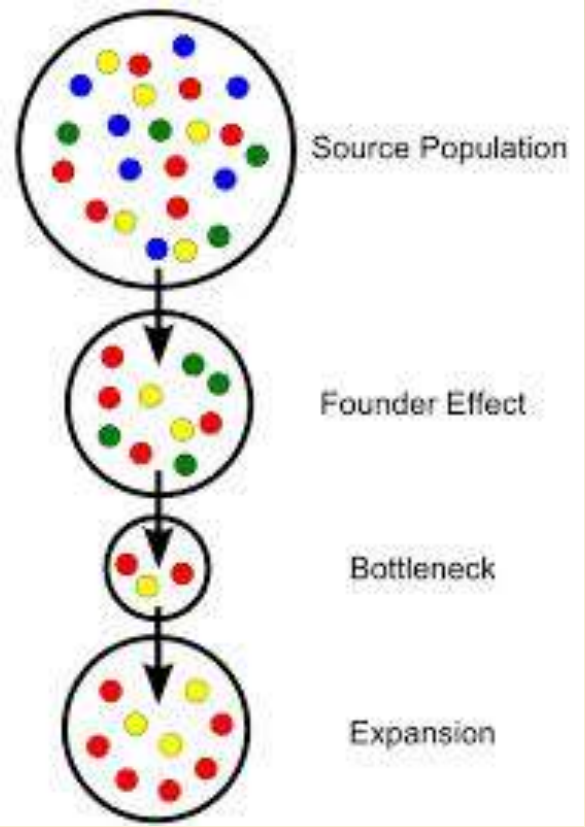
Population Genetics



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

What is evolution?

Change in population over time

***Biological* evolution**

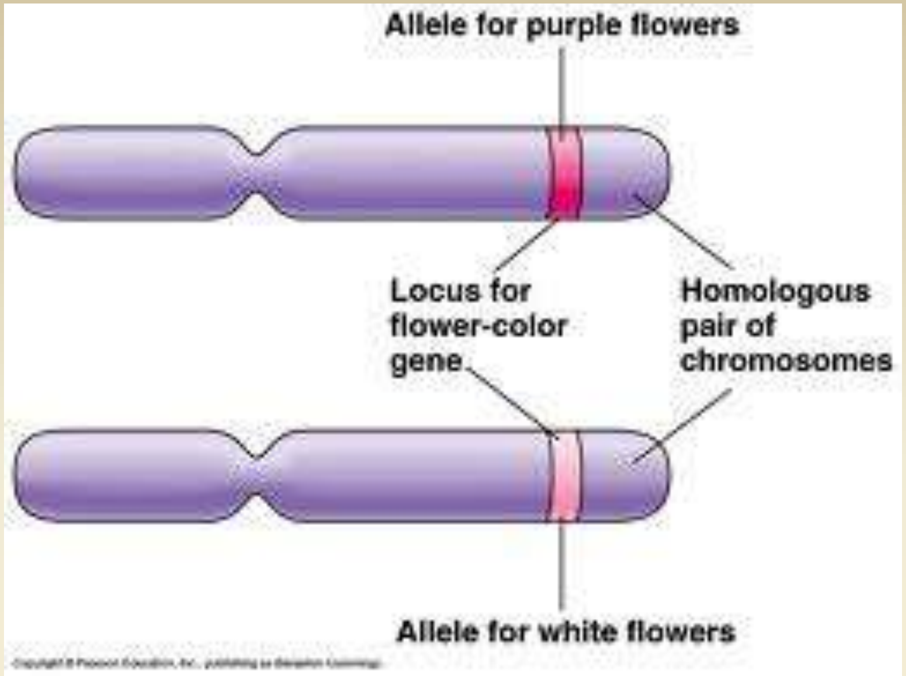
change in allele frequencies in populations
genetic changes in populations

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

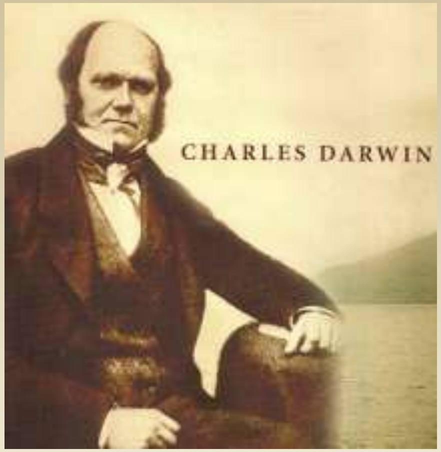


An allele can be dominant, recessive, or co-dominant with the others.

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

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

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

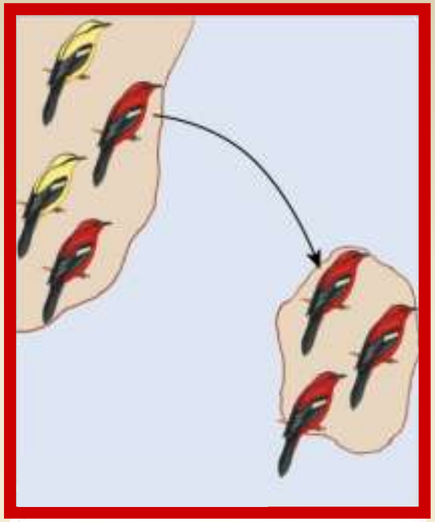
The Hardy-Weinberg Principles Describes a Population at *Genetic Equilibrium*

Genetic equilibrium requires:

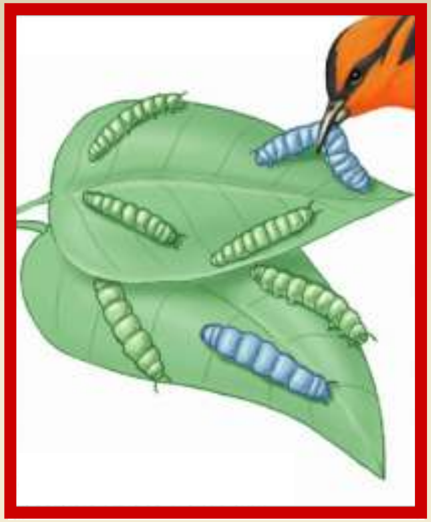
1. The population size is very large
2. Random mating is occurring
3. No mutation occurs
4. No selection occurs
5. No alleles transfer in or out of the population (no migration)

5 Agents of evolutionary change

Genetic Drift

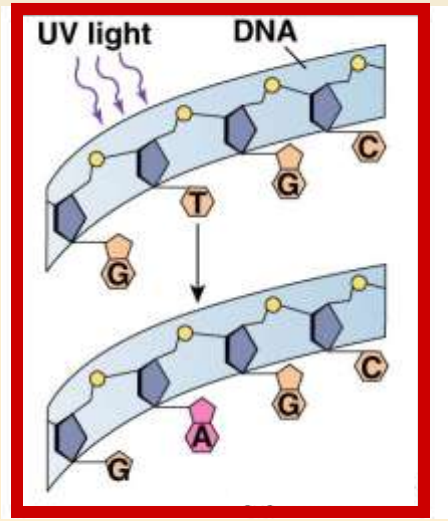


Selection



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

Adaptation

The consequence of natural selection is **adaptation**

Adaptation: some heritable aspect of form, function, behavior or development that improves the odds for surviving or reproducing in a given environment.

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

Deme is a locally interbreeding subset of the population

The deme is most often the focal point for evolutionary change

Consider the California yarrow, *Achillea borealis* and *A. lanulosa*

A. borealis

A. lanulosa

Demes Can Adapt to Local Conditions

Average height, timing and length of growing season, temperature and drought tolerance varied among the populations on an east-west transect

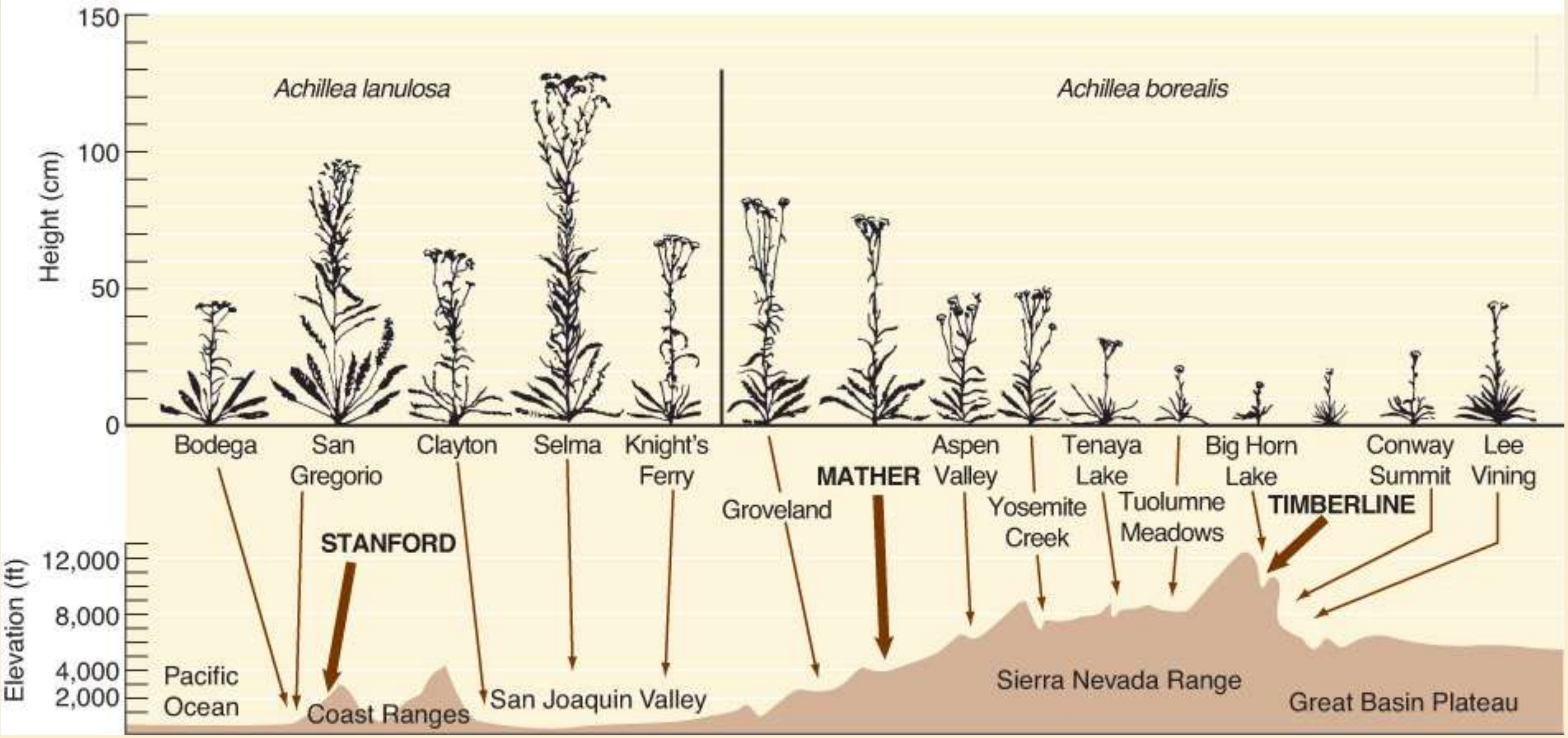


Figure B01A: Locations of the populations and indication of genetic differences that have evolved

Adapted from Clausen, J. D., D. Keck, and W. M. Hiesey, Carnegie Inst. Wash. Publ., No. 581, 1948: 1–219.

Demes Have Different Gene Pools

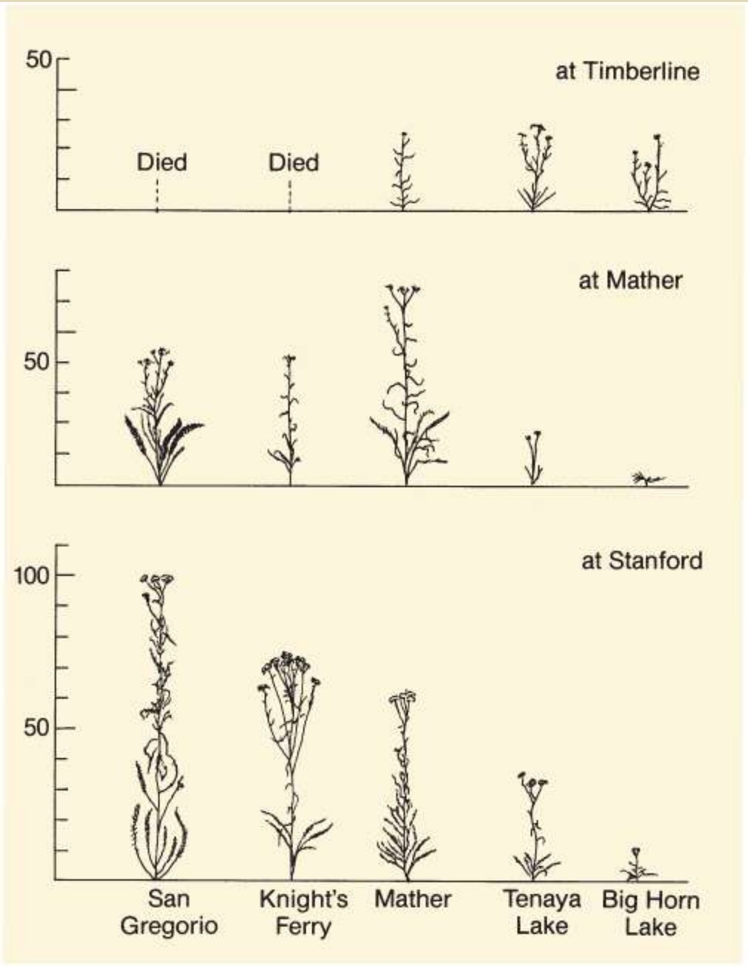


Figure B02: Responses of clones from representative of the common yarrow

Adapted from Clausen, J. D., D. Keck, and W. M. Hiesey, Carnegie Inst. Wash. Publ. No. 581, 1–219.

