Big Idea 1: The process of evolution drives the diversity and unity of life.

Enduring understanding 1.A: Change in the genetic makeup of a population over time is evolution.

# Preface

# Why do species go extinct?

• Short Answer- They were unable to meet life's challenges!

#### Recall Living Organisms Must

- Obtain Nutrients
- Obtain & Conserve Water
- Eliminate Wastes
- Exchange Gases

- Sense & Respond to their environment
- Thermoregulate
- Defend themselves
- Reproduce

# Why do species go extinct?

 Long Answer- The environment changes, which in turn effects the species ability to meet one or more of life's challenges and as a group the species is unable to adapt in time to the new conditions.

#### **Environmental Changes & New Conditions**

- Changing Climates
- Habitat Loss
- Being Out-Competed (by better adapted organisms)
- Over-Exploitation & Hunting

# How do species continue to meet life's challenges?

 The population as a whole adapts and changes over time with the inevitable changing conditions...They Evolve! Essential knowledge 1.A.1: Natural selection is a major mechanism of evolution.

a. According to Darwin's theory of natural selection, competition for limited resources results in differential survival. Individuals with more favorable phenotypes are more likely to survive and produce more offspring, thus passing traits to subsequent generations.

Variation exists among individuals in a

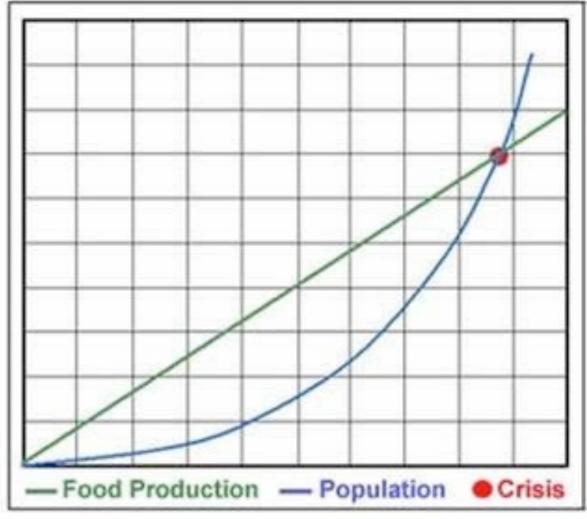
population!



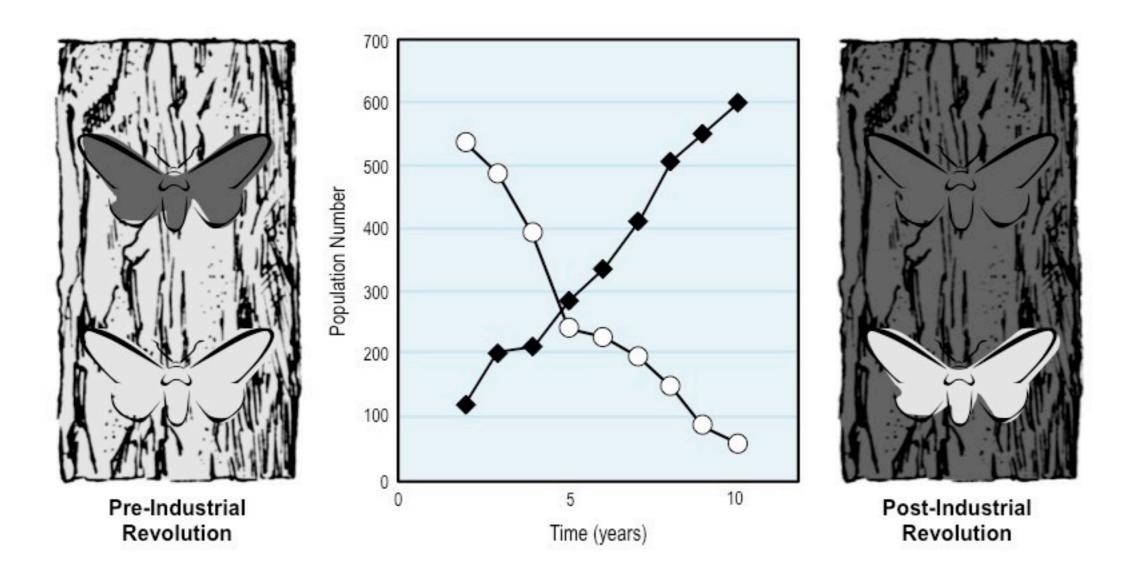


 Populations produce more offspring than the environment can support, thus some will fail to survive and/or reproduce!





