1) The Smallest Unit of Evolution

- One misconception is that organisms evolve, in the Darwinian sense, during their lifetimes
- Natural selection acts on individuals, but only populations evolve
- Genetic variations in populations contribute to evolution
- **Microevolution** is a change in allele frequencies in a population over generations

2) Population genetics provides a foundation for studying evolution

- Microevolution is change in the genetic makeup of a population from generation to generation

3) Mutation and sexual reproduction produce the **genetic variation** that makes evolution possible

- **Mutation** and **sexual reproduction**, produce the variation in gene pools that contributes to differences among individuals
- Variation in individual **genotype** leads to variation in individual **phenotype**
- Not all phenotypic variation is heritable
- Natural selection can only act on variation with a genetic component

4) Variation Within a Population

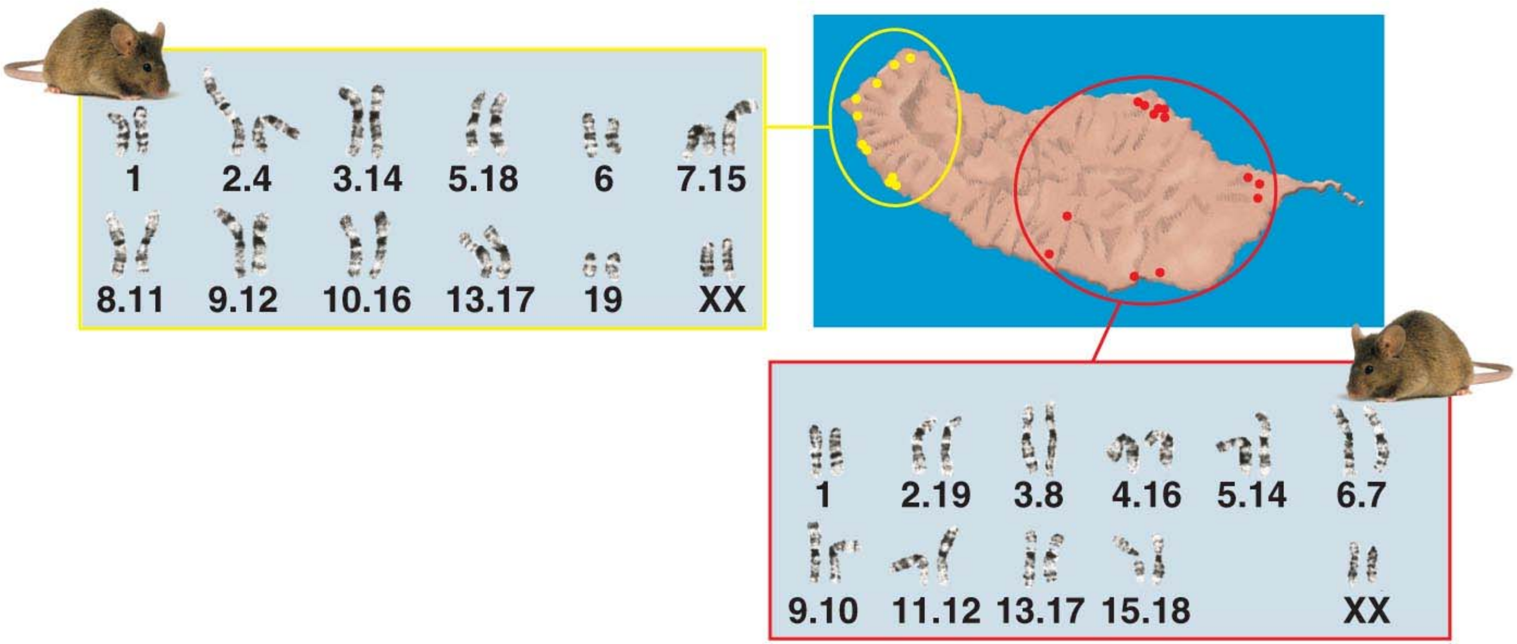
- Both discrete and quantitative characters contribute to variation within a population
- *Discrete characters* can be classified on an either-or basis (like either purple or white flowers, for example)
- *Quantitative characters* vary along a continuum within a population (height among humans, for example)