- Individual yarrow plants were drawn from five different locales, cut and grown vegetatively into mature individuals
- These clones of individuals were then planted at different locations
- They showed considerable differences in fitness; some even dying in a different locale

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

The Hardy-Weinberg Principle

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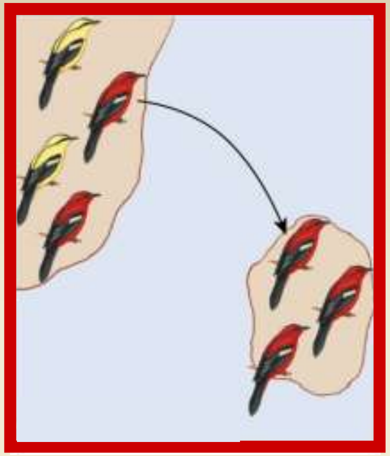
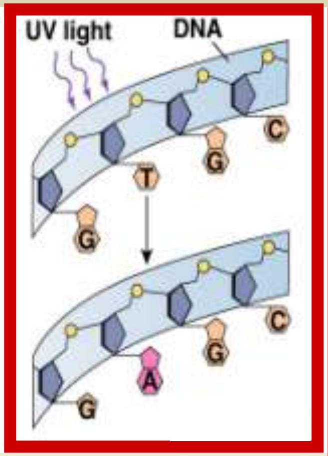
5 Agents of evolutionary change

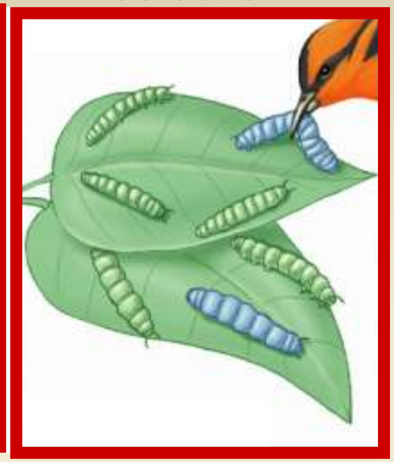
Genetic Drift



Mutation



Selection



Non-random mating

Gene Flow

A population ***not*** in Hardy-Weinberg equilibrium is one in which allele frequencies are changing generation to generation due to one or more of the five evolutionary agents are operating in a population

GENETIC DRIFT

Agents of Evolutionary Change

Small Population Size:

- When a population is large, then allele frequencies are very unlikely to change due to random sampling error
- When a population is small, then, just by chance, some individuals fail to mate at all, not because they are unfit
- When a population is small, then, just by chance, some offspring fail to survive to reproduce, not because they are unfit
- When a population is small, gene frequencies may change due to these sorts of random effects – this is called **genetic drift**

California condors



The theory of genetic drift was developed in the 1930s by Sewall Wright, and is sometimes referred to as the **Sewall Wright effect**.

Small Population Size

Hypothesis: how often will you get tails?

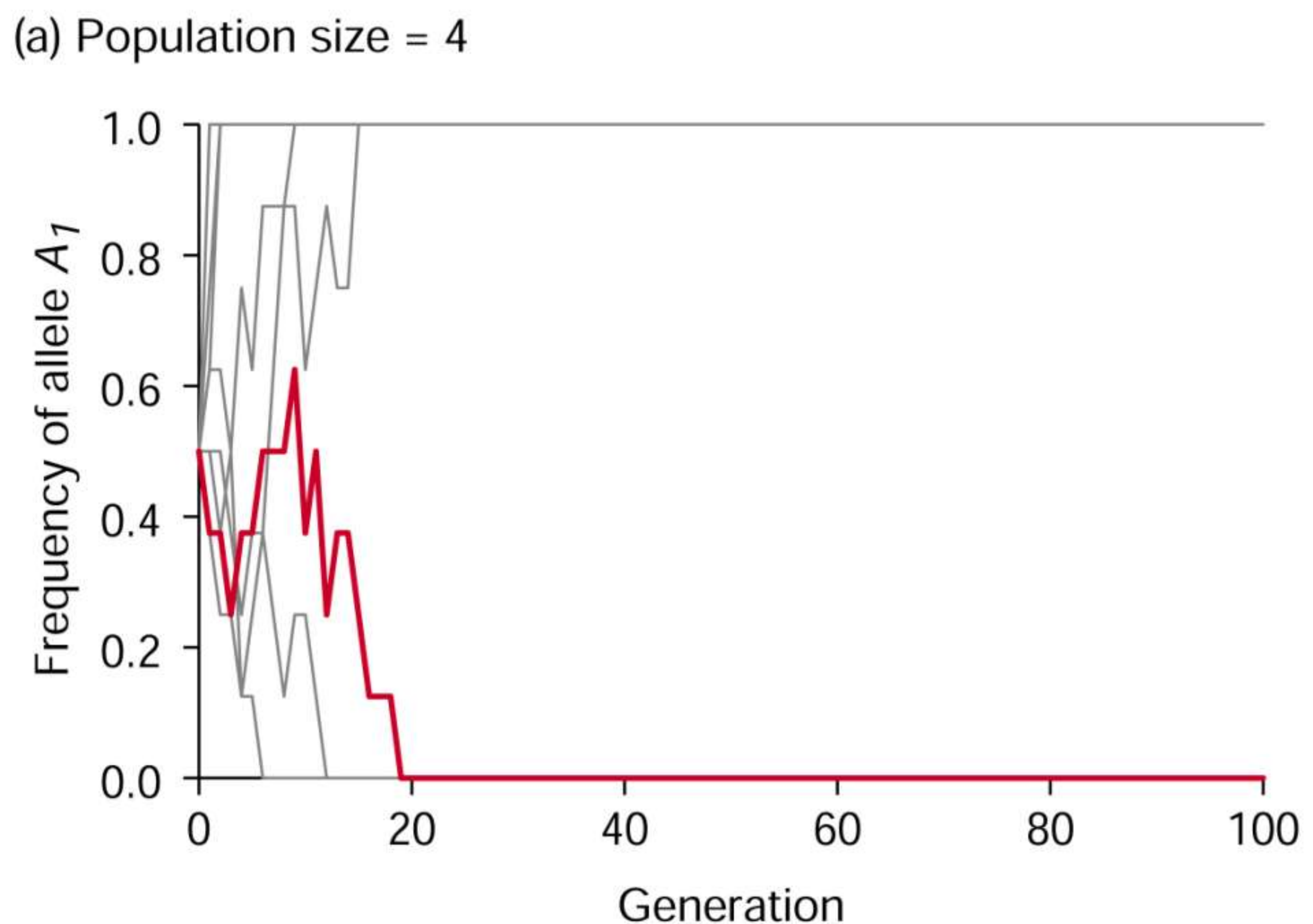
Flip a coin ten times:

- # heads
- # tails

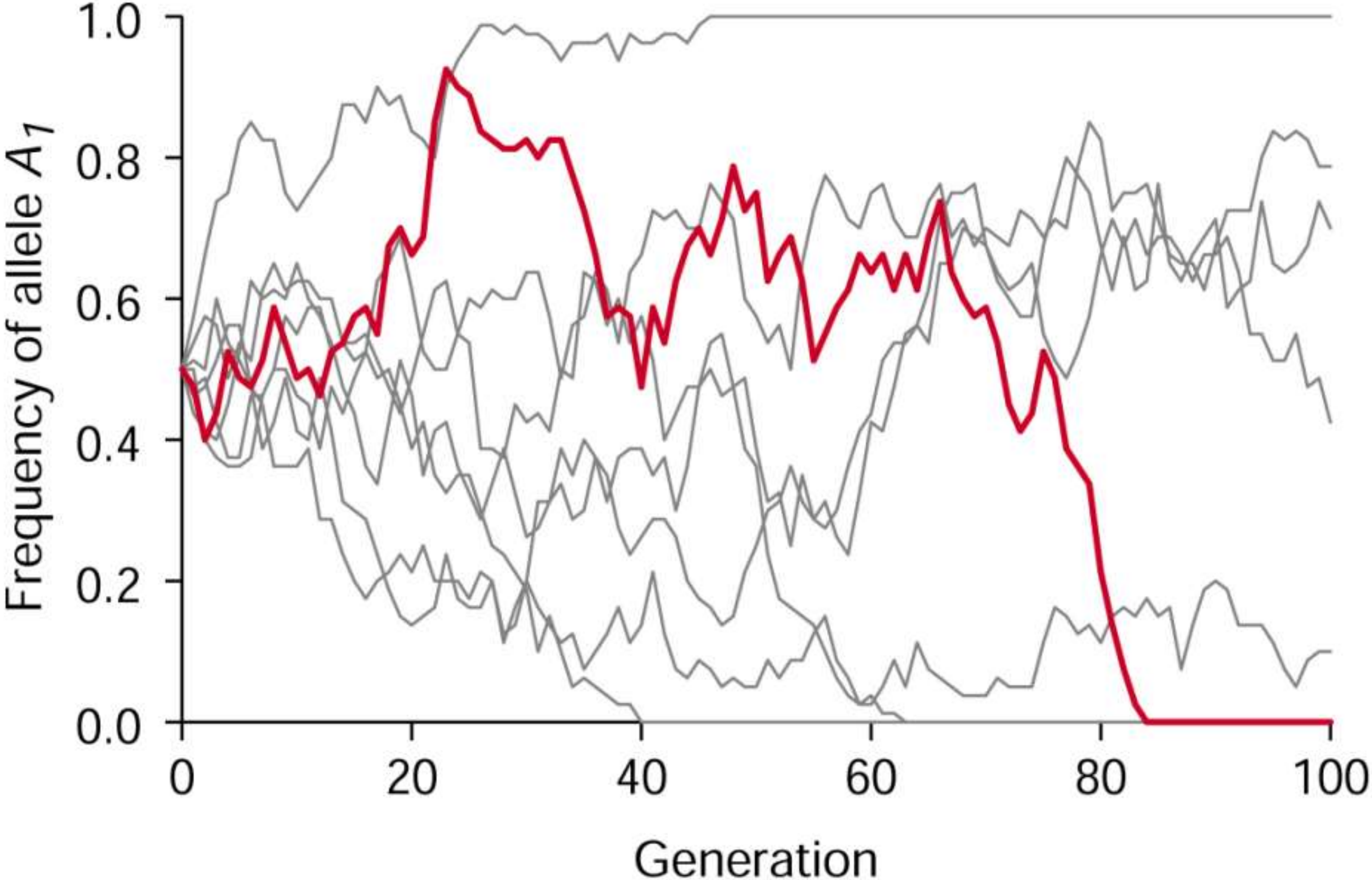
Everyone flip a coin ten times:

- # heads
- # tails

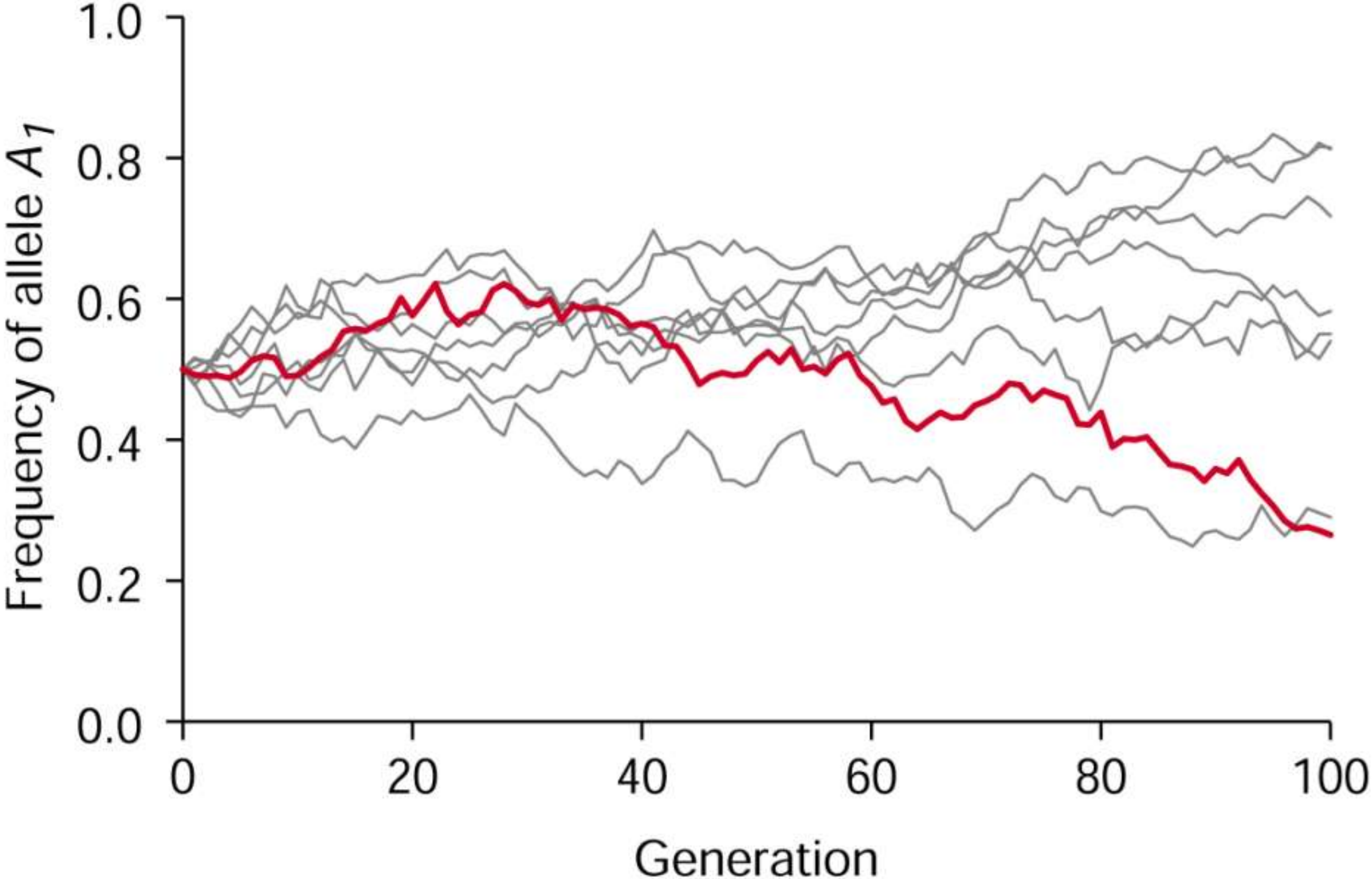
The difference between 10 and 200 events: **sampling error**


(a) Population size = 4
Frequency of allele A_1
1.0
0.8
0.6
0.4
0.2
0.0
0
20
40
60
80
100
Generation

(b) Population size = 40



(c) Population size = 400



Conclusions from simulations

- Populations follow unique paths
- Genetic drift has strongest effects on small populations.
- Given enough time, even in large populations genetic drift can have an effect.
- Genetic drift leads to fixation or loss of alleles, which increases homozygosity and reduces heterozygosity.

Genetic Drift

Genetic drift = the alteration of the gene pool of a small population due to chance.

Two factors may cause genetic drift:

- **Bottleneck effect** may lead to reduced genetic variability following some large disturbance that removes a large portion of the population. The surviving population often does not represent the allele frequency in the original population
- **Founder effect** may lead to reduced variability when a few individuals from a large population colonize an isolated habitat.