 Individuals with inherited traits that give them a better chance of surviving and reproducing in a particular environment tend to leave more offspring!



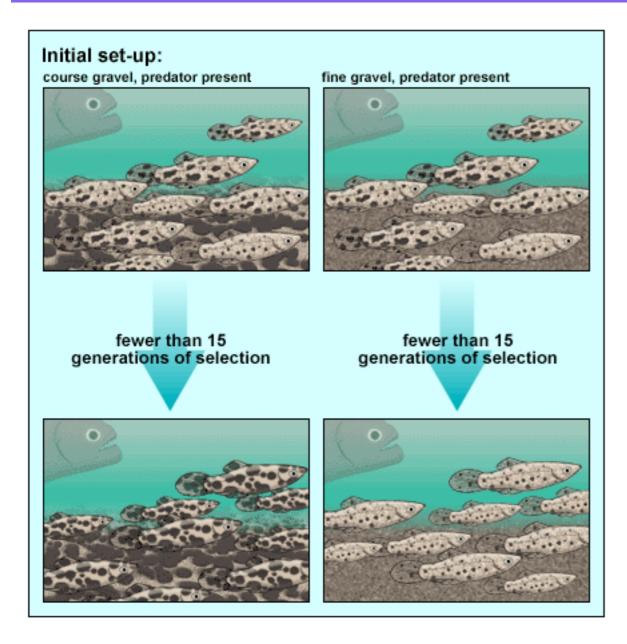
 The unequal ability of individuals to survive and reproduce will lead to accumulation of favorable traits and less favorable traits will diminish in the population over \*time!

\*Darwin noted that if artificial selection could produce dramatic changes in a relatively short time frame then given enough time nature could fashion the same dramatic changes

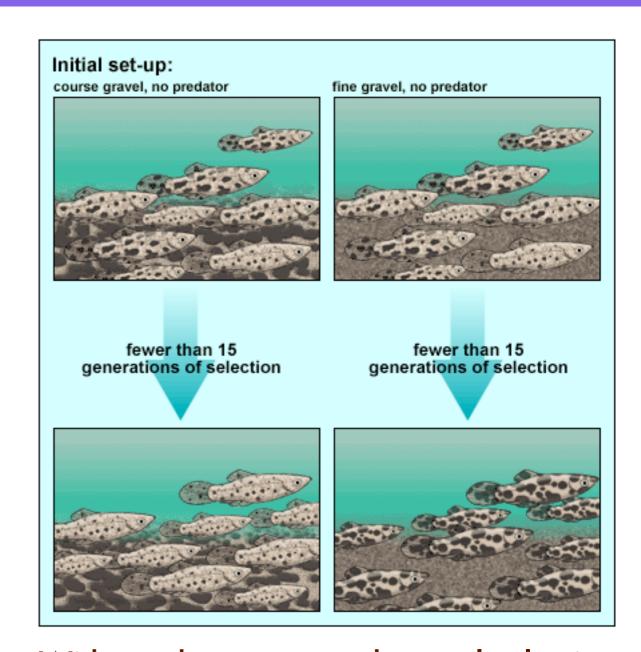
#### Darwin's Mechanism

- NATURAL SELECTIONdifferential reproductive success!
- Beneficial traits are subject to the environment, a beneficial trait for one may be deleterious to another.
  - (should the environment change so to might the beneficial traits)
- Natural selections can only amplify or diminish the traits that differ in a population.
   (no variation = no selection)
- Populations evolve not individuals.

#### Natural Selection in Action



The predator serves as the strongest selective pressure, resulting in a population that blends with its surroundings to avoid being eaten



With predator removed sexual selection serves as the strongest pressure, resulting in a population with males standing out against their surroundings

# What exactly is evolving?

- The POPULATION as a whole adapts and changes over time with the inevitable changing conditions...POPULATIONS Evolve!
- The POPULATION is the smallest unit of evolution.
  - The Individual can change through growth and development but only populations evolve.
  - Traits effect the survival and reproductive success of individuals but the effects of natural selection are only seen in the larger group over time.

Essential knowledge 1.A.1: Natural selection is a major mechanism of evolution.

b. Evolutionary fitness is measured by reproductive success.

- <u>Fitness</u>- contribution an individual makes to the gene pool of the next generation relative to others.
  - <u>Relative Fitness</u>- contribution that a genotype makes to the gene pool of the next generation for same locus relative to others.
  - Be careful natural selection acts on phenotypes/traits not genotypes.
    - The organism as a "whole" is selected.
  - Thus the fitness of any one allele is dependent on the entire genetic make up of the organism and the environment at that point in time.
    - This can sometimes result in strange consequences...