5) Measuring Genetic Variation

- Population geneticists measure polymorphisms in a population by determining the amount of heterozygosity at the gene and molecular levels
- **Average heterozygosity** measures the average percent of loci that are heterozygous in a population
- Nucleotide variability is measured by comparing the DNA sequences of pairs of individuals

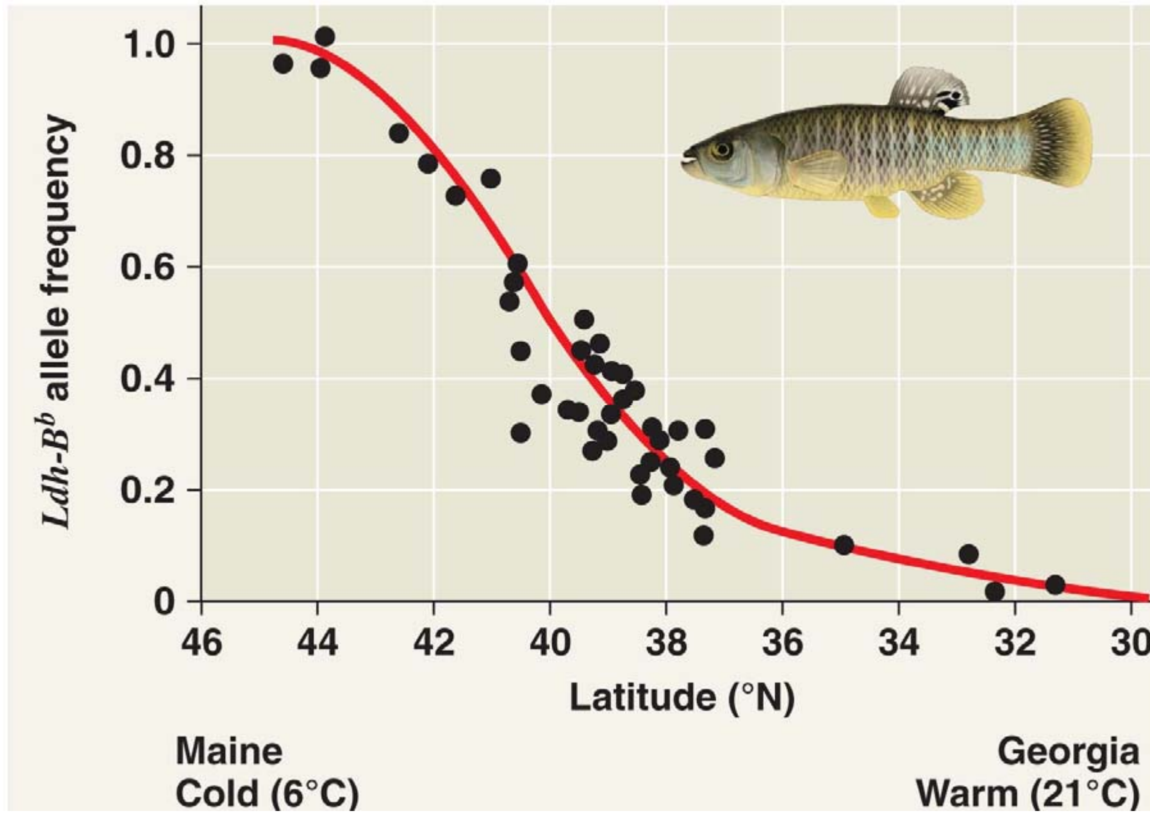
6) *Variation Between Populations*

- Most species exhibit **geographic variation**, differences between gene pools of separate populations or population subgroups



7) Clinal variation of traits

- Some examples of geographic variation occur as a **cline**, which is a graded change in a trait along a geographic axis



8) Mutation

- **Mutations** are changes in the nucleotide sequence of DNA
- Mutations cause new genes and alleles to arise
- Only mutations in cells that produce gametes can be passed to offspring