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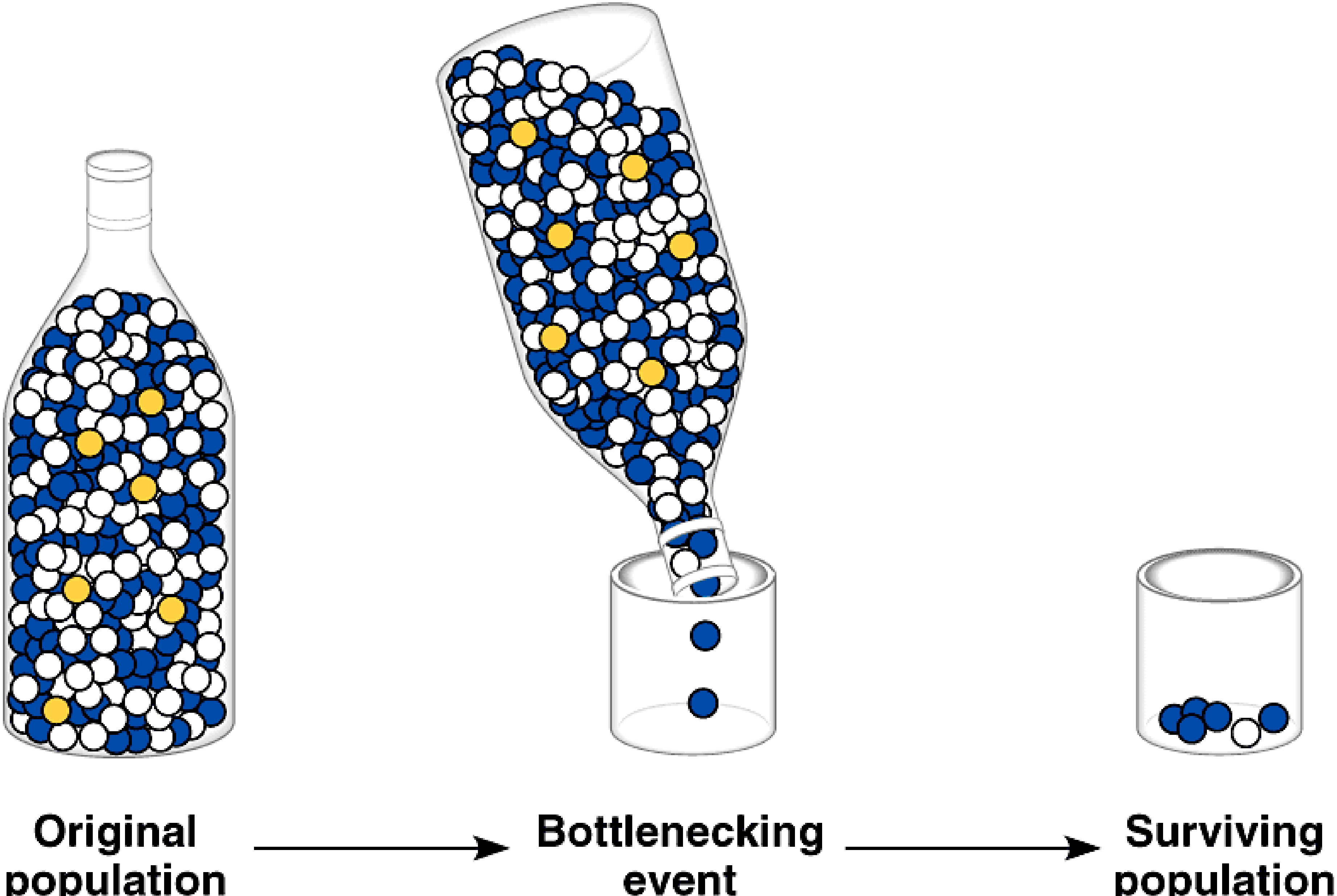
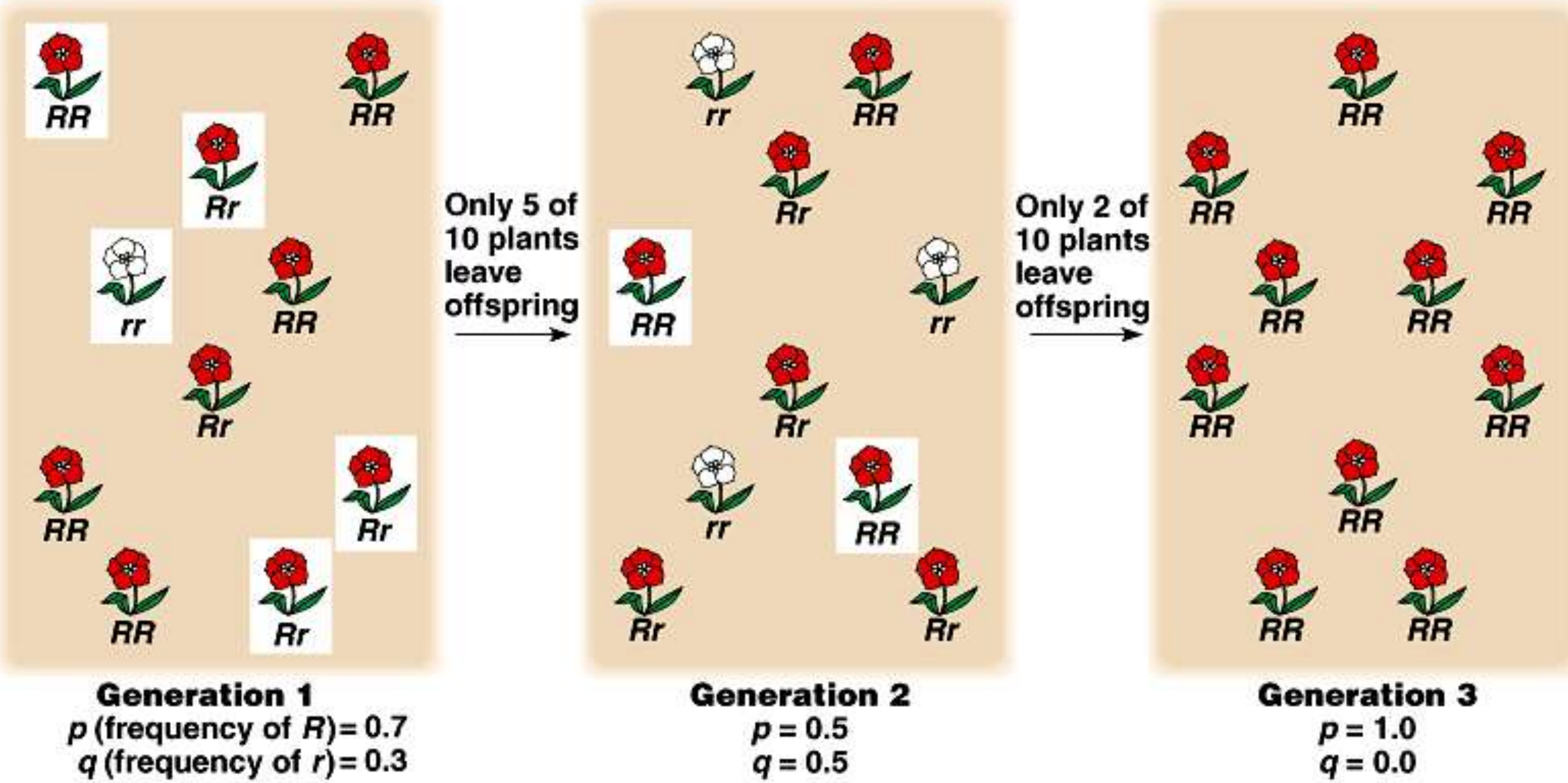
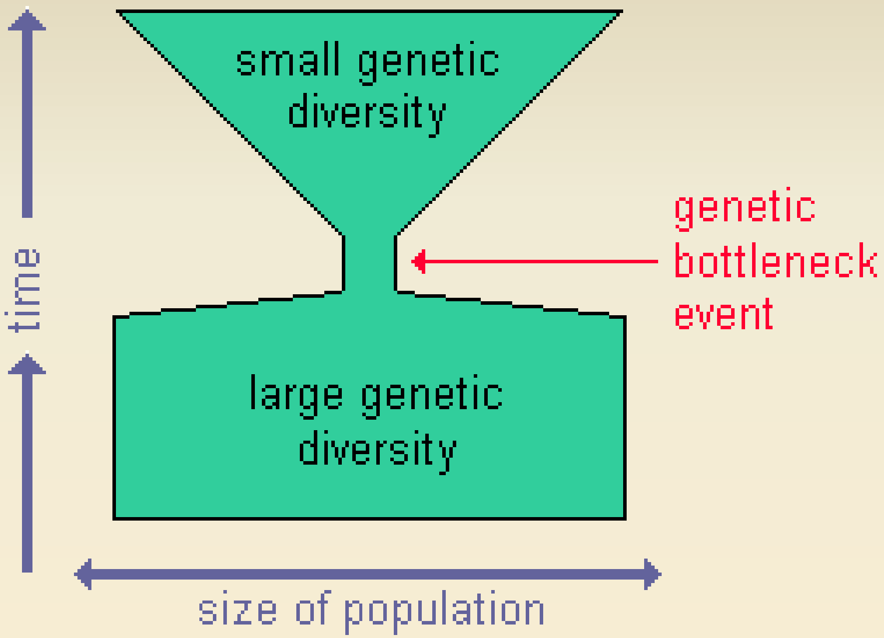

Original population
Bottlenecking event
Surviving population


RR
RR
Rr
rr
RR
Rr
RR
Rr
RR
Rr
Only 5 of 10 plants leave offspring
rr
RR
Rr
RR
rr
Rr
rr
RR
Rr
Rr
Only 2 of 10 plants leave offspring
RR
RR
RR
RR
RR
RR
RR
RR
RR
RR
Generation 1
p (frequency of R) = 0.7
q (frequency of r) = 0.3
Generation 2
p = 0.5
q = 0.5
Generation 3
p = 1.0
q = 0.0

Bottlenecks



A population bottleneck is essentially the same phenomenon as the founder effect, except that in a bottleneck, the entire species is wiped out except for a small group of survivors.

The allele frequencies in the survivors determines the allele frequencies in the population after it grows large once again.

Cheetahs, which have very little genetic variation, are presumed to have gone through several genetic bottlenecks.

The Bottleneck Effect

The actions of people sometimes cause bottlenecks in other species.

- N. California elephant seal population reduced to 20-100 individuals in the 1890s.
- Current population > 30,000.
- Variation *drastically* reduced – 24 genes with 1 allele.

Bottlenecks

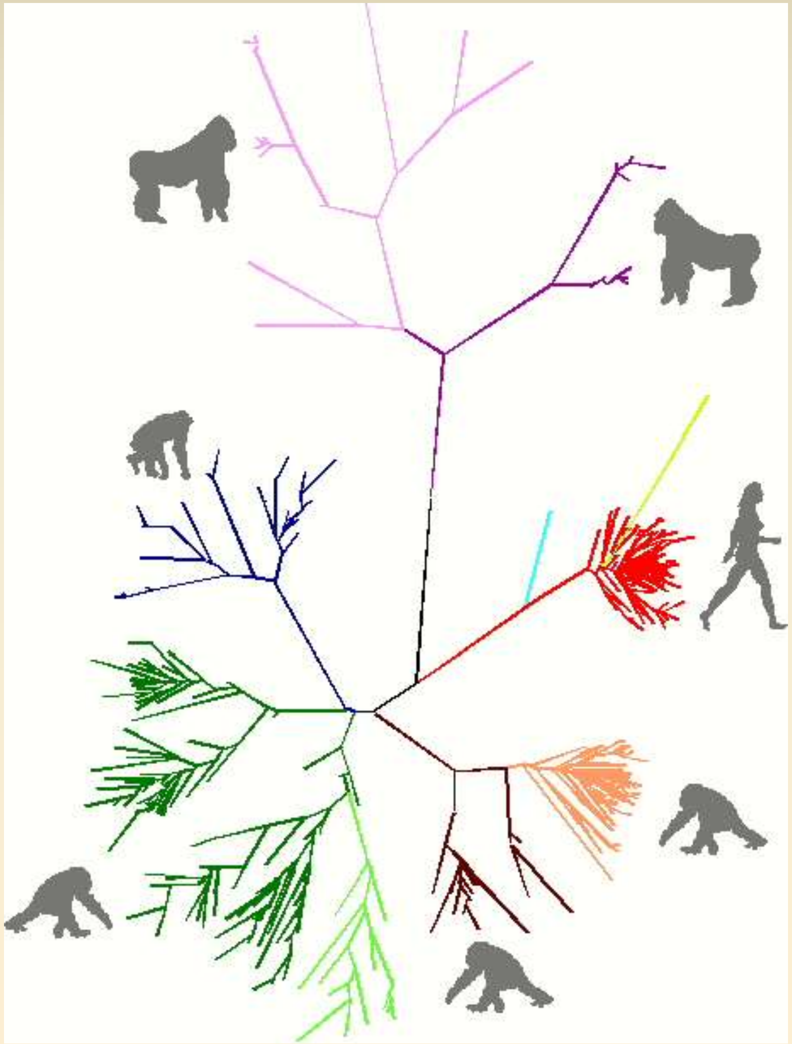
- Human Example: Pingalop atoll is an island in the South Pacific.
- Typhoon in 1780 killed all but 30 people.
- One of survivors was a man who was heterozygous for the recessive genetic disease achromatopsia. This condition caused complete color blindness.
- Today the island has about 2000 people on it, <u>nearly all descended from these 30 survivors. About 10% of the population is homozygous for achromatopsia</u> This implies an allele frequency of about 0.26.

Human Bottleneck

The human population is thought to have gone through a population bottleneck about 100,000 years ago.

There is more genetic variation among chimpanzees living within 30 miles of each other in central Africa than there is in the entire human species.

The tree represents mutational differences in mitochondrial DNA for various members of the Great Apes (including humans).



Toba Supervolcano Bottleneck?

The Toba eruption has been linked to a genetic bottleneck in human evolution about 50,000 years ago, due to the effects of the eruption on the global climate.

Human populations sharply decreased to 3,000–10,000 surviving individuals.

Founder Effect Example

Founder effect example: the Amish are a group descended from 30 Swiss founders who renounced technological progress. Most Amish mate within the group. One of the founders had Ellis-van Crevald syndrome, which causes short stature, extra fingers and toes, and heart defects. Today about 1 in 200 Amish are homozygous for this syndrome, which is very rare in the larger US population.

Note the effect inbreeding has here: the problem comes from this recessive condition becoming homozygous due to the mating of closely related people.

Figure 38-12 AN AMISH CHILD WITH ELLIS–VAN CREVELD SYNDROME.
The child has shortened limbs and six fingers on each hand. All the Amish with this syndrome are descendants of a single couple that helped found the Amish community in Lancaster County, Pennsylvania, in 1744. Because of inbreeding in the isolated community, the recessive trait is now common.

Native Americans provide a possible example. Most North American tribes lack the gene governing type B blood. However, that gene is widespread in Mongolia, their ancestral home. The group that migrated across the Bering Strait may have been small, and lacked this gene.

Native American populations also show a high incidence of albinism. Can this also be attributed to founder effects?

Irish Potato Blight - *Phytophthora*

Potato Blight

- *Phytophthora infestans*
- great Irish famine of 1845-1849
  - 1,000,000 died
- Origin of *P. infestans*
  - Mexico = highest genetic diversity; likely origin
  - Ireland = decreased genetic diversity due to founder effect
  - Decreased genetic differentiation in other regions
    - Europe, North America

Bottleneck Effect: Elephant seal

Reduction of a population's gene pool produced when a few members survive the widespread elimination of a species.

Año Nuevo
State Park

The Effects of Population Size

In large populations, the effect of genetic drift is minimal.

In small populations, genetic drift may be the primary evolutionary force.

The effect of population size is determined by monitoring the frequency of heterozygotes, or the **heterozygosity** of a population over time.