• This type fitness is not all about "physical fitness".

6 Kids





**Greater Evolution Fitness** 



This fitness is all about reproductive success and the traits that make one fit can be very subtle.

This type fitness is not all about pure survival.

3 Kids, dies at age 35

0 Kids, dies at age 98



Greater Evolution Fitness



A long life span does not always equate to evolutionary fitness.

- Lastly sometimes alleles that do have no value or even slightly maladaptive can be perpetuated if the organism's overall fitness is high.
  - This explains have old useless DNA persists in a genome over time.
- It can go the other way as well... sometimes alleles that do have value are not perpetuated if the organism's overall fitness is low or by some accident dies suddenly.

Essential knowledge 1.A.1: Natural selection is a major mechanism of evolution.

C. Genetic variation and mutation play roles in natural selection. A diverse gene pool is important for the survival of a species in a changing environment.

# Evolution-Ingredients

- For evolution to occur we need at least two very important ingredients: time & variation.
- TIME
  - (in general) more time = greater changes
  - Darwin's mechanism was slow and gradual, it required vast tracks of time
  - Today we think "some" change might happen rapidly in bursts

#### VARIATION

• remember the force behind evolution is natural selection, the very word itself "selection " implies variation otherwise there is nothing to "select"

#### **Evolution-Variation**

- Individual variation occurs in all species.
  - some variation we can see- hair color
  - some variation we can see- blood groups
- Individual variation reflect genetic variation.
  - different phenotypes result from different genotypes

When certain traits are favored over others it is in fact the genes, the "blueprints" for those traits that are being being favored. Thus GENES provide the raw material for evolution.

## Variation in a Population

- Biologists can quantify genetic variation at two levels.
- Gene Variability
  - measures variation among genes themselves
- Nucleotide Variability
  - measures variation among the nucleotides that make up the genes

## Variation in a Population

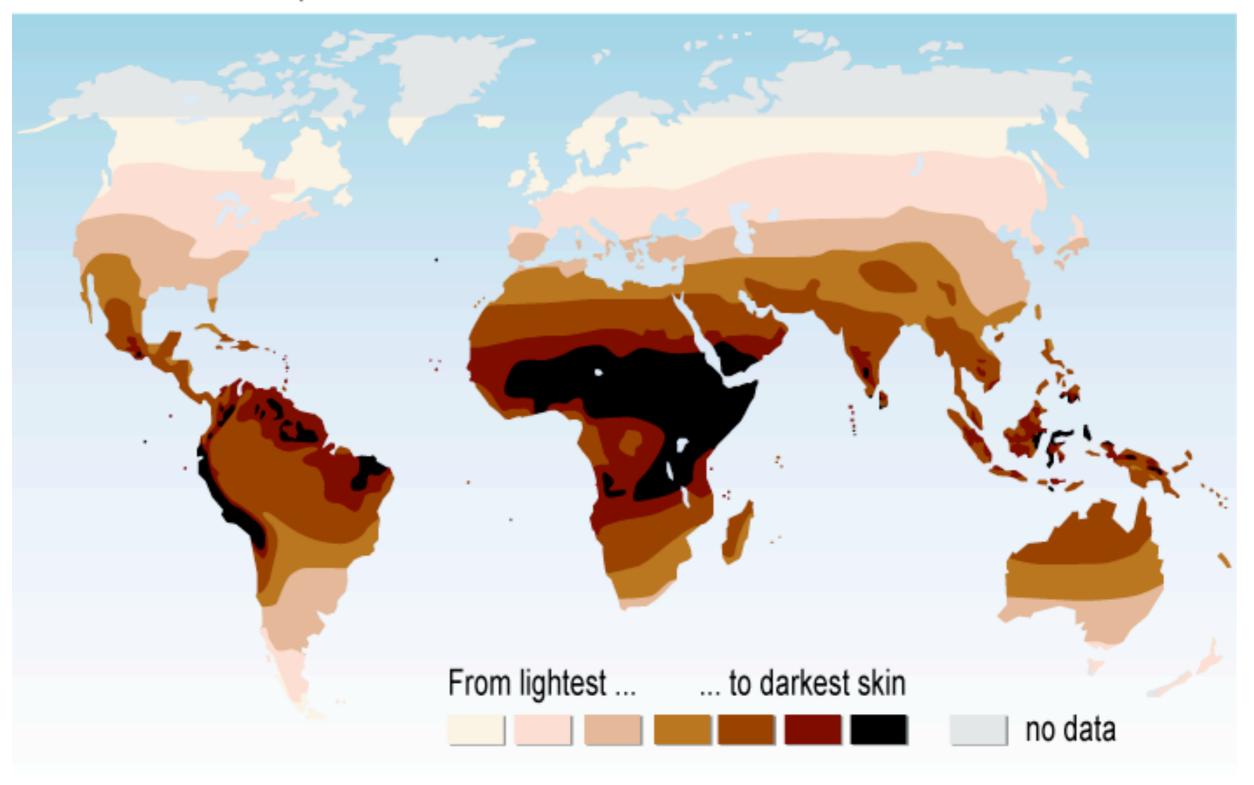
- Gene Variability
  - measures average heterozygosity, the percent of loci(gene positions) that are heterozygous (Aa as opposed to homozygous AA or aa)
- Nucleotide Variability
  - compares the DNA sequences of two individuals from a population and averages the differences seen in many of these comparisons