9) Point Mutations

- A point mutation is a change in one base in a gene
- The effects of point mutations can vary:
  - Mutations in noncoding regions of DNA are often harmless
  - Mutations in a gene might not affect protein production because of redundancy in the genetic code
  - Mutations that result in a change in protein production are often harmful
  - Mutations that result in a change in protein production can sometimes increase the fit between organism and environment

10) Mutations That Alter Gene Number or Sequence

- Chromosomal mutations that delete, disrupt, or rearrange many loci are typically harmful
- Duplication of large chromosome segments is usually harmful
- Duplication of small pieces of DNA is sometimes less harmful and increases the genome size
- Duplicated genes can take on new functions by further mutation

11) Mutation Rates

- Mutation rates are low in animals and plants
- The average is about one mutation in every 100,000 genes per generation
- Mutations rates are often lower in prokaryotes and higher in viruses

12) Sexual Reproduction

- Sexual reproduction can shuffle existing alleles into new combinations
- In organisms that reproduce sexually, recombination of alleles is more important than mutation in producing the genetic differences that make adaptation possible

13) Gene Pools and Allele Frequencies—some definitions

- A **population** is a localized group of individuals capable of interbreeding and producing fertile offspring
- A **gene pool** consists of all the alleles for all loci in a population
- A locus is *fixed* if all individuals in a population are homozygous for the same allele

14) Calculating allele frequency in a population:

- For diploid organisms, the total number of alleles at a locus is the total number of individuals x 2
- The total number of dominant alleles at a locus is 2 alleles for each homozygous dominant individual plus 1 allele for each heterozygous individual; the same logic applies for recessive alleles

15) Calculating allele frequency, cont.

- By convention, if there are 2 alleles at a locus, p and q are used to represent their frequencies
- The frequency of all alleles in a population will add up to 1, so, $p + q = 1$

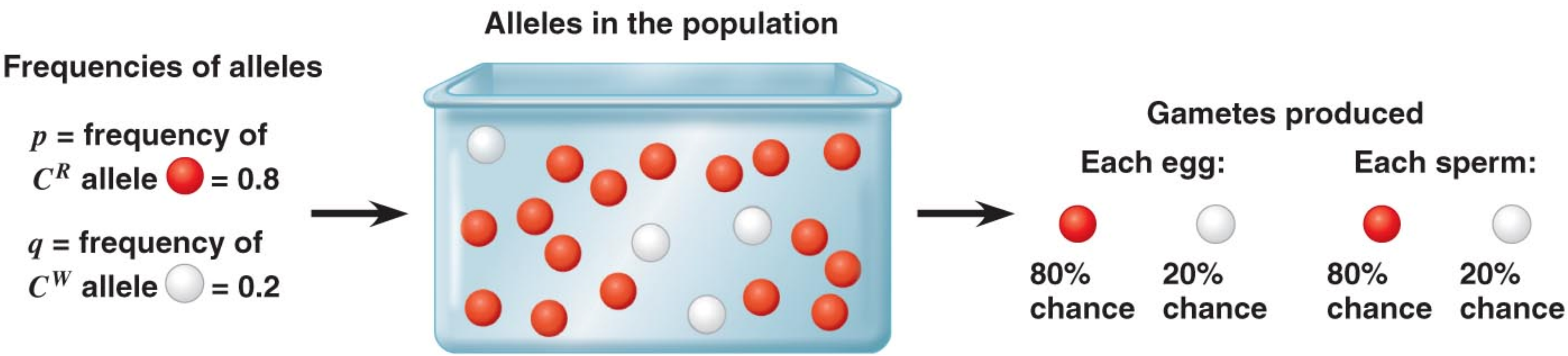
16) Introducing the Hardy-Weinberg Principle

- The Hardy-Weinberg principle describes a population that is <u>not evolving</u>
- If a population does <u>not</u> meet the criteria of the Hardy-Weinberg principle, it can be concluded that the population is evolving

17) The Hardy-Weinberg Principle, cont.