Inbreeding

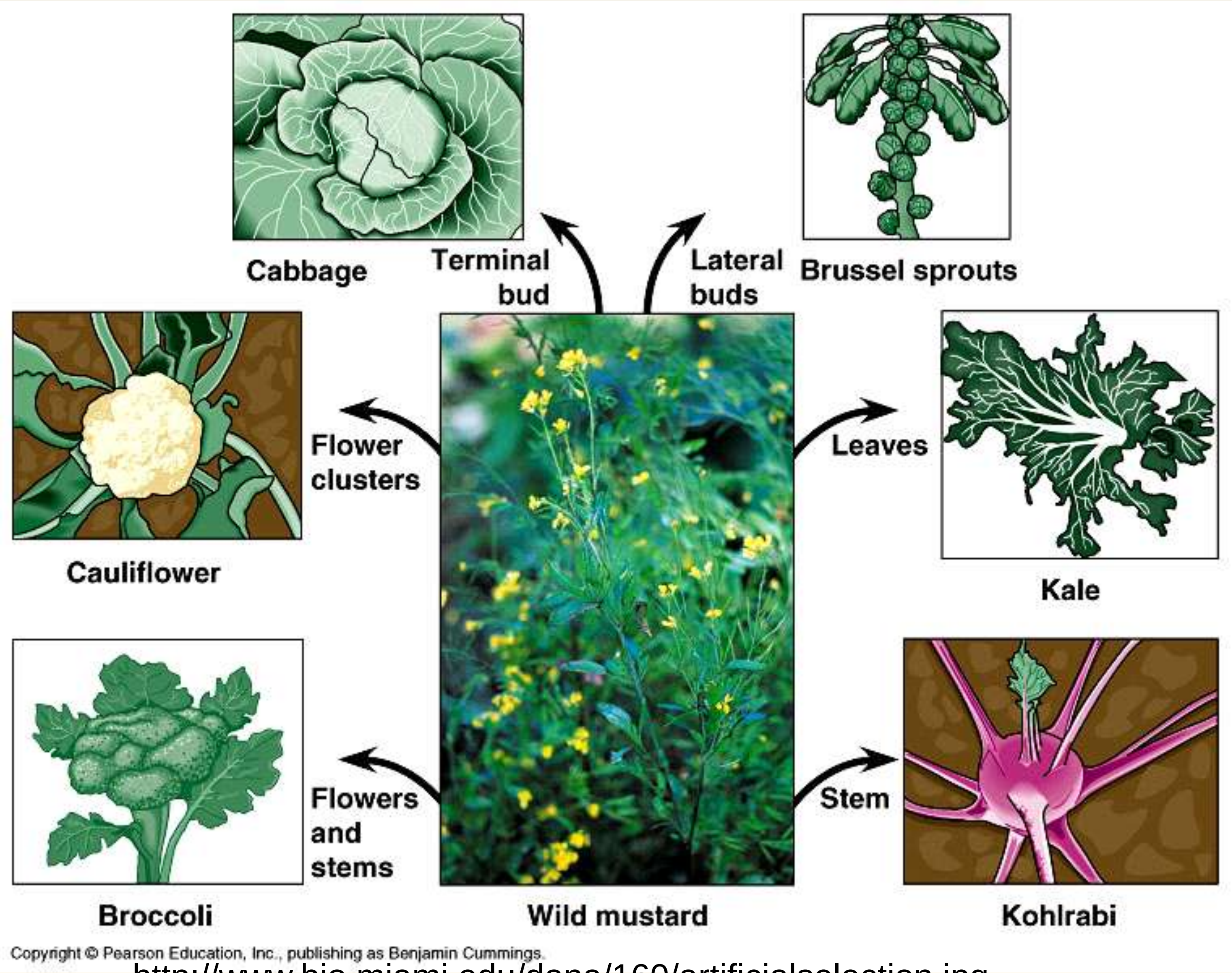
- Mating between relatives or selfing in plants
  - Inevitable in smaller populations
  - Occurs in nature because of proximity of relatives – example: natural stands of tree whose relatives are proximal because of limited seed dispersion
- If there is no natural selection, an inbreeding population will acquire an *increase in the frequency of homozygotes without a change in allele frequency in the population*

SELECTION

Artificial Selection

http://www.gly.uga.edu/railsback/1122/1122Dogs.jpeg

Artificial Selection



http://www.bio.miami.edu/dana/160/artificialselection.jpg

Natural Selection

Some individuals in a population, because of their phenotypic characteristics, produce more offspring that themselves live to reproduce.

- Natural selection can favor, disfavor, or conserve the genetic make-up of a population.

Agents of Natural Selection

Abiotic factors

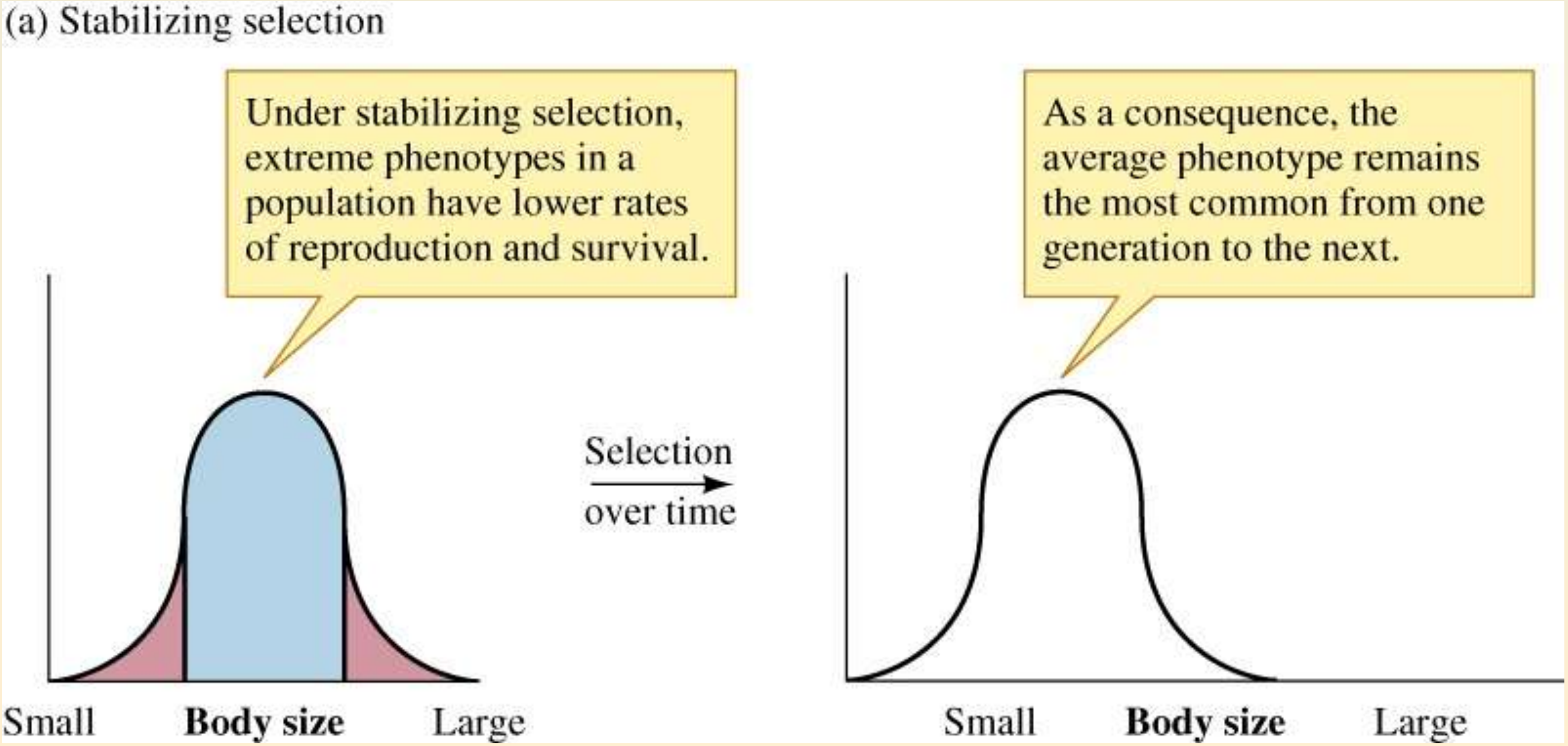
- Climate
- Geology
- Other non-living factors

Biotic factors

- Competition
  - Food, Shelter, Mating
- Interspecies Interactions
  - Predation
- Sexual Selection

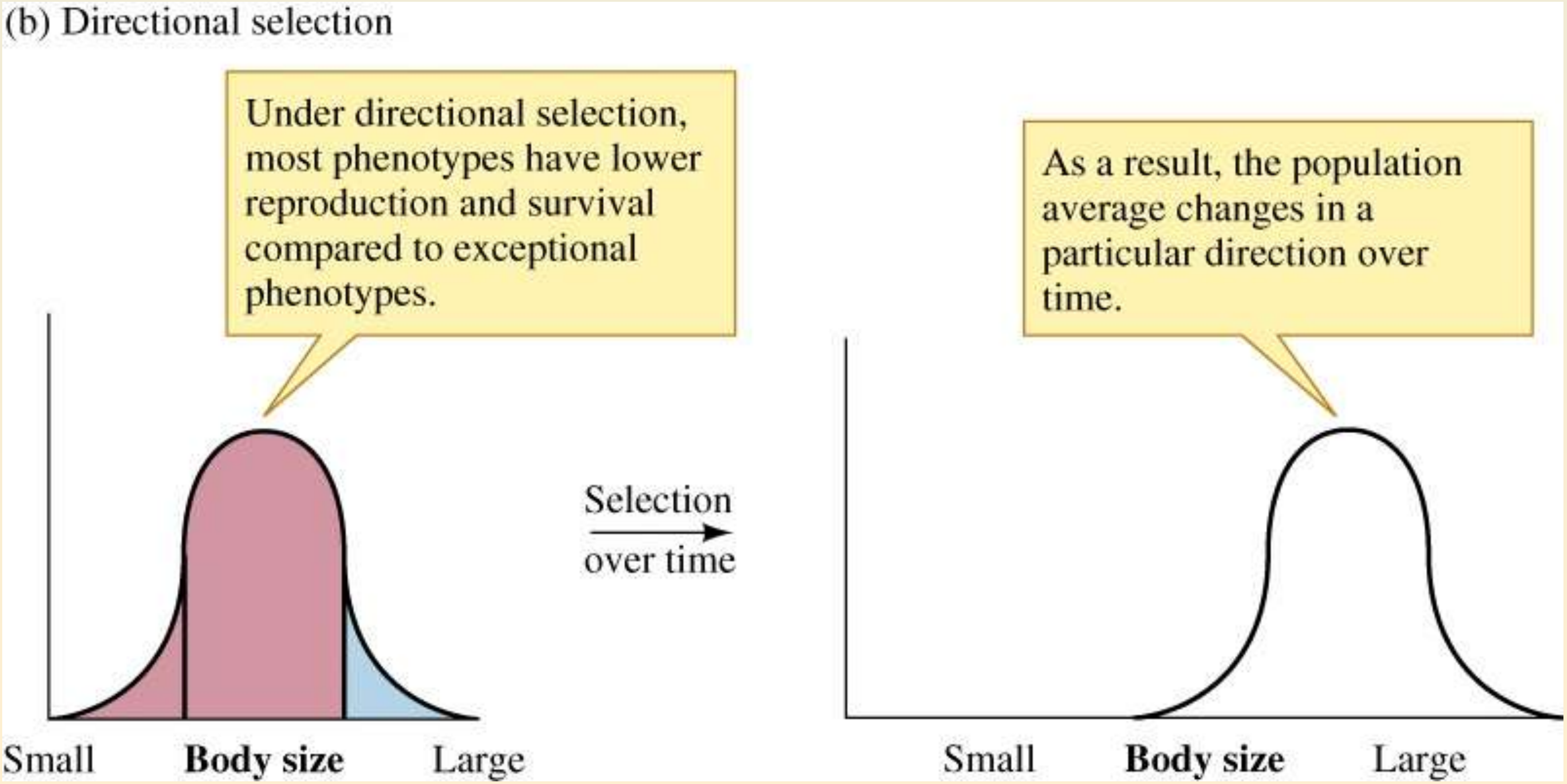
Stabilizing Selection

Stabilizing selection acts to impede changes in a population by <u>acting against extreme phenotypes</u> and <u>favoring average</u> phenotypes.



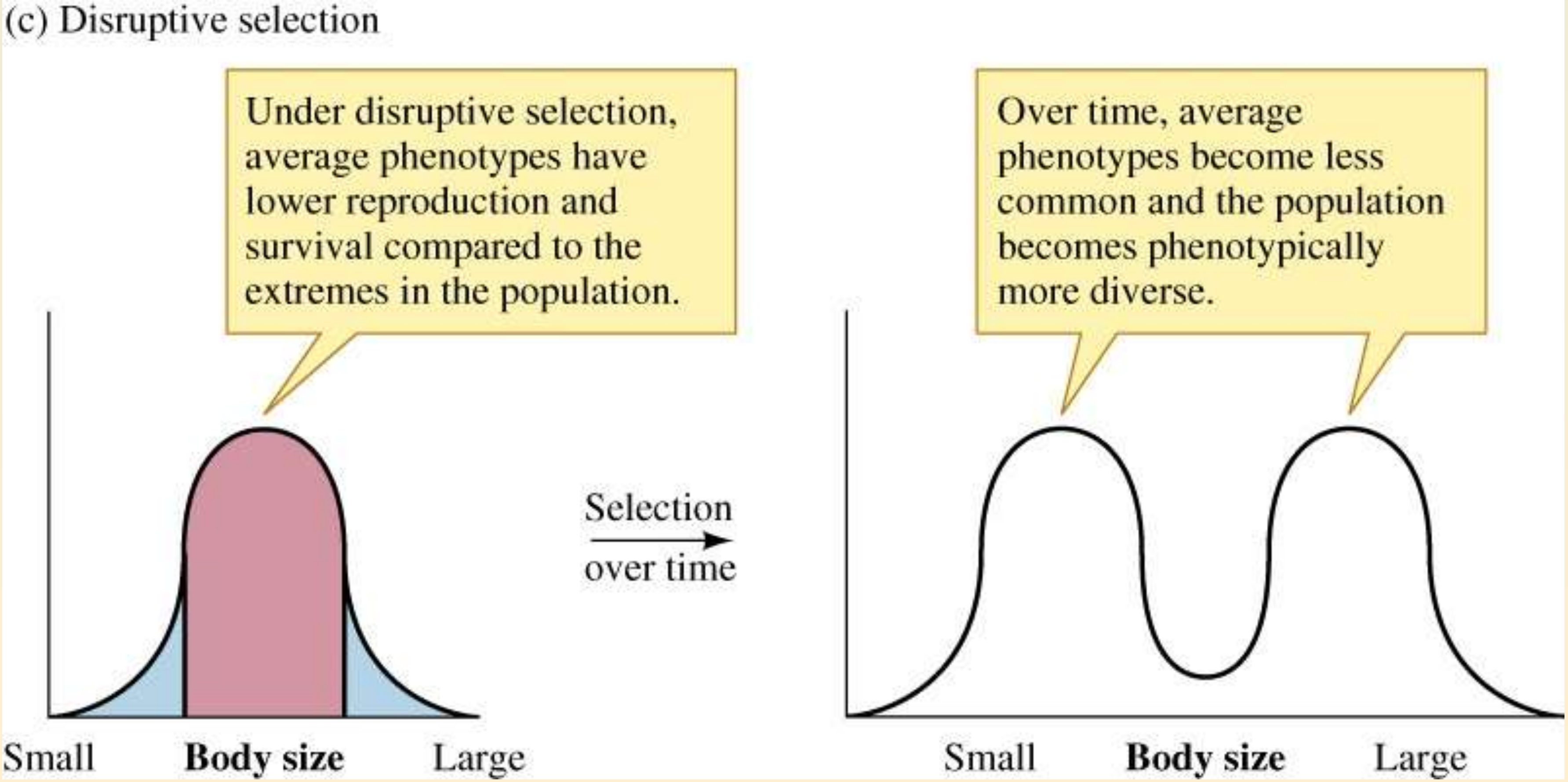
Directional Selection

Directional selection leads to changes in phenotypes by favoring an extreme phenotype over other phenotypes in the population.



