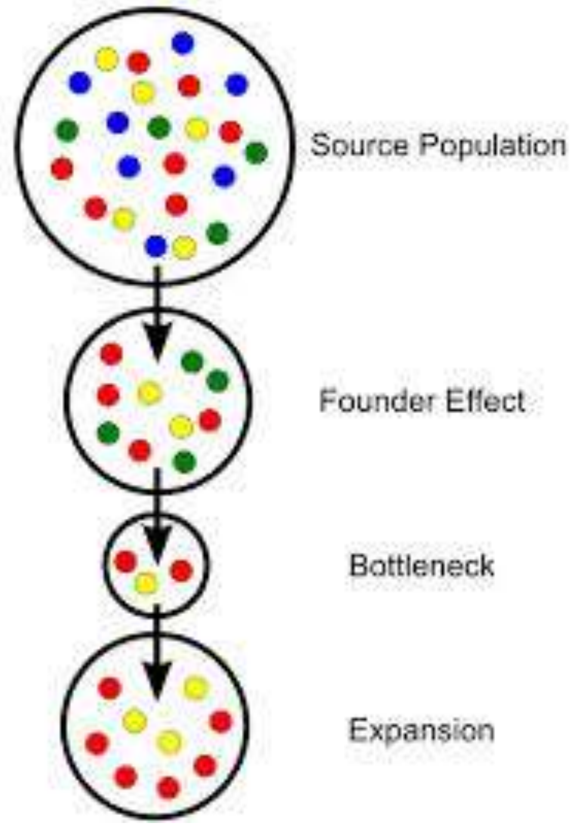
Population Genetics



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

What is evolution?

Change in population over time

Biological evolution

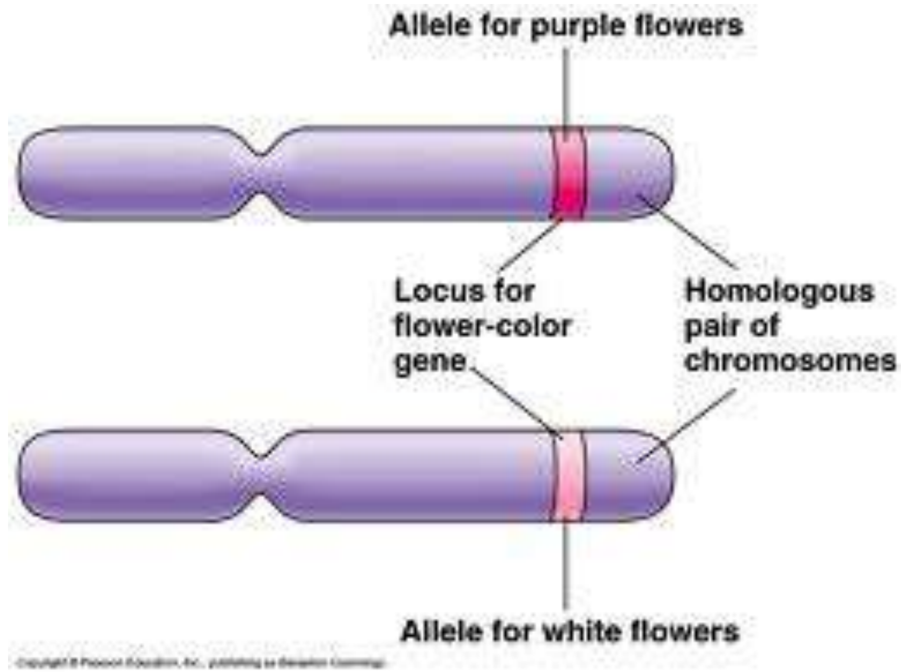
change in allele frequencies in populations
genetic changes in populations

Microevolution

changes within populations and species due to natural selection and other evolutionary forces (mutation, drift)
processes eventually leading toward speciation

Macroevolution

big changes between species, genera, families, phyla
takes place over long periods of time

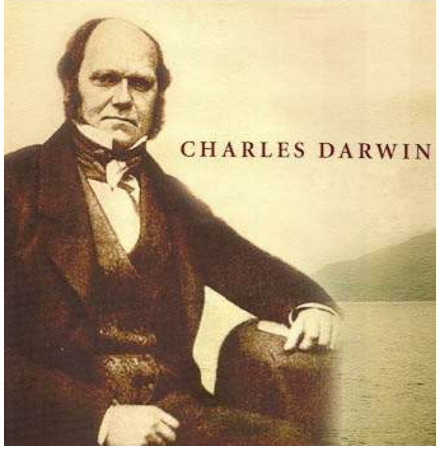


An allele can be dominant, recessive, or co-dominant with the others.