- The **Hardy-Weinberg principle** formally describes a state in a population where the frequencies of alleles and genotypes remain constant from generation to generation (and is therefore not evolving)
- In a given population where gametes contribute to the next generation randomly, allele frequencies will not change
- Mendelian inheritance preserves genetic variation in a population

18) Selecting alleles at random from a gene pool:



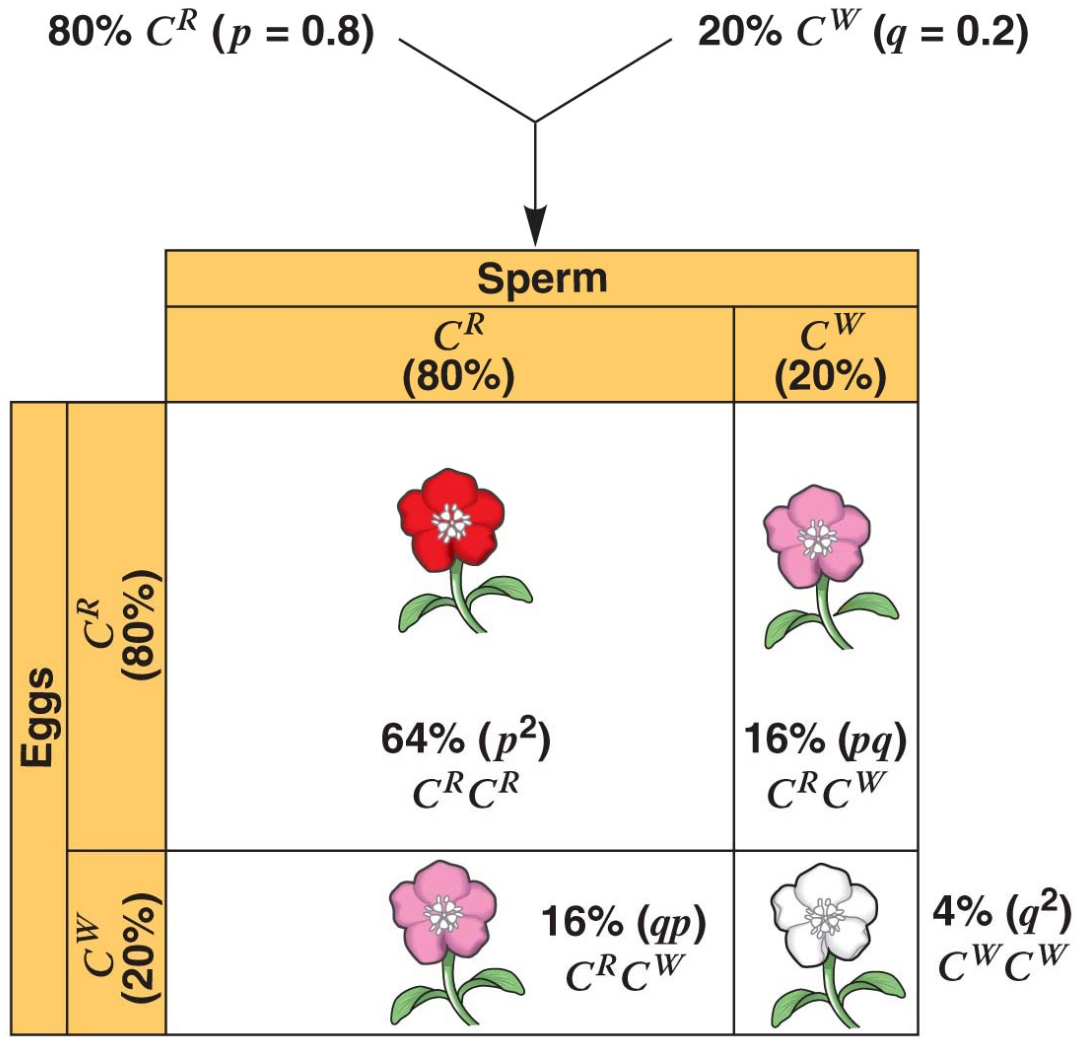
- 16 out of 20 are the red allele (C^R), so 16/20 = 80% = a frequency of 0.8, and the white allele (C^W) = 4/20 = 20% = 0.2 frequency.
- Assuming random mating, there is an 80% chance the egg carries a C^R and a 20% chance it carries the C^W allele, and by the same reasoning, the sperm would have an 80% chance of carrying a C^R and a 20% chance of carrying the C^W allele.