Disruptive Selection

Disruptive selection creates bimodal distributions by favoring two or more extreme phenotypes over the average phenotype in a population.

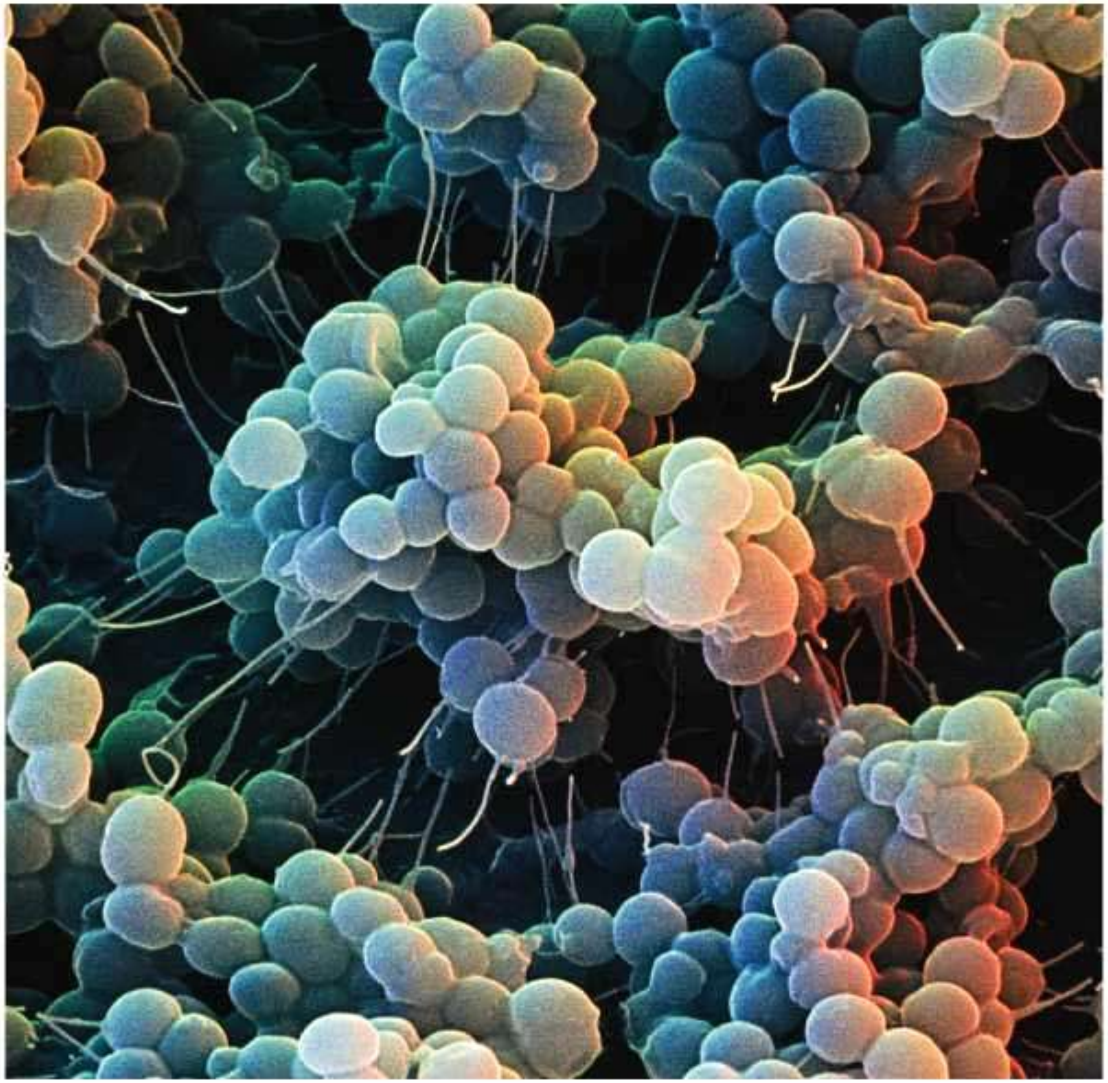


Natural Selection for Pesticide Resistance

http://www.museums.org.za/bio/insects/cockroaches/

http://home-supplies.best-emporium.com/cat-125/Cleaning-Sanitation/Cleaning-Chemicals/Rodenticide-Insecticides

Staphylococcus aureus: antibiotic resistance



Bernard Kettlewell (1907-1979) and Industrial Melanism

Bernard Kettlewell   Light and dark Peppered moths

Kettlewell performed extensive field studies in Britain in the 1950s to test the hypothesis that bird predators were altering the frequencies of the color morphs based on the moths' contrast to their backgrounds, such as tree bark, when they were at rest

Peppered Moth *Biston betularia*

- The Peppered Moth *(Biston bettularia)* occurs in two forms:
  - a typical wildtype white/speckled form
  - a melanic or black form
- The dark form is caused by a dominant mutation that occurs spontaneously
- Peppered moths rest on trees and depend on camouflage for protection

Peppered Moth *Biston betularia*

- In unpolluted areas, trees are covered in lichens and the light form of the moth is hard to see
- In mid 1800's, air pollution in British cities covered trees with coal dust and soot
- In Victorian era cities, the dark form became common and the light form rare

Peppered Moth *Biston betularia*

- In the year 1848, 5% of the population was dark colored moths while 95% was light colored
- In the year 1895, 98% was dark colored while 2% was light colored
- In the year 1995, 19% was dark colored while 81% was light colored

Why Doesn't Natural Selection Eliminate All Genetic Variation in Populations?

- Natural selection does tend to reduce variability in populations by eliminating less fit alleles
- Mechanisms which counteract that elimination to preserve genetic variation include:
  - The diploid condition preserves variation by "hiding" recessive alleles (Bb)
  - **Balanced polymorphisms** (2 or more phenotypes are stable in the population) may result from:
    1. heterozygote advantage: Aa superior to aa and AA
    2. frequency-dependent selection
    3. variation within the environment for a population

Natural selection does not produce perfect organisms

Evolution is limited by historical constraints (*e.g.*, humans have back problems because our ancestors were 4-legged).

Adaptations are compromises. (Humans are athletic due to flexible limbs, which often dislocate or suffer torn ligaments.)

Not all evolution is adaptive. Chance probably plays a huge role in evolution and not all changes are for the best.

Selection edits existing variations. New alleles cannot arise as needed, but most develop from what already is present.

MIGRATION

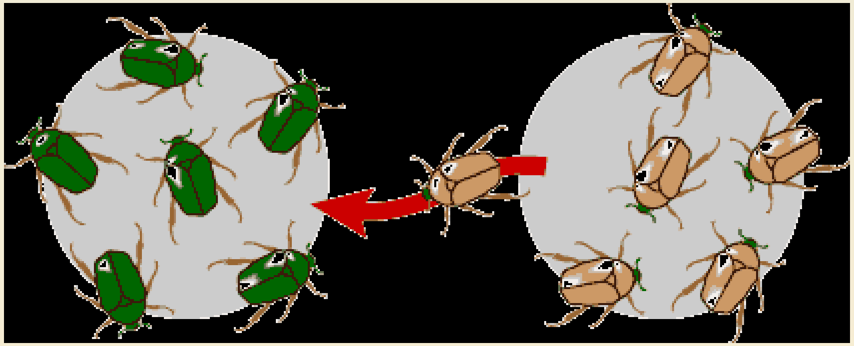
Migration

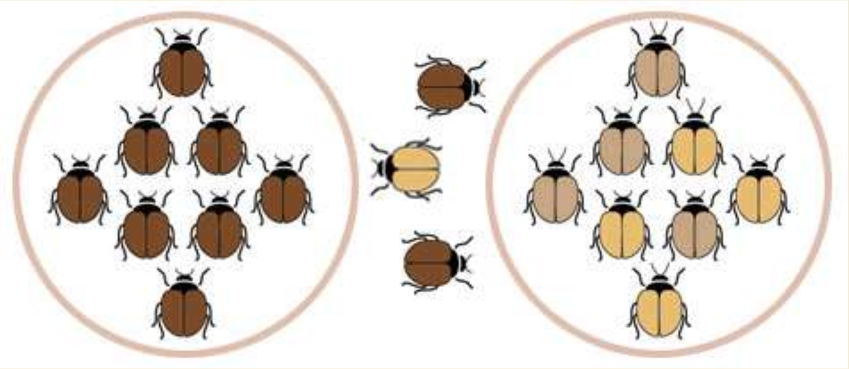
Migration is the movement of individuals in or out of a population. Migration is necessary to keep a species from fragmenting into several different species.

Even as low a level as one individual per generation moving between populations is enough to keep a species unified.

- The physical flow counters the effects of:
  - mutation
  - natural selection
  - genetic drift

Migration – the movement of breeding individuals into or out of isolated populations – results in evolutionary change because alleles move with the individuals. We call this movement **gene flow**.

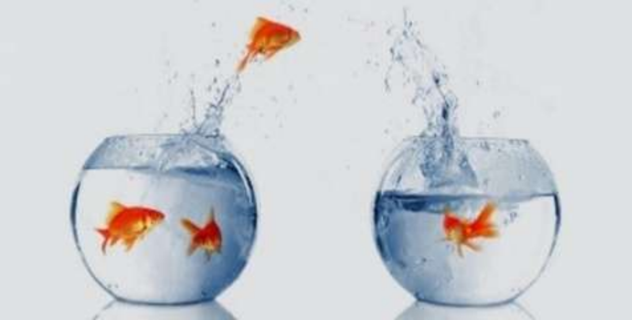




If enough migration occurs, the original isolates, with their inherent limited genetic variability, may fuse and form a new larger population with increased genetic variability.

Gene Flow: A movement of alleles from one population to another

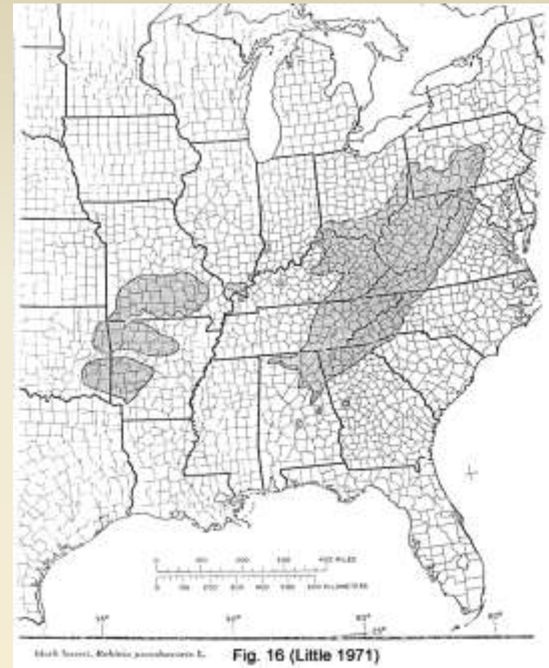
- Migration of individuals or gametes between populations
- Migration can be a powerful agent for evolutionary change
- Migration tends to homogenize allele frequencies between populations
- But migration is adding or removing alleles from the gene pool, so migration is going to change gene frequencies in the populations experiencing immigration or emigration



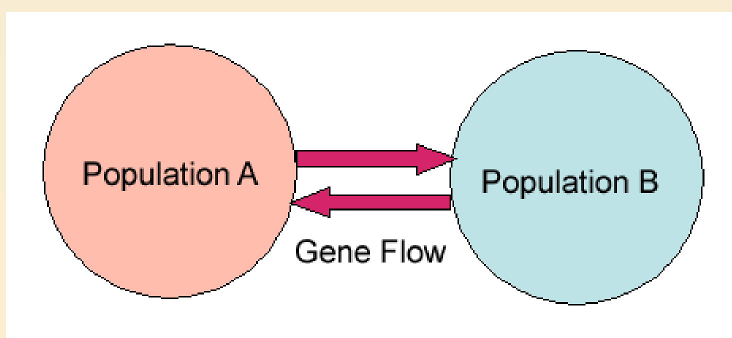
Conservation Genetics and Populations

Geocarpon minimum

Robinia pseudoacacia



How did the distribution get that way?



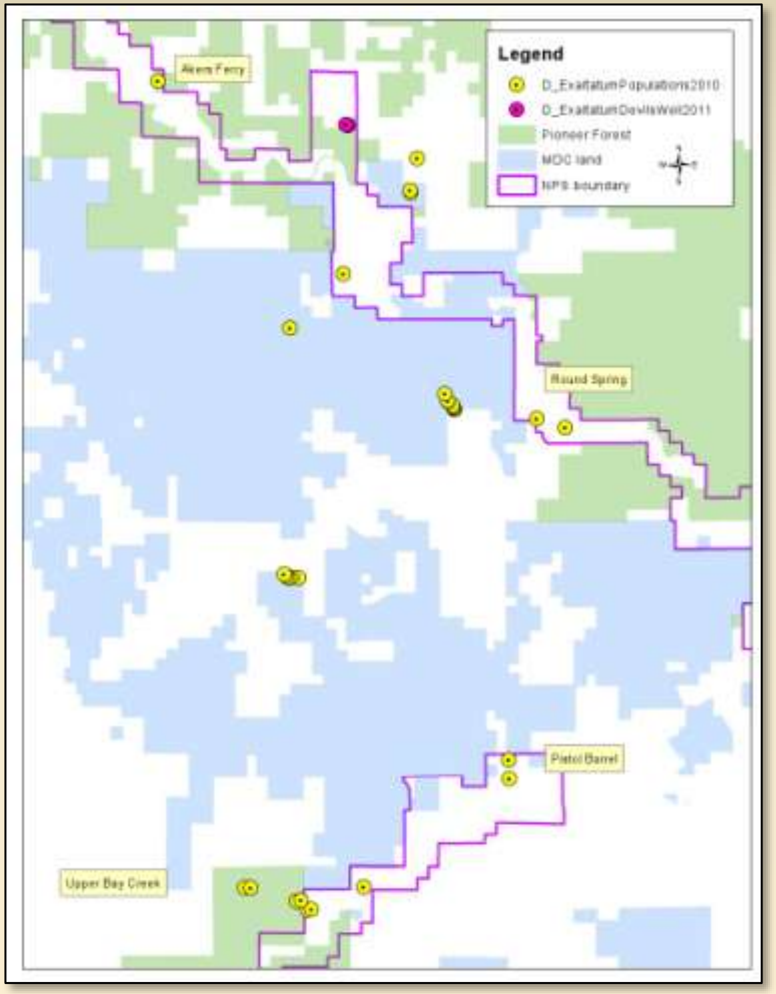
Is gene flow interrupted?