If the two alleles are different, and one is **dominant**, then the character expressed is the one of the dominant allele.

If they are **co-dominant**, then a bit of everything is expressed.

For a **recessive** allele to be expressed, it has to be either alone or present on both chromosomes of a homologous pair.



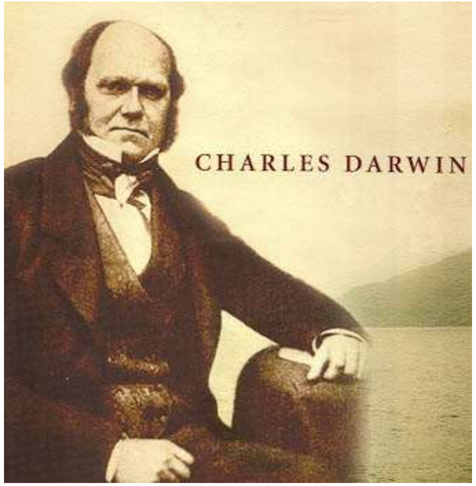
"Preservation of Favored Races in the Struggle for Life" = Natural Selection

1. There is **variation** in morphology, function or behavior between individuals.
2. Some traits are more **adaptive** than others.
3. Traits are **heritable**.
4. Individuals that are more **"fit"** live to reproduce or **reproduce more**.
5. Less adaptive traits become less common in populations

Fitness

A fundamental concept in evolutionary theory is "fitness", which can defined as the ability to survive and reproduce. Reproduction is key: to be evolutionarily fit, an organism must pass its genes on to future generations.

Basic idea behind evolution by natural selection: the more fit individuals contribute more to future generations than less fit individuals. Thus, the genes found in more fit individuals ultimately take over the population.



Darwin never understood heredity.

Pangenesis, Blending Inheritance.

Believed in slow, gradual change.

Mendel published on peas (1866)

Copy of Mendel's paper found in Darwin's library years after his death. Darwin did read German; but no evidence Darwin ever read Mendel's paper, or understood it

Mendel's Laws rediscovered 1900, but the "Gradualists" thought these were important only in exceptional cases.

Many believed large mutations were source of variation for new species.

What are the sources of variation in a gene pool?

This was a puzzle to Darwin and contemporaries.

Fertilization - combines alleles from two parents

Independent assortment in meiosis (metaphase I) - mixes paternal and maternal chromosomes

Crossing over at meiosis - recombines alleles in chromosomes

Changes in chromosome structure - deletions, insertions, inversions, translocation

Gene mutation - creates new alleles, important in the long run.

Variation

- Mendel’s genetics rediscovered, 1900
  - Correns, Tschermak, DeVries
- Years of dispute over cause of evolution
  - Natural selection vs. genetics
- Neo-Darwinists - 1920s –
  - Ronald Fisher, J.B.S. Haldane, Sewall Wright
- “Modern Synthesis” 1930’s
  - Genetics and natural selection reconciled