19) Hardy-Weinberg Equilibrium, cont.

- Hardy-Weinberg equilibrium describes a population (gene pool) in which random mating occurs, therefore allele frequencies do not change
- If p and q represent the relative frequencies of the only two possible alleles in a population at a particular locus, then
  - $p^2 + 2pq + q^2 = 1$
  - where p^2 and q^2 represent the frequencies of the homozygous genotypes and $2pq$ represents the frequency of the heterozygous genotype

20) Punnet square illustration of flower color

- Note that the gametes for each generation are drawn at random from the gene pool of the previous generation (slide 18)
- Mendelian processes alone do NOT alter frequencies of alleles or genotypes
- As long as random mating occurs, the frequencies remain in Hardy-Weinberg equilibrium (slide 21)



21) Note the continuance (constancy) of the genotype ratios in each generation (64% C^RC^R, 32% C^RC^W, & 4% C^WC^W)

64% C^RC^R, 32% C^RC^W, and 4% C^WC^W

Gametes of this generation:

64% C^R + 16% C^R = 80% C^R = 0.8 = p

4% C^W + 16% C^W = 20% C^W = 0.2 = q

64% C^RC^R, 32% C^RC^W, and 4% C^WC^W

Gametes of this generation:

64% C^R + 16% C^R = 80% C^R = 0.8 = p

4% C^W + 16% C^W = 20% C^W = 0.2 = q

Genotypes in the next generation:

64% C^RC^R, 32% C^RC^W, and 4% C^WC^W plants

22) The Hardy-Weinberg Equilibrium results of the preservation of allele and genotype frequencies

- The Hardy-Weinberg theorem describes a hypothetical population
- In real populations, allele and genotype frequencies <u>do</u> change over time

23) When the equilibrium is disturbed, then the population evolves

- The five conditions for non-evolving populations (which are not often met in nature):
  - Extremely large population size
  - No gene flow
  - No mutations
  - Random mating
  - No natural selection

24) Evolution can be occurring at one gene loci and non-evolving a others

- Natural populations are commonly in H-W Equilibrium for specific genes
- In natural populations some loci can be evolving while other loci are in H-W Equilibrium

25) Applying the Hardy-Weinberg Principle

- We can assume the locus that causes phenylketonuria (PKU) is in Hardy-Weinberg equilibrium given that:
  - The PKU gene mutation rate is low
  - Mate selection is random with respect to whether or not an individual is a carrier for the PKU allele

26) The PKU story, cont.

- Natural selection can only act on rare homozygous individuals who do not follow dietary restrictions
- The population is large
- Migration has no effect as many other populations have similar allele frequencies

27) The PKU story, cont.

- The occurrence of PKU is 1 per 10,000 births
  - $q^2 = 0.0001$
  - $q = 0.01$
- The frequency of normal alleles is
  - $p = 1 – q = 1 – 0.01 = 0.99$
- The frequency of carriers is
  - $2pq = 2 \times 0.99 \times 0.01 = 0.0198$
  - or approximately 2% of the U.S. population

28) So what, then, can alter allele frequencies in a population?

- Three major factors alter allele frequencies and bring about most evolutionary change:
  - Natural selection
  - Genetic drift
  - Gene flow