Conservation Genetics and Populations

Delphinium exaltatum

U.S. Distribution

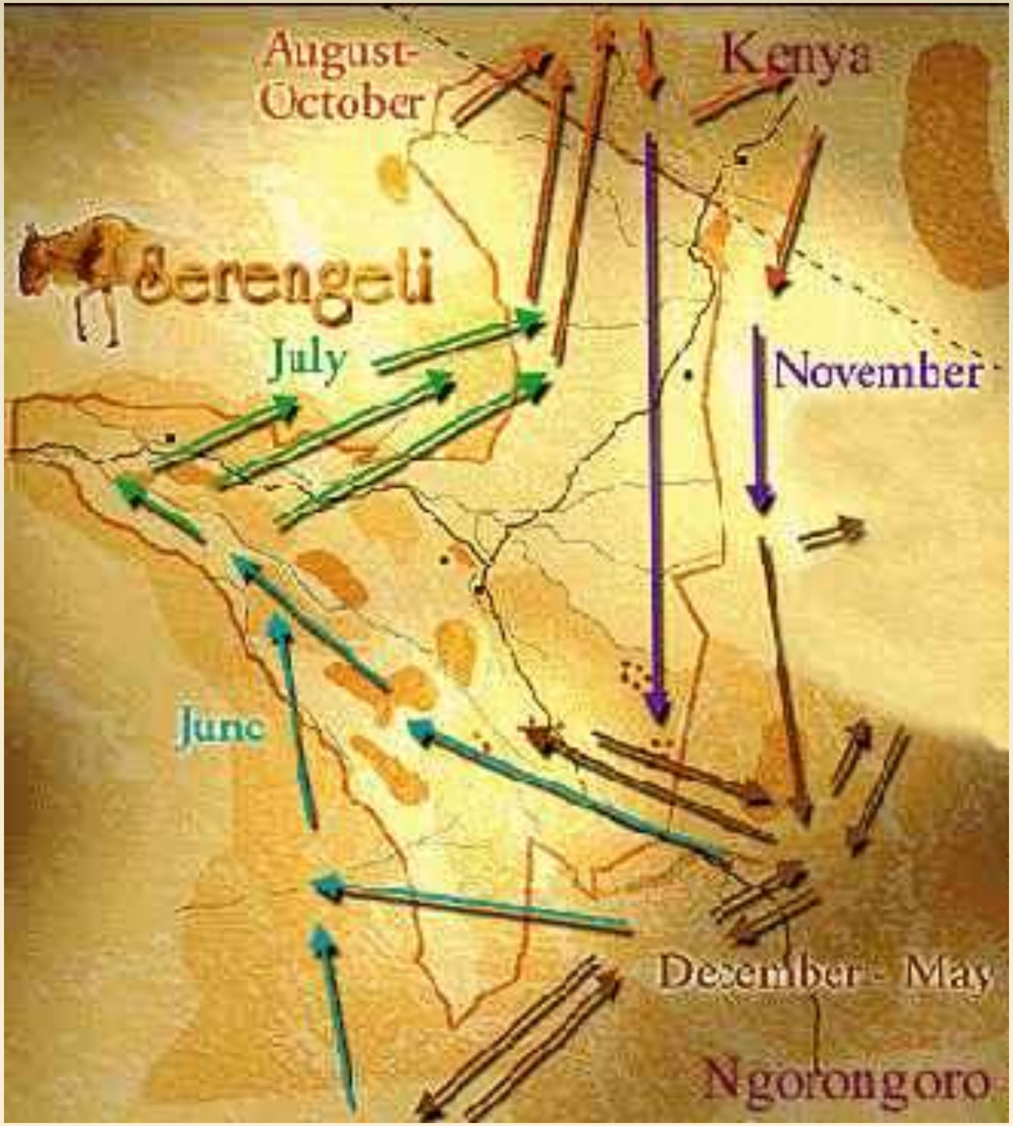
Shannon Co., Missouri



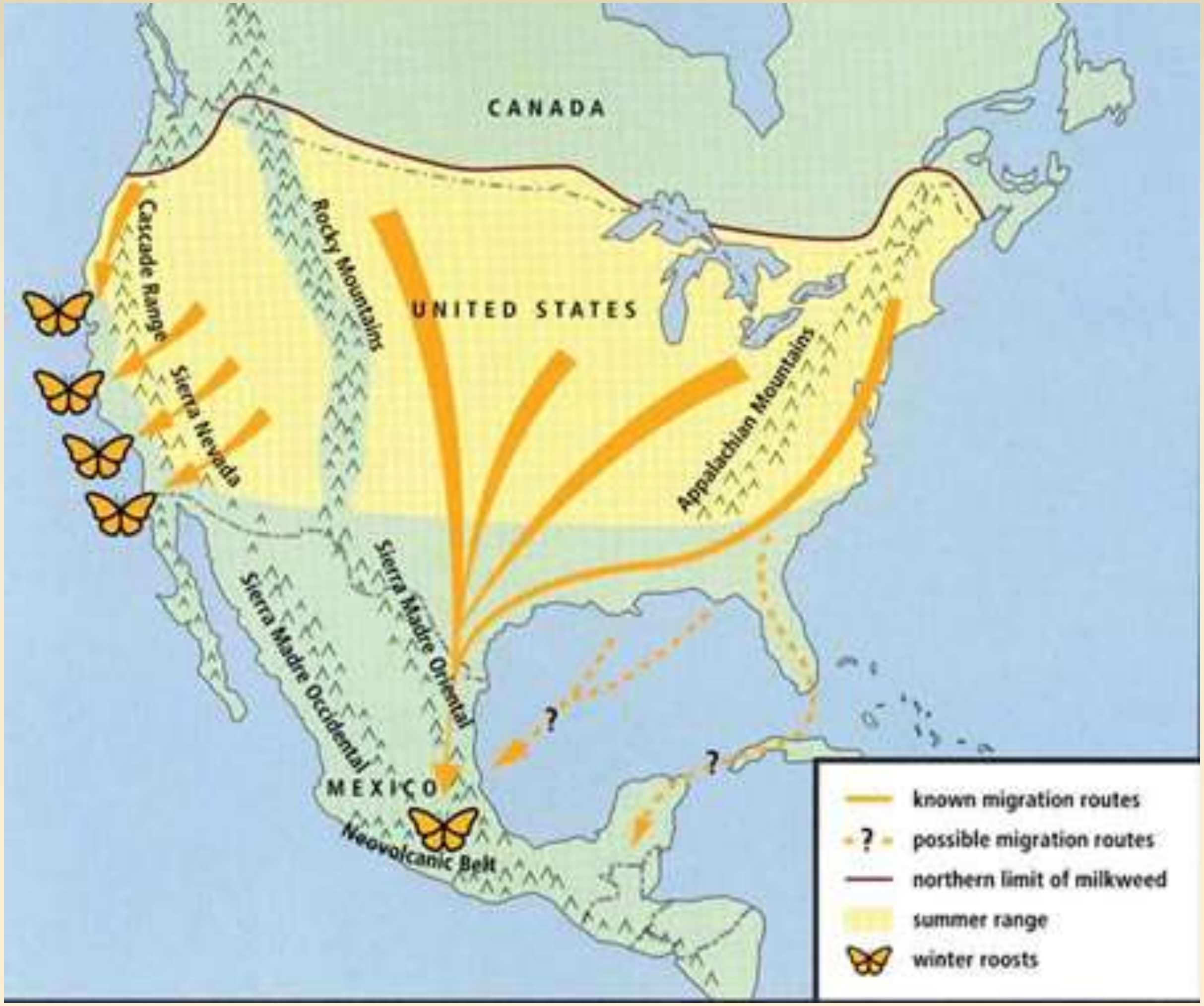
Scale, Populations and Metapopulations

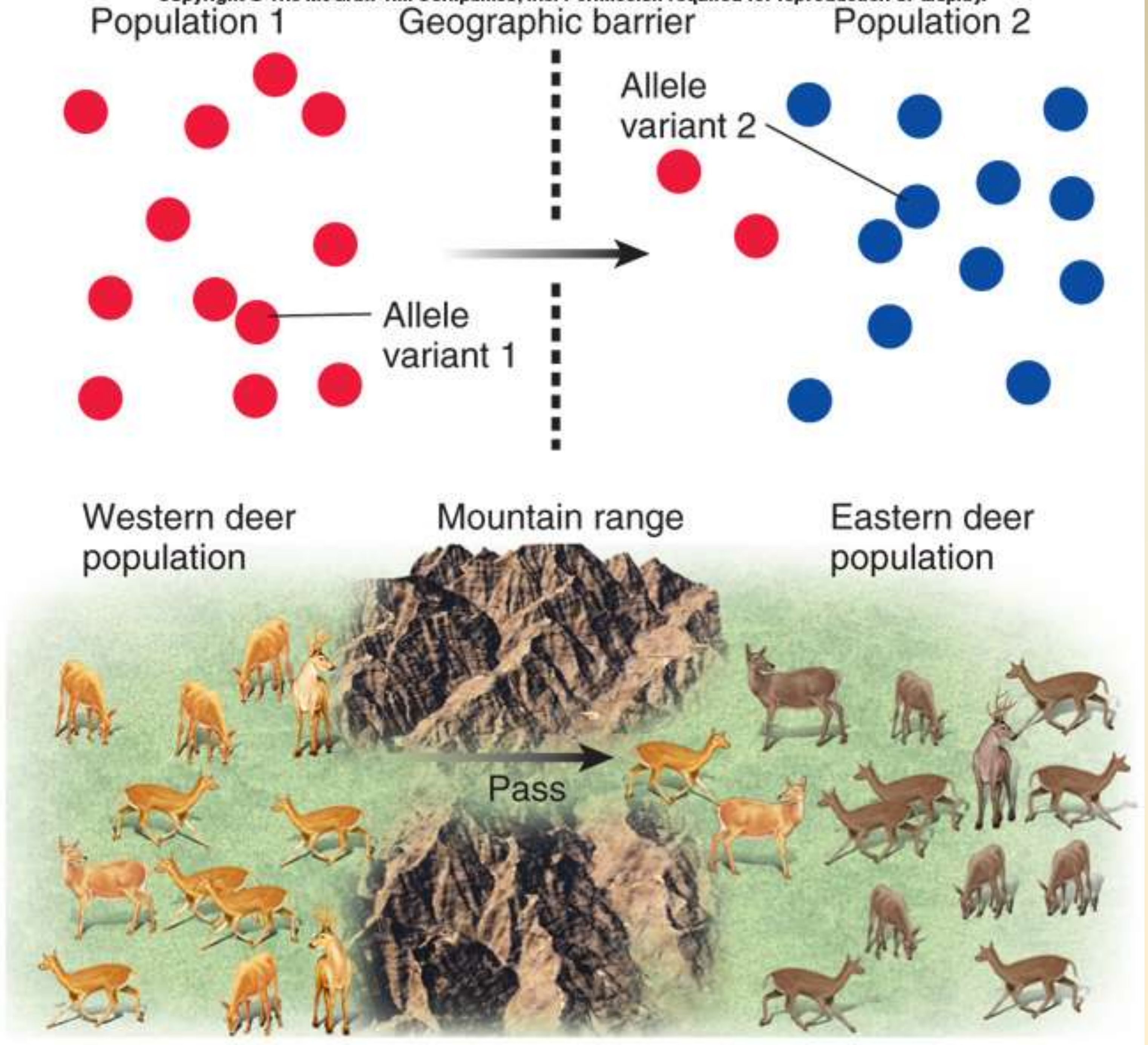
Seasonal Migration

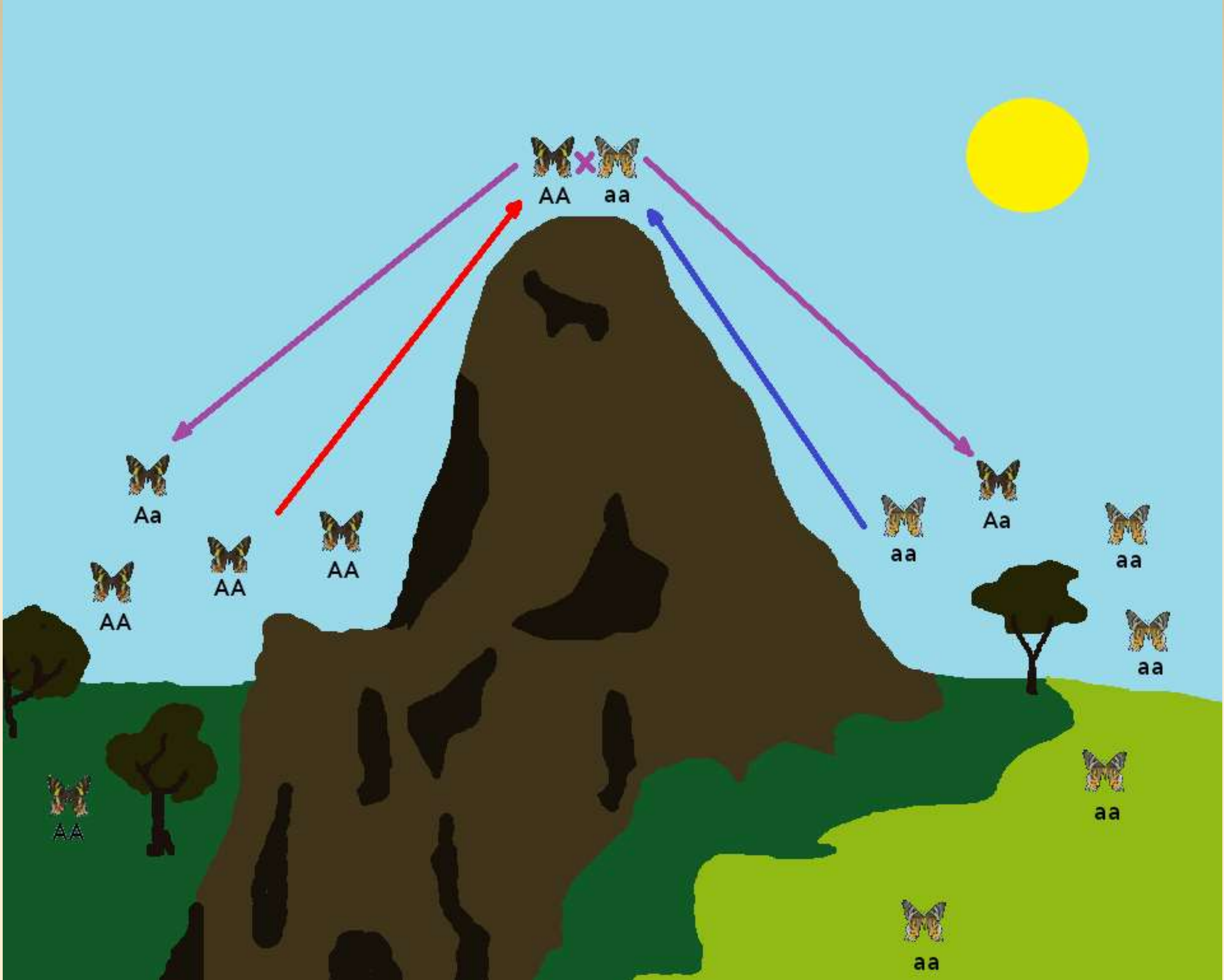
Seasonal Migration



Seasonal Migration




Population 1
Geographic barrier
Population 2
Allele variant 2
Allele variant 1
Western deer population
Mountain range
Eastern deer population
Pass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What is dispersal?

Simply, the movement of organisms away from their birthplace.

Often, confined to a particular life history stage.

Don’t confuse with *dispersion*, which refers to the position of individual organisms with respect to others in the population.