Neo-Darwinists - 1920s

Ronald Fisher, J.B.S. Haldane, Sewall Wright

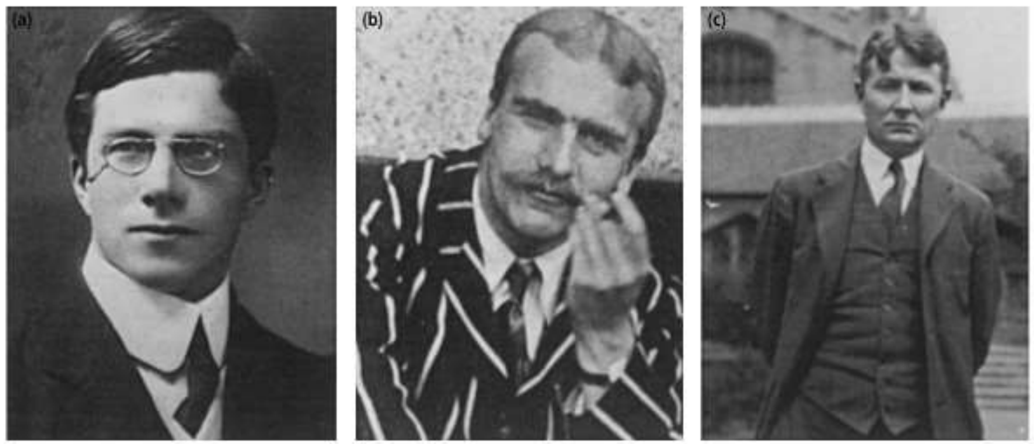
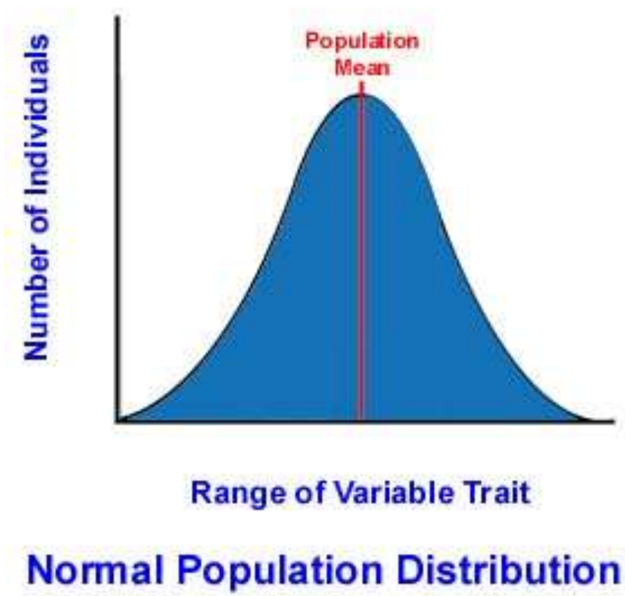


Figure 1.8
(a) Ronald Aylmer Fisher (1890–1962) in 1912, as a Steward at the First International Eugenics Conference.
(b) J.B.S. Haldane (1892–1964) in Oxford, UK in 1914.
(c) Sewall Wright (1889–1988) in 1928 at the University of Chicago.

Shift in emphasis from individuals to populations

Worked out the statistical foundation of population genetics



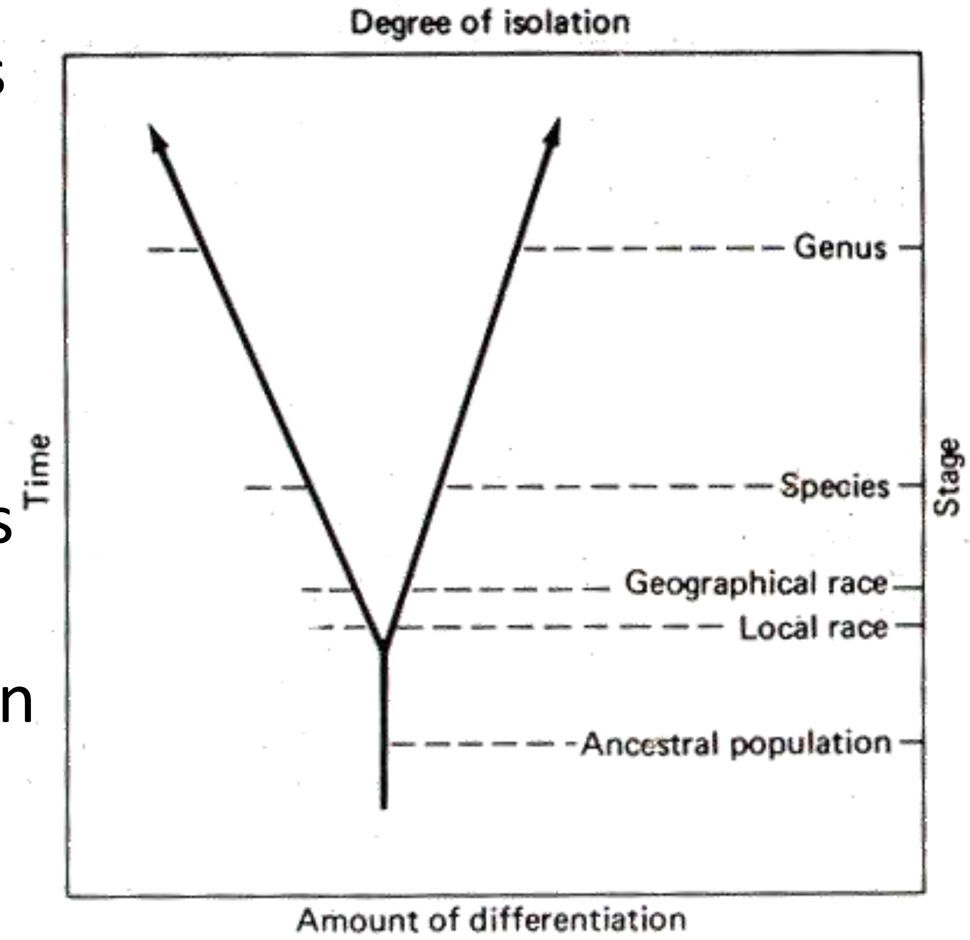
Population Genetics and Gene (Allele) Frequencies in Populations