Both of these require biotechnology such as gel electrophoresis, PCR and restriction fragment analysis

### Variation Between Populations

- Biologists can also quantify genetic variation between two different populations... *geographic variation*.
- Differences are usually found
  - sometimes differences are purely random (genetic drift, discussed shortly)
  - other times the differences are likely a result of natural selection favoring certain traits over others
    - example- clines

# Skin colour map for indigenous people Predicted from multiple environmental factors



Source: Chaplin G.®, Geographic Distribution of Environmental Factors Influencing Human Skin Coloration, American Journal of Physical Anthropology 125:292-302, 2004; map updated in 2007.

#### Sources of Variation

- Recall: Evolution requires variation...
- BUT natural selection reduces variation in a population over time...
- THUS there must be mechanism(s) that generate(s) variation in a population otherwise populations would lose their ability to evolve

SEX & MUTATIONS... Generate Variation!

#### Sex Sex Sex

- Sexual Reproduction shuffles existing genes (alleles) deals them at random to produce unique individuals.
- The "shuffling mechanisms" include...
  - Crossing Over
  - Independent Assortment
  - Random Fertilization

Ultimately all the different genes that sex reshuffles originated in a population by way of a mutation.

# Mutations...gotta have them!

- Any and all new alleles (genes) arise from mutations.
- Mutations- are change in nucleotide sequence in an organisms DNA
  - Mutations are random
    - Only mutations that occur in gametes (sperm/eggs) can be inherited and passed generation after generation
    - Somatic (body cells) mutations can not be inherited and thus die with the individual.