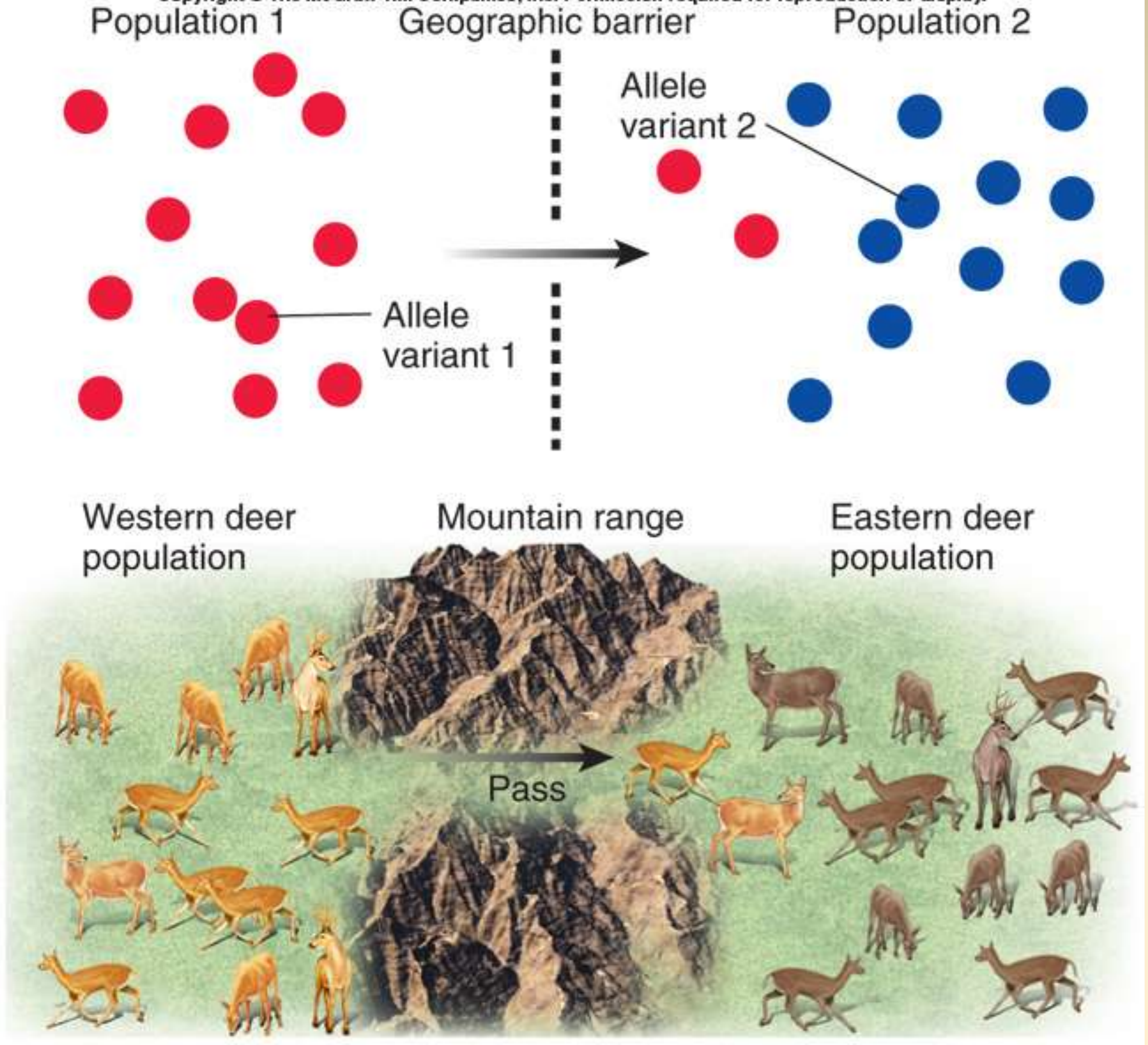
Dispersal is an ecological process that plays an adaptive role in the life history of the organism involved.

In other words, the fitness of the organism is increased in some way through the process of dispersal.

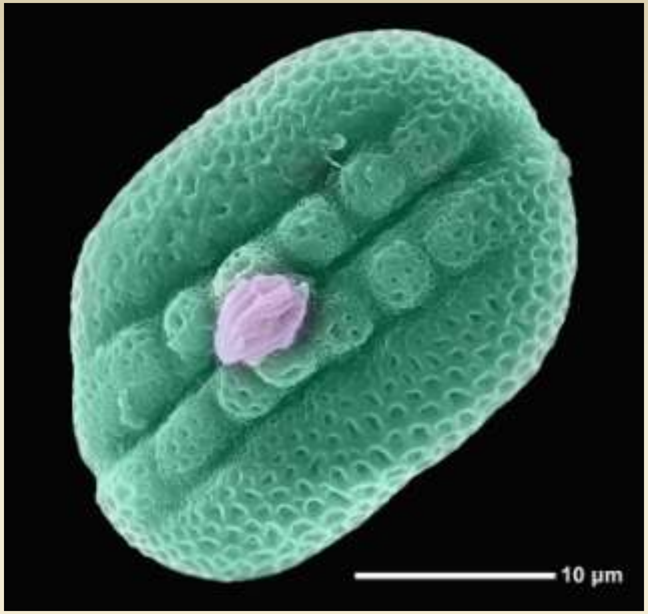
Why?

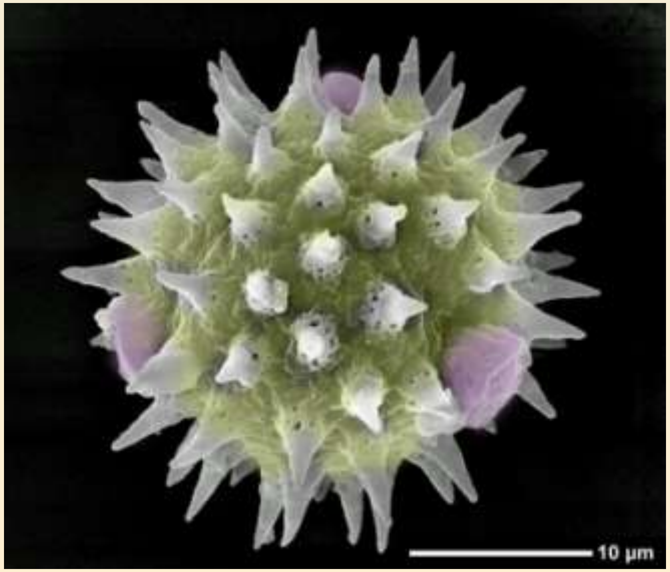
There's always a trade-off. Dispersing individuals probably face reduced interspecific competition, but there's always the chance of finding a less suitable environment.

Look at it this way. The "parental" environment was obviously good enough to allow the parent organisms to reproduce. Leaving that environment is risky, but it must be worth the risk.

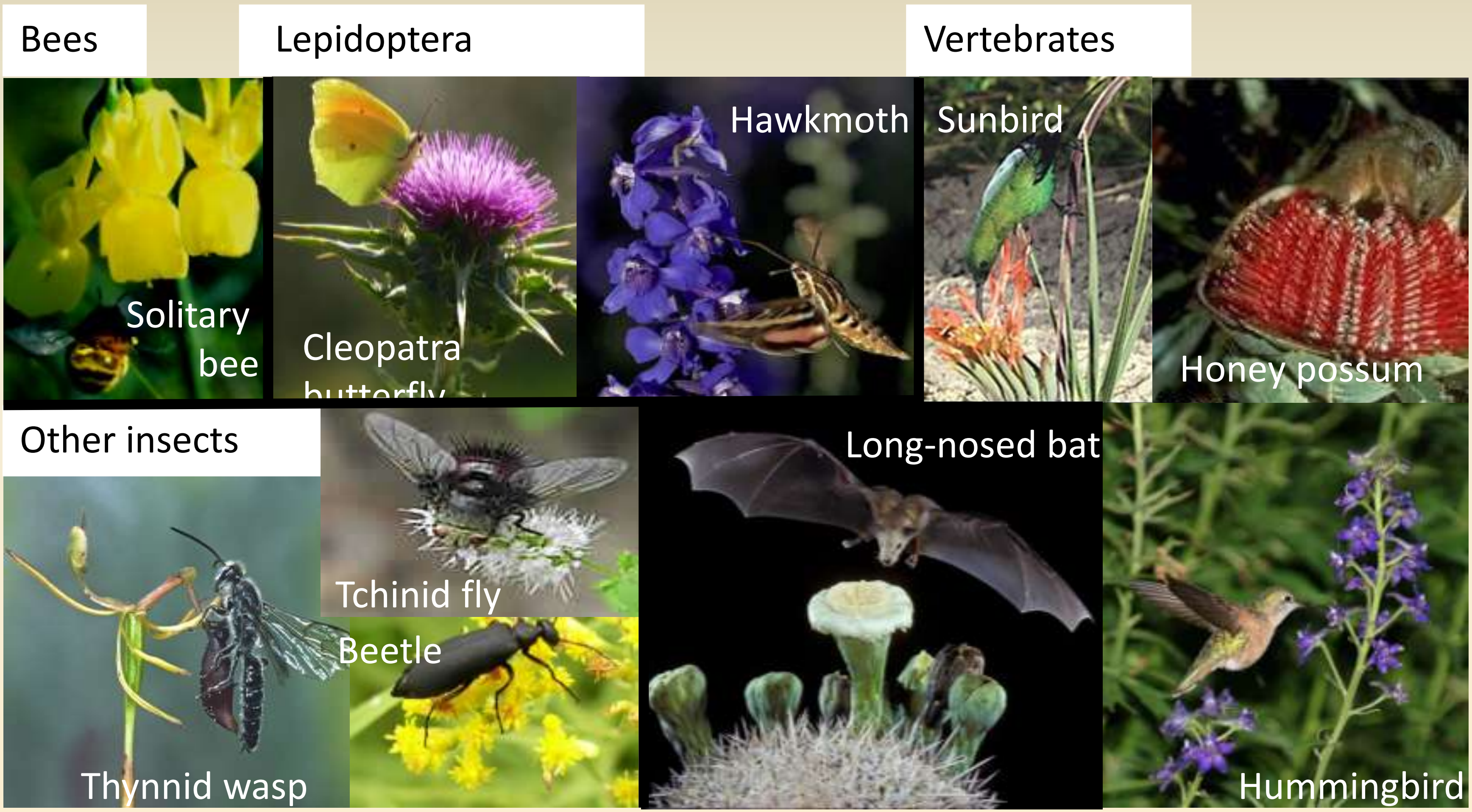

Population 1
Geographic barrier
Population 2
Allele variant 2
Allele variant 1
Western deer population
Mountain range
Eastern deer population
Pass

Dispersal - Geneflow





Pollen dispersal agents: biotic



Seed dispersal agents

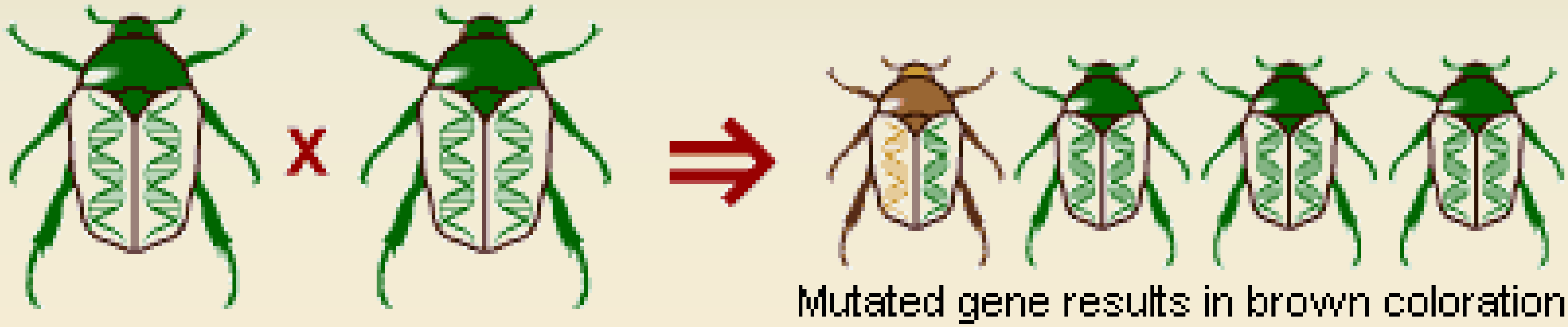
Migration

Migration can be thought of as combining two populations with different allele frequencies and different numbers together into a single population.

After one generation of random mating, the combined population will once again be in H-W equilibrium.

MUTATION

Mutation

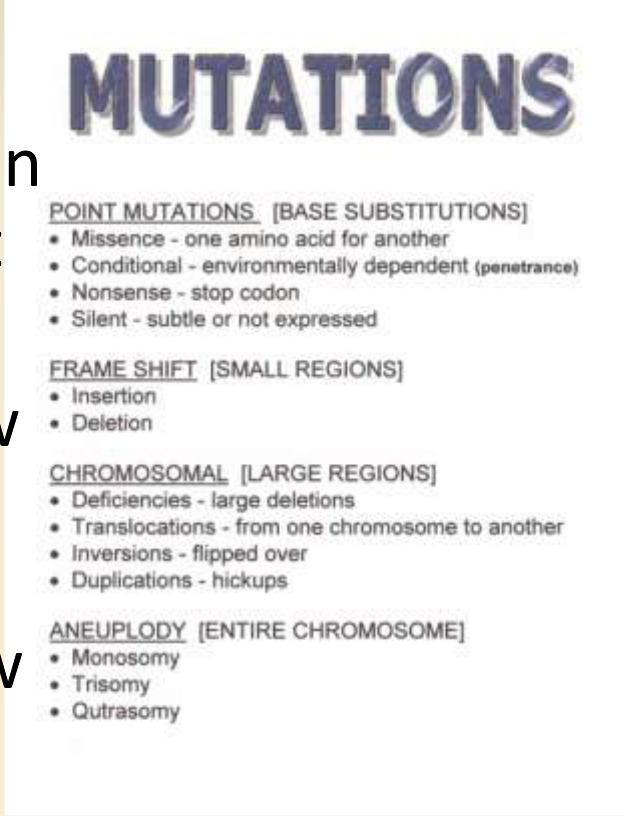


Types of Mutations

- Point mutation
  - Synonymous – no change in a.a.
  - Nonsynonymous – change a.a.
- Frame-shift mutation
- Stop mutation
- Chromosome Fusion
- Trinucleotide Repeats

Mutation: Changes in a cell's DNA

- Mutation is the ultimate source of genetic variation
- Since a new mutation transforms an allele into a different allele, it must also change allele frequencies
- Mutation rates are generally so low they have little effect on Hardy-Weinberg proportions of common alleles in the short term, over a few hundred generations



Mutations are rare (1/100,000 gametes)

Mutations can be:

- Helpful
  - Depends on environment
- Neutral
- Harmful
  - Lethal mutation

Resistance to DDT in Common Houseflies

Rapid genetic changes in many insect populations exposed to pesticides such as dieldrin and DDT