The neo-Darwinian theory

- Evolution is a population phenomenon
- Evolution occurs when there is a change in gene (now allele) frequencies in a population because of various natural forces such as mutation, selection and genetic drift
- These changes in allele frequencies lead to differences among populations, species, and higher clades
- This population genetics view of evolution became known as neo-Darwinian theory with its emphasis on the frequency of genes in populations

The Neo-Darwinian synthesis helped explain how mutation led to variation and how selection led to adaptation.

Change begins in populations and leads to higher level differences, those between races, species, genera and higher categories.



Review some terms…..

Gene – a discrete unit of hereditary information consisting of a specific nucleotide sequence in DNA

Alleles – alternative forms of a gene (Aa, Bb)

Allele frequency = proportion of any specific allele in a population

Allele frequencies are estimated from genotype frequencies

Genotype – the genetic makeup of an individual

Genotype frequency - proportion of individuals in a population with a specific genotype

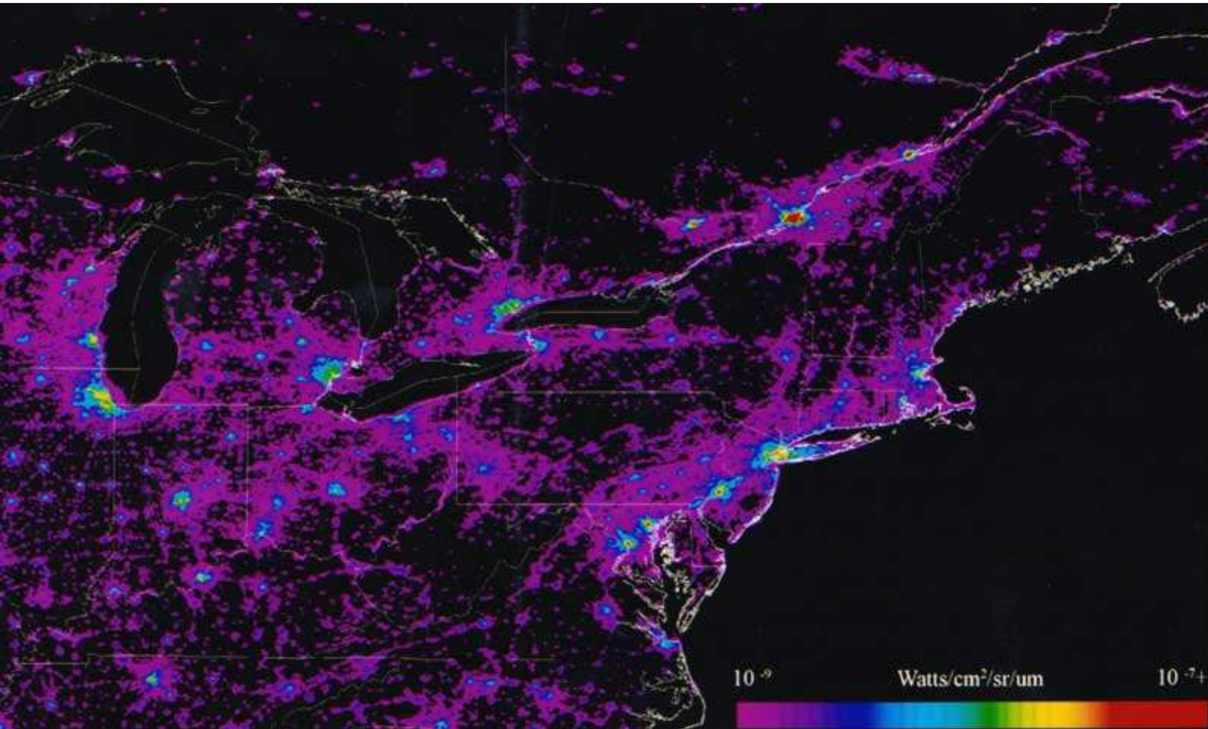
Genotype frequencies may differ from one population to another