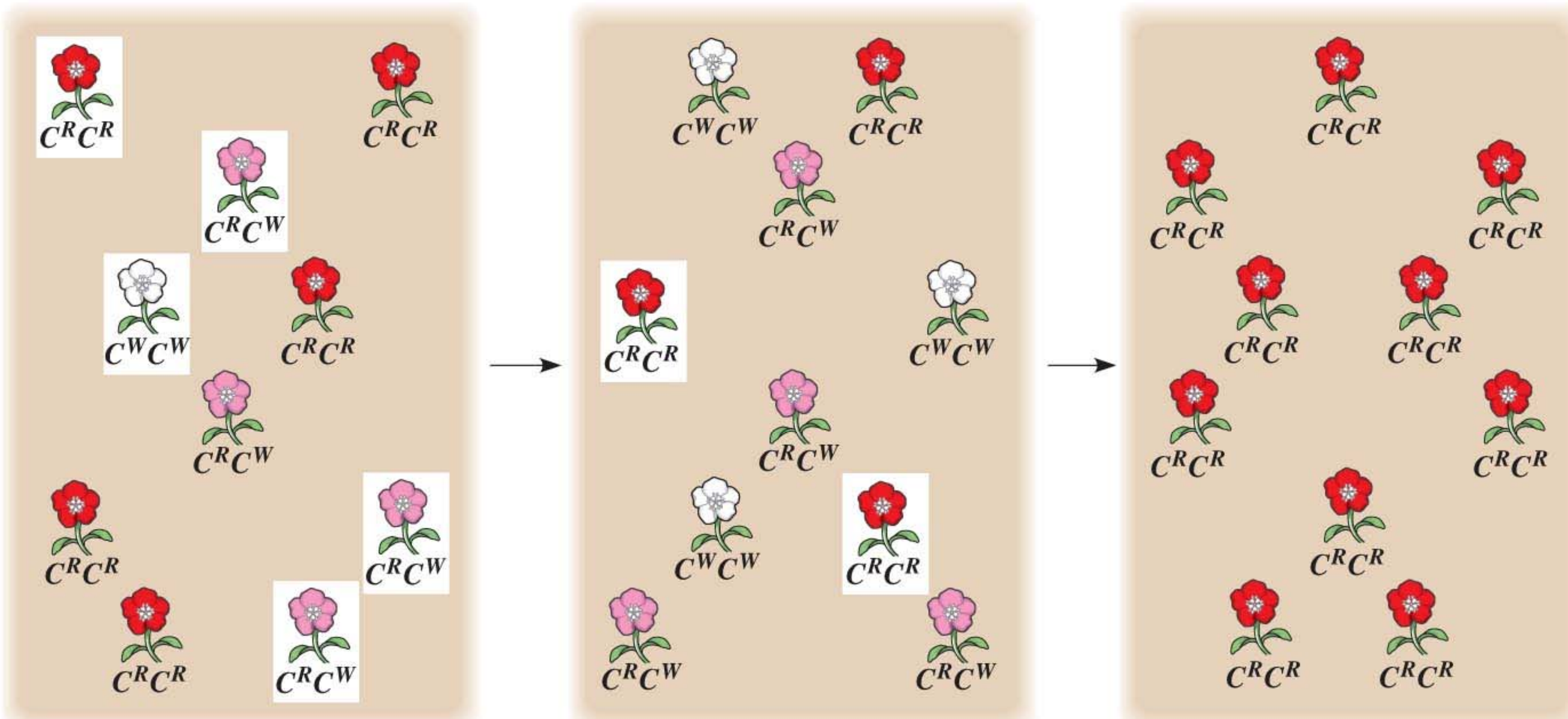
29) Natural selection

- Differential success in reproduction results in certain alleles being passed to the next generation in greater proportions

30) Genetic Drift

- The smaller a sample, the greater the chance of deviation from a predicted result
- **Genetic drift** describes how allele frequencies fluctuate unpredictably from one generation to the next
- Genetic drift tends to reduce genetic variation through losses of alleles

31) A graphic example of genetic drift in a small population (10 plants)