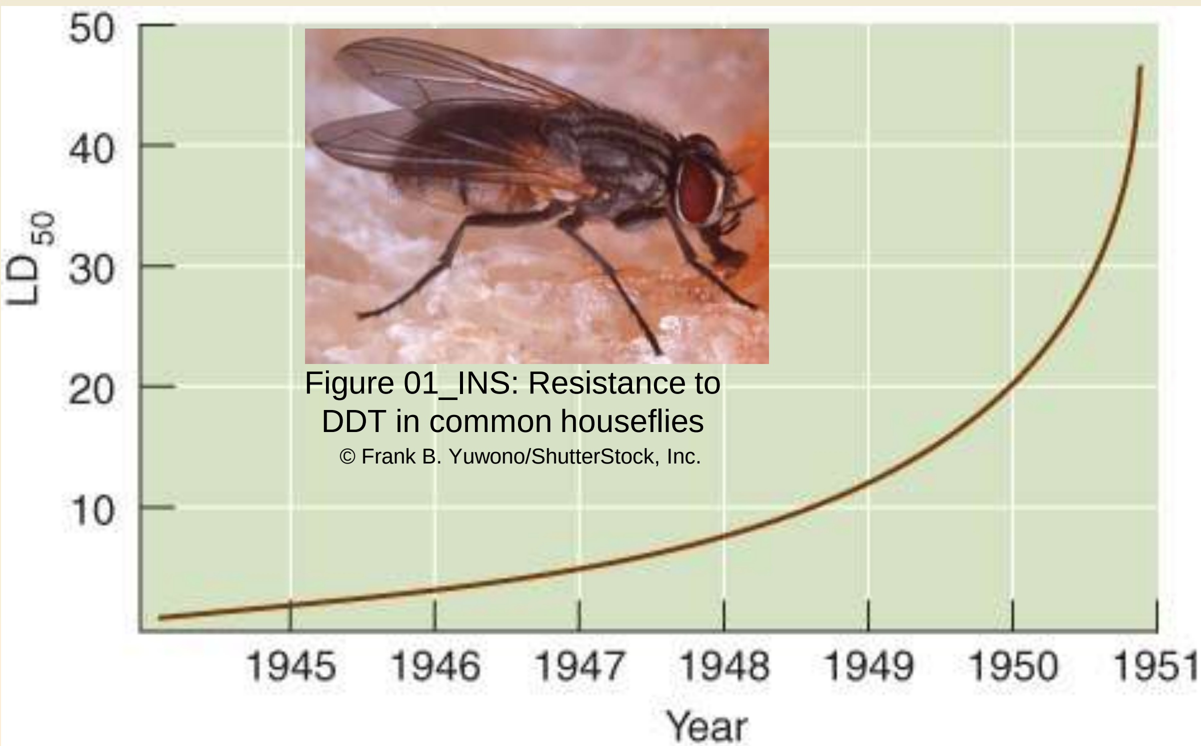


Figure 01_INS: Resistance to DDT in common houseflies

- Both dieldrin and DDT are neurotoxins; DDT opens sodium channels and both DDT and dieldrin stimulate Ach synthesis and release

- Resistance develops when mutant neuron membrane proteins arise or the receptor numbers change

Adapted from Strickberger, M. W. Genetics, Third edition. Macmillan, 1985; based on data from Decker, G. E., and W. N. Bruce, Amer. J. Trop. Med. Hygiene 1 (1952): 395-403.

Mutation

Mutation is unavoidable. It happens as a result of radiation in the environment: cosmic rays, radioactive elements in rocks and soil, etc., as well as mutagenic chemical compounds, both natural and artificially made, and just as a chance event inherent in the process of DNA replication.

However, the rate of mutation is generally quite low: for any given gene, about 1 copy in 10^4 - 10^6 is a new mutation.

Mutations provide the necessary raw material for evolutionary change, but by themselves new mutations do not have a measurable effect on allele or genotype frequencies.

Nonrandom Mating

Random Mating is required for the Hardy-Weinberg Equilibrium

The members of the population mate with each other without regard to their phenotypes and genotypes

The members of the population are (relatively) equally likely to mate with any other member of the population of the opposite sex, i.e., have relatively equal access to all members of the population

<u>Examples</u>

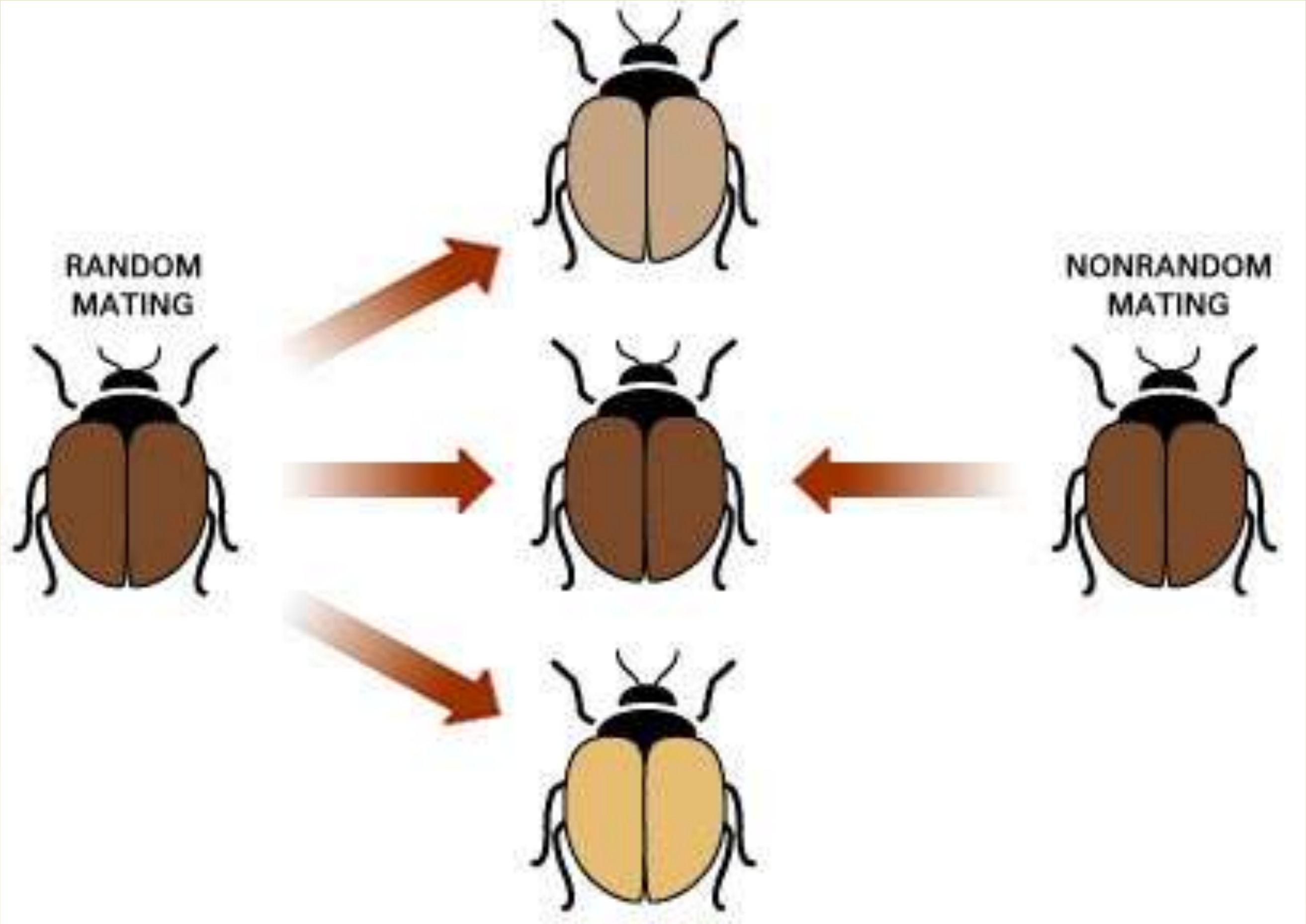
Humans who mate without regard to ABO and Rh blood types

This example shows that a particular gene can meet the H-W equilibrium, even though the species does not

Wind-pollinated plant species and many aquatic species who release their eggs and sperm into the water

Animals who are members of large mobile schools, herds or flocks

Nonrandom Mating: mating between specific genotypes shifts genotype frequencies



Nonrandom Mating: mating between specific genotypes shifts genotype frequencies

Do humans mate randomly? How do people pick a mate?

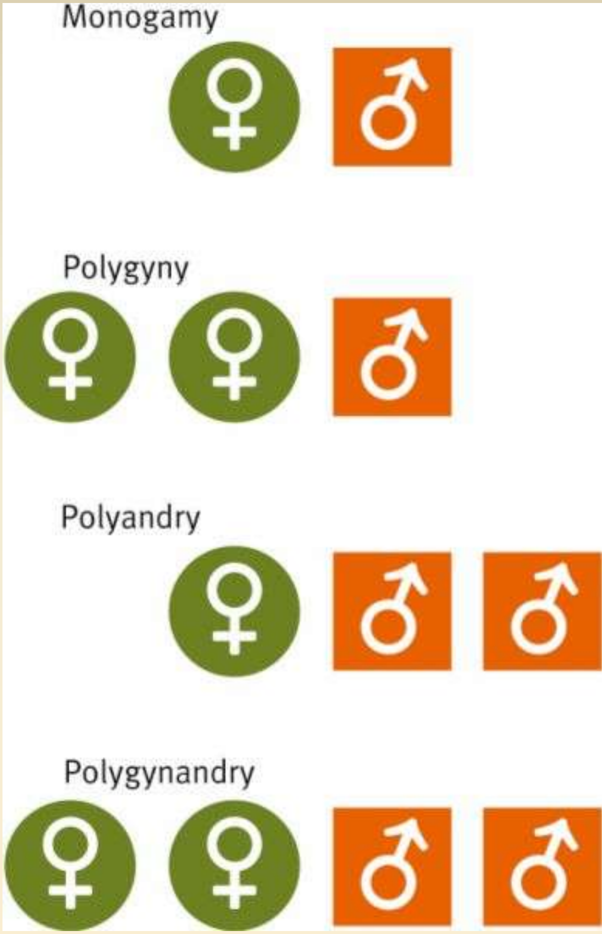
Ladies, would you prefer to mate and produce offspring with one of these males over another? If so, you are practicing non-random mating based on phenotypic characteristics





Classification of mating systems

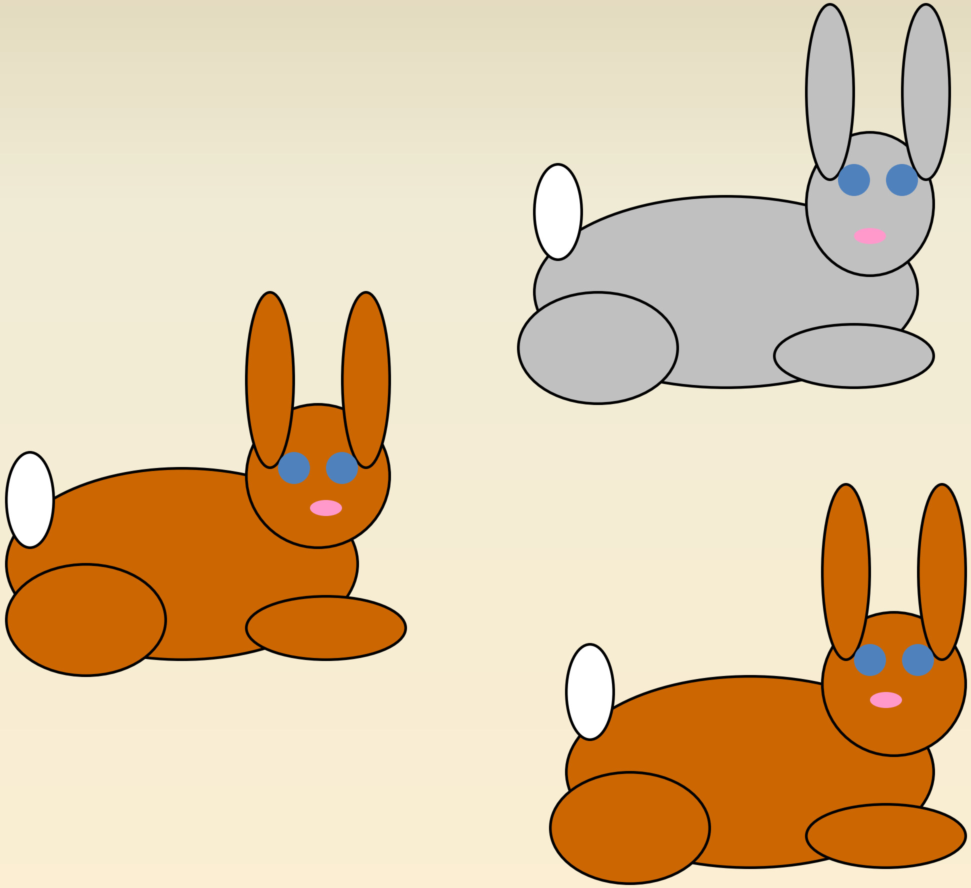
- Monogamy, polygamy, polyandry (Darwin 1871)
- Monogamy, resource defense polygyny, harem defense polygyny, explosive mating assemblage, leks, female access polyandry, etc. (Emlen & Oring 1977)
- Promiscuous
- Self pollination
- Asexual reproduction
  - Apomixis: parthenogenesis in animals and apogamy in plants



Non-random mating: 3 types

- Assortive – shifts genotype frequency
  - Organisms choose a mate with the same genotype as themselves.
  - Organisms choose a mate with a different genotype from themselves.
- Self-fertilization – shifts genotype frequency
  - Organism mates with itself
- Sexual selection –shifts allele frequency
  - Some genotypes mate more successfully than others

Assortive – shifts genotype frequency



Self-fertilization – shifts genotype frequency

Sexual selection –shifts allele frequency

http://subjunctive.net/photoblog/2003/peacock-wooing-peahen.jpg

Sexual selection leads to differences between sexes

Sexual dimorphism is the difference in appearance between males and females of a species.

Intrasexual selection is the direct competition between members of the same sex for mates of the opposite sex.

- This gives rise to males most often having secondary sexual equipment such as antlers that are used in competing for females.

Intersexual selection (mate choice), one sex is choosy when selecting a mate of the opposite sex.

-This gives rise to often amazingly sophisticated secondary sexual characteristics; *e.g.*, peacock feathers.


Benjamin
Cummings


Benjamin
Cummings

The fullest meaning of "random mating" implies that any gamete has an equal probability of fertilizing any other gamete, including itself. In a sexual population, this is impossible because male gametes can only fertilize female gametes.

More or less random mating in a sexual population is achieved in some species of sea urchin, which gather in one place and squirt all of their gametes, male and female, out into the open sea. The gametes then find each other and fuse together to become zygotes.

In animal species, mate selection is far more common than random fertilization. A very general rule is "assortative mating", that like tends to mate with like: tall people with tall people, short people with short people, etc. This rule is true for externally detectable phenotypes such as appearance, but invisible traits like blood groups are usually close to H-W equilibrium in the population.

Assortative mating is most easily analyzed as a tendency for inbreeding. You are more like your relatives than you are to random strangers. Thus you are somewhat more likely to mate with a distant relative than would be expected by chance alone.

And then, from across the room,
their eyes met.

Founder Effects

- Establishment of a population by a few individuals can profoundly affect genetic variation
  - Conseq[illegible] of [illegible] eff[illegible]ts
    - Fewer [illegible]eles
    - Fixed [illegible]
    - Modified allele frequencies compared to source pop
      - Perhaps due to “new environment”

End