Phenotype – the physical and physiological traits of an organism

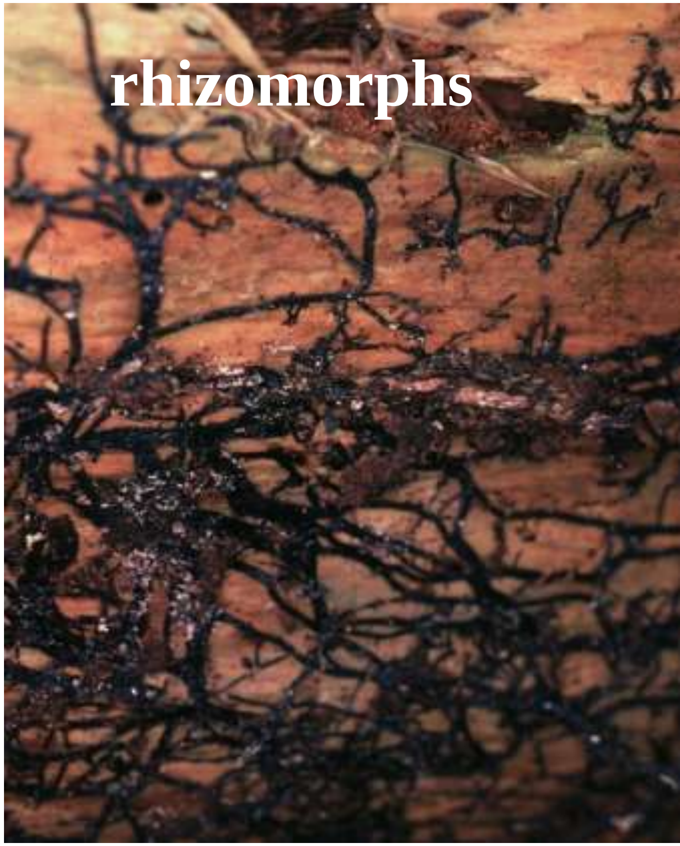
Populations

Group of individuals of the same species that can interbreed with one another

Some species occupy a wide geographic range and are divided into discrete populations


10^{-9}
Watts/cm^2/sr/um
10^{-7}+

Giant clones, all one individual



World’s Biggest Individual?
Giant fungud
Connected underground

Armillaria ostoyae
2,400 years, killing trees as it grows. Now about 880 hectares of the Malheur National Forest in eastern Oregon.

Poplars, Rocky Mountains, one tree, many trunks, connected underground

Population - a localized group of individuals of the same species.

Species - a group of populations whose individuals have the ability to breed and produce fertile offspring.

Individuals near a population center are, on average, more closely related to one another than to members of other populations.

Population - group of organisms of the same species living in the same geographical area

Subpopulation - any of the breeding groups within a population among which migration is restricted

Local population - subpopulation within which most individuals find their mates

Gene Pool

The **gene pool** is all of the genes and different alleles in a population

We study genetic variation within the gene pool and how variation changes from one generation to the next

Emphasis is often on variation in alleles between members of a population at certain loci of interest

Allele Frequencies

A population's gene pool includes all the alleles for all the loci present in the population

- Diploid organisms have a maximum of two different alleles at each genetic locus
- Typically, a single individual therefore has only a small fraction of the alleles for a given locus that are present in the population as a whole



Genes in Natural Populations Are Usually Polymorphic

Polymorphism – many traits display variation within a population

Due to 2 or more alleles at a locus that influence a phenotype

Polymorphic gene/locus - 2 or more alleles

Monomorphic gene/locus– predominantly a single allele

Single nucleotide polymorphism (SNPs)

- Smallest type of genetic change in a gene
- Most common – 90% of the variation in human gene sequences

Large, healthy populations exhibit a high level of genetic diversity

Polymorphisms are the raw material for evolution

Gene frequencies - the proportion of each kind of allele in the whole population.

