genetic variation through recombination can be much faster than through mutation

Two Forces Control the Fate of Genetic Variation in Populations

- Genetic drift (chance)
- Natural Selection

Genetic drift

- stochastic process—occurs randomly through time

- by chance, alleles may be lost from population→"fixation" of genotypes in population

- theoretically significant only in small populations (with or without random mating)

Selection

- genotypes from mutation & recombination express differential survival and reproduction = selection

- human actions on economically important plant or animal populations = artificial selection

- environmental conditions (broadly defined) acting on wild plant or animal populations or species = natural selection

Selection

- "most fit" = genotype or phenotype with greater average reproductive output over its lifespan than other genotypes or phenotypes
- dependent on place, time and environmental conditions, as well as genetics
- organisms are "selectional mosaics"
- survival and reproductive output—sum of all genetic processes and genetic-environmental interactions in each individual

Measuring fitness differences

- fitness = relative reproductive output
- fecundity = reproductive output
- viability = probability of survival from egg to adult
- Fitness = viability + fecundity
- can evaluate fitness etc. by comparing mutants or phenotype variants with wild type in laboratory or common garden
- some single-gene or other mutations result in obvious fitness differences, others do not

Measuring fitness (viability) at different temperatures

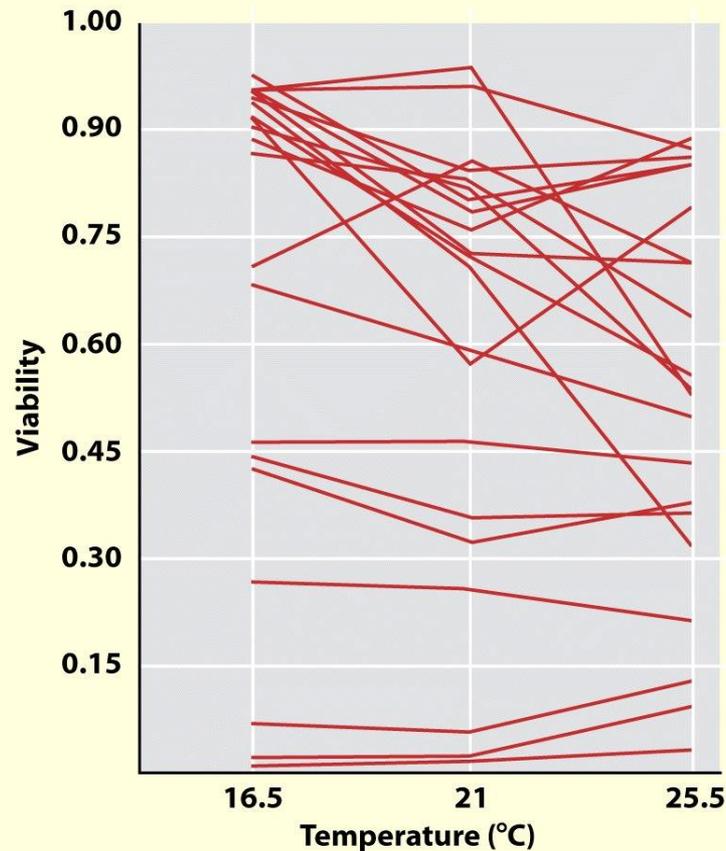**Measuring fitness at different temperatures**

Figure 17-8 *Introduction to Genetic Analysis*, Ninth Edition, W. H. Freeman and Company