Generation 1
p (frequency of C^R) = 0.7
q (frequency of C^W) = 0.3

Generation 2
$p = 0.5$
$q = 0.5$

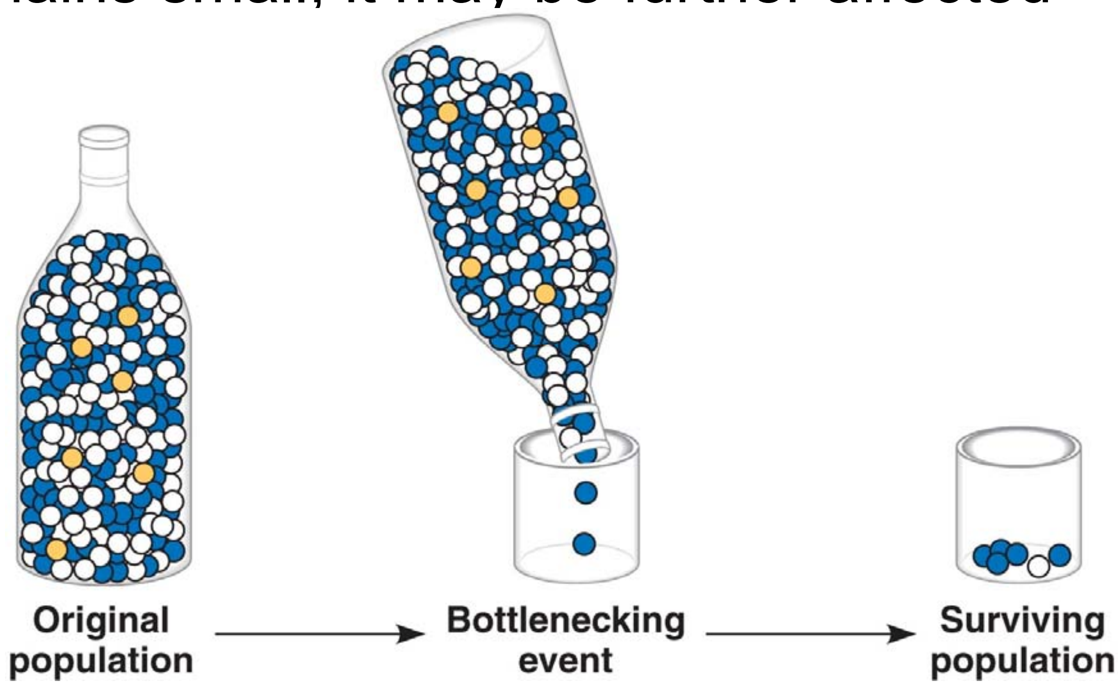
Generation 3
$p = 1.0$
$q = 0.0$

32) Two special examples of genetic drift—the Founder and Bottleneck effects

- The **founder effect** occurs when a few individuals become isolated from a larger population
- Allele frequencies in the small founder population can be different from those in the larger parent population
- High incidence of *retinitis pigmentosa* in residents of Tristan da Cunha

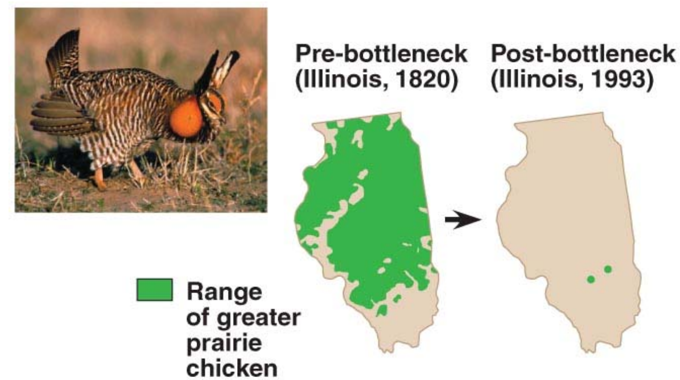
33) The Bottleneck Effect

- The **bottleneck effect** is a sudden reduction in population size due to a change in the environment
- The resulting gene pool may no longer be reflective of the original population's gene pool
- If the population remains small, it may be further affected by genetic drift



34) Impact of Genetic Drift on the Greater Prairie Chicken

- Loss of prairie habitat caused a severe reduction in the population of greater prairie chickens in Illinois
- The surviving birds had low levels of genetic variation, and only 50% of their eggs hatched



(a)

Location	Population size	Number of alleles per locus	Percentage of eggs hatched
Illinois			
1930–1960s	1,000–25,000	5.2	93
1993	<50	3.7	<50
Kansas, 1998 (no bottleneck)	750,000	5.8	99
Nebraska, 1998 (no bottleneck)	75,000–200,000	5.8	96
Minnesota, 1998 (no bottleneck)	4,000	5.3	85

(b)

35) Summarizing the effects of Genetic Drift

1. Genetic drift is significant in small populations
2. Genetic drift causes allele frequencies to change at random
3. Genetic drift can lead to a loss of genetic variation within populations
4. Genetic drift can cause harmful alleles to become fixed