To get these proportions we count the total number of individuals in the population and estimate the relative frequencies of the alleles.

p = frequency of dominant allele

q = frequency of recessive allele

p + q = 1

Example

90 TT, 60 Tt, 50 tt = 200 individuals, 400 alleles

240 of these are T (freq.=0.60), 160 are t (freq.=0.40)

90 TT, 60 Tt, 50 tt = 200 individuals, 400 alleles

240 of these are T

90+90+60=240

<u>freq. T= 240/400 = 0.60</u>

160 are t

50+50+60=160

<u>freq. t =160/400 =0.40</u>

Allele frequencies define gene pools

500 flowering plants

480 red flowers — **20 white flowers**

320 RR — **160 Rr** — **20 rr**

As there are 1000 copies of the genes for color,
the allele frequencies are (in both males and females):

320 x 2 (RR) + 160 x 1 (Rr) = 800 R; 800/1000 = 0.8 (80%) R
160 x 1 (Rr) + 20 x 2 (rr) = 200 r; 200/1000 = 0.2 (20%) r

Genes in Populations Hardy-Weinberg

The Hardy-Weinberg Principle

Hardy-Weinberg Principle/equilibrium

G. H. Hardy (1877-1947)

English mathematician

Wilhelm Weinberg (1862-1937)

German physician & geneticist

The Hardy-Weinberg Principle

Working independently just a few years after the rediscovery of Mendelian genetics they concluded that:

The original proportions of the genotypes in a population remain constant from generation to generation as long as five assumptions are met.

The Hardy-Weinberg Principle

Five assumptions: If:

1. The population size is very large
2. Random mating is occurring
3. No mutation occurs
4. No selection occurs
5. No alleles transfer in or out of the population (no migration)

Then allele frequencies in the population will remain constant through future generations

Simplifying Assumptions for The Hardy-Weinberg Principle

1) diploid organisms
2) sexual reproducing organisms
3) generations are non-overlapping
4) all genotypes equally viable

If these assumptions are not met, it complicates the mathematics for the analyses

Whether or not these assumptions are all met, biologists can use Mendelian ratios and Hardy-Weinberg analysis to measure rates of evolution

The Hardy-Weinberg Principle

- p = frequency for first allele in the population
- q = frequency for second allele in the population
- Calculate allele frequencies with a binomial equation:

$$p + q = 1$$

- because there are only two alleles:

  p + q must always equal 1 (100% of the alleles)

[Note: more alleles can be handled, with three alleles: $p + q + r = 1$]

The Hardy-Weinberg Principle

Calculate genotype frequencies with a binomial expansion

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

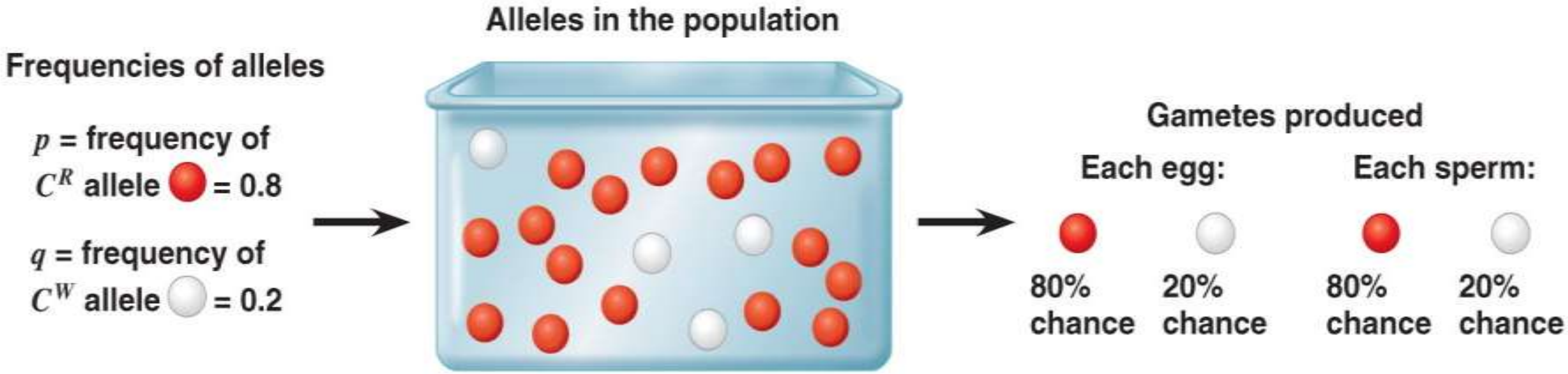
- p^2 = individuals homozygous for first allele
- $2pq$ = individuals heterozygous for the alleles
- q^2 = individuals homozygous for second allele