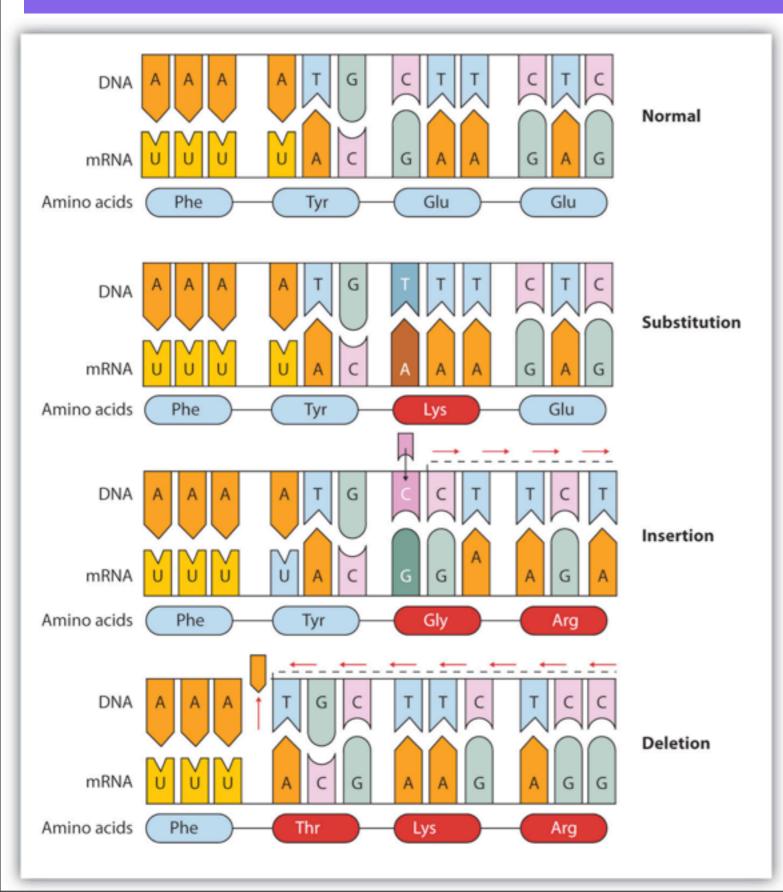
#### Effects of Mutations

- Mutations change nucleotide sequences, which in turn changes the protein product, which in turn alters the phenotype (trait).
- Most mutations are neutral or perhaps slightly harmful.
- On rare occasions a mutation results in a new trait that provides a selective advantage to the individual and should they reproduce to their offspring.
  - Over time this new gene becomes more common.

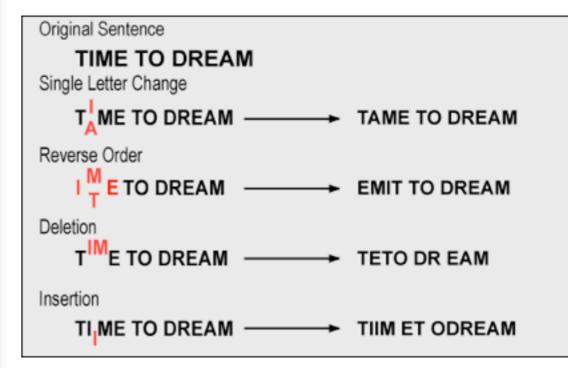
#### Mutations

- Mutations can occur in any number of ways.
  - Gene Mutations (small scale)
    - substituting one nucleotide for another
    - adding or deleting a nucleotide
  - Chromosomal Mutations (large scale)
    - rearranging chromosomal pieces (moving an entire gene or genes) in a genome
    - deleting or duplicating chromosomal pieces (deleting or duplicating an entire gene or genes) in a genome
    - disrupting an entire gene or genes at one time

#### Gene Mutations



The first two are called "point" mutations, it may or may not effect 1 amino acid

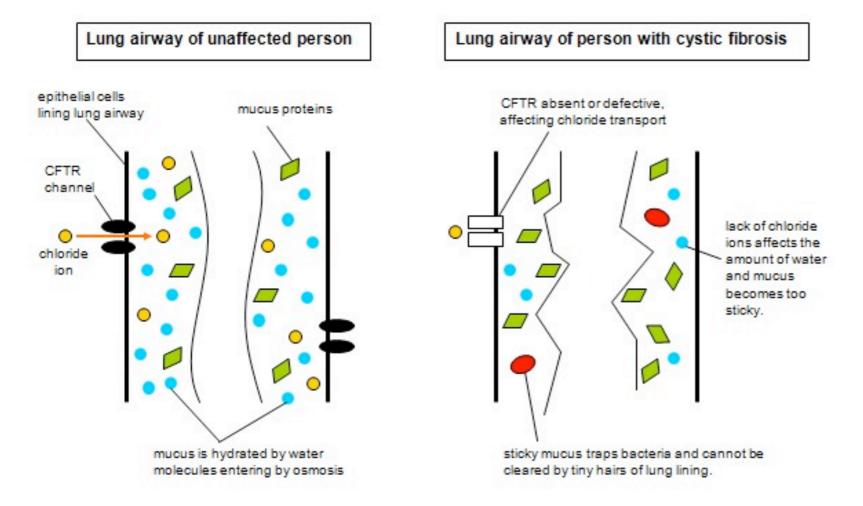


The last two are called "frameshift" mutations, it moves the entire reading frame, they have dramatic effects

#### Gene Mutations

Most mutations are neutral, have no effect on the fitness of the individual. Occasionally a mutation will provide a selective advantage and other rare times in results in a detrimental phenotype, reducing fitness. Here is an example of such a case

#### **CYSTIC FIBROSIS**



1 amino acid is lost out 508 and this results in cystic fibrosis

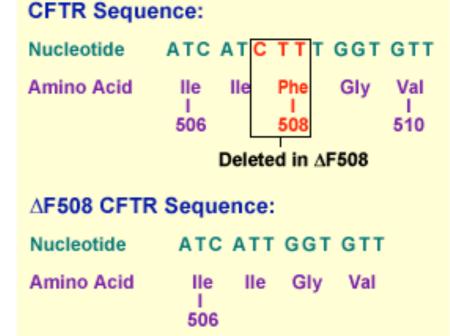