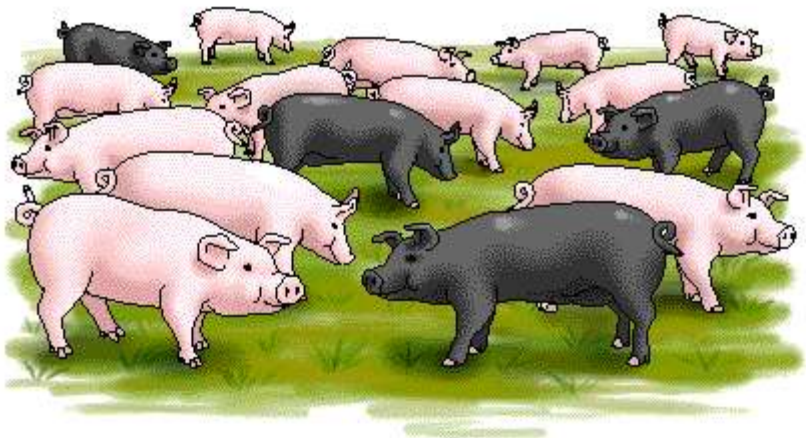
- because there are three phenotypic classes:

  $p^2 + 2pq + q^2$ must always equal 1

Hardy-Weinberg Principle/equilibrium

Allele frequencies remain unchanged generation to generation





Black (b) is recessive to white (B)

Bb and BB pigs “look alike”
so can’t tell their alleles by observing their phenotype.

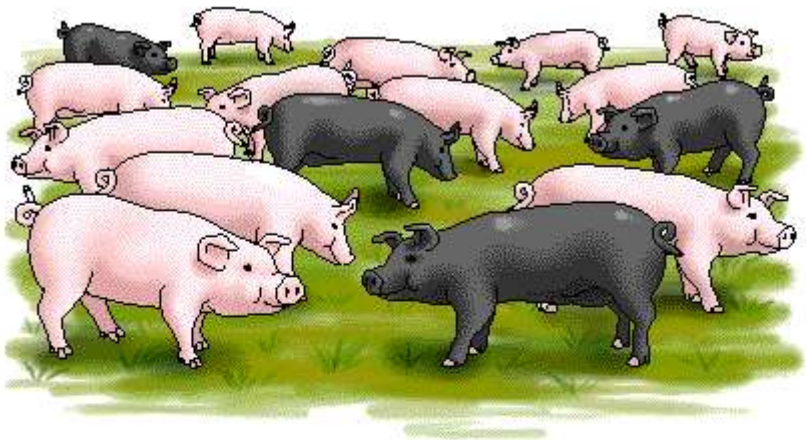
ALWAYS START WITH RECESSIVE alleles.
p= dominant allele   q = recessive allele

4/16 are black.

So bb or q^2 = 4/16 or 0.25

$q = \sqrt{0.25} = 0.5$

http://www.phschool.com/science/biology_place/labbench/lab8/samprob1.html



Once you know q

you can figure out p

. . . $p + q = 1$

$p + q = 1$

$p + 0.5 = 1$

$p = 0.5$

Now you know the **allele** frequencies.

The frequency of the recessive (b) allele $q = 0.5$

The frequency of the dominant (B) allele $p = 0.5$

WHAT ARE THE GENOTYPIC FREQUENCIES?

You know pp from problem

bb or $q^2 = 4/16 = 0.25$

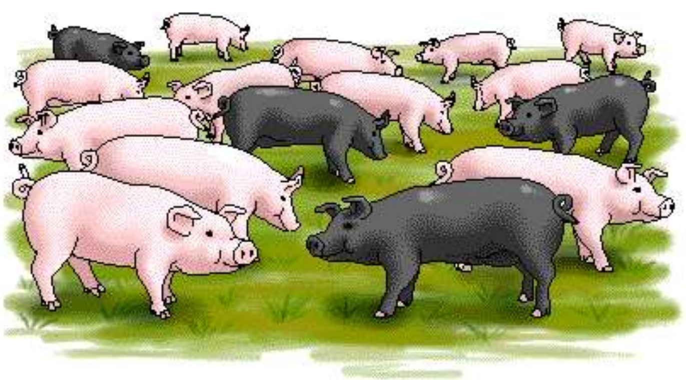
BB or $p^2 = (0.5)^2 = 0.25$

Bb = $2pq = 2\,(0.5)\,(0.5) = 0.5$

25% of population are bb

25% of population are BB

50% of population are Bb



http://www.phschool.com/science/biology_place/labbench/lab8/samprob1.html

The Hardy-Weinberg Theorem

Used to <u>describe a non-evolving population</u>.

Shuffling of alleles by meiosis and random fertilization have no effect on the overall gene pool.

Natural populations are not expected to actually be in Hardy-Weinberg equilibrium.

<u>Deviation from H-W equilibrium usually results in evolution.</u>

Understanding a non-evolving population, helps us to understand how evolution occurs.

The Hardy-Weinberg Principle

Five assumptions: If:

1. The population size is very large
2. Random mating is occurring
3. No mutation occurs
4. No selection occurs
5. No alleles transfer in or out of the population (no migration)

Then allele frequencies in the population will remain constant through future generations

But we know that evolution does occur within populations.

Evolution of a population = microevolution.

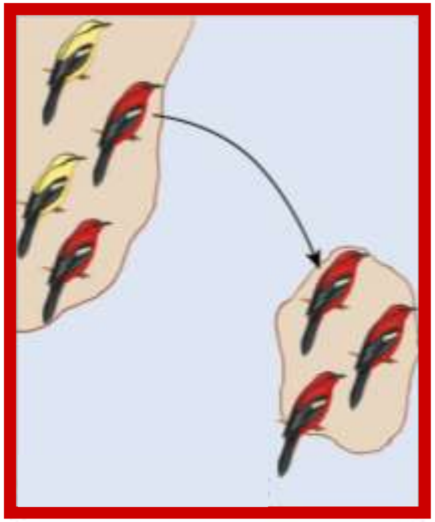
Microevolution refers to changes in allele frequencies in a gene pool from generation to generation. Represents a gradual change in a population.

Causes of microevolution:

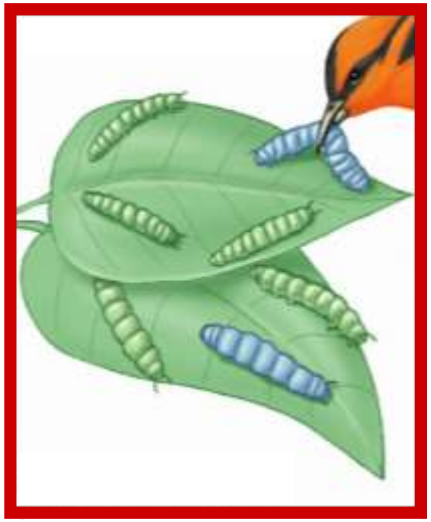
1) Genetic drift
2) Natural selection
3) Gene flow
4) Mutation
5) Nonrandom mating

5 Agents of evolutionary change

Genetic Drift

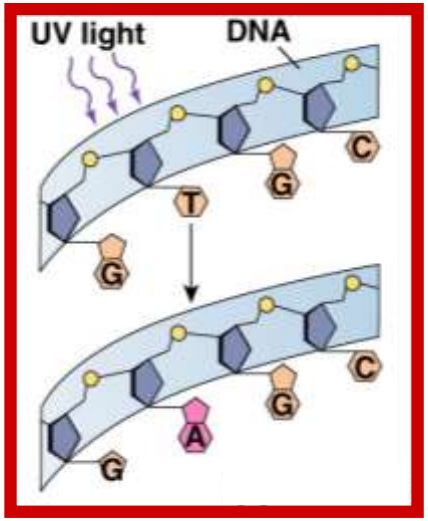


Selection



Gene Flow

Mutation



Non-random mating

Departures from HW Equilibrium

- Check Gene Diversity = Heterozygosity
  - If high gene diversity = different genetic sources due to high levels of migration
- Inbreeding - mating system “leaky” or breaks down allowing mating between siblings
- Asexual reproduction = check for clones
  - Risk of over emphasizing particular individuals
- Restricted dispersal = local differentiation leads to non-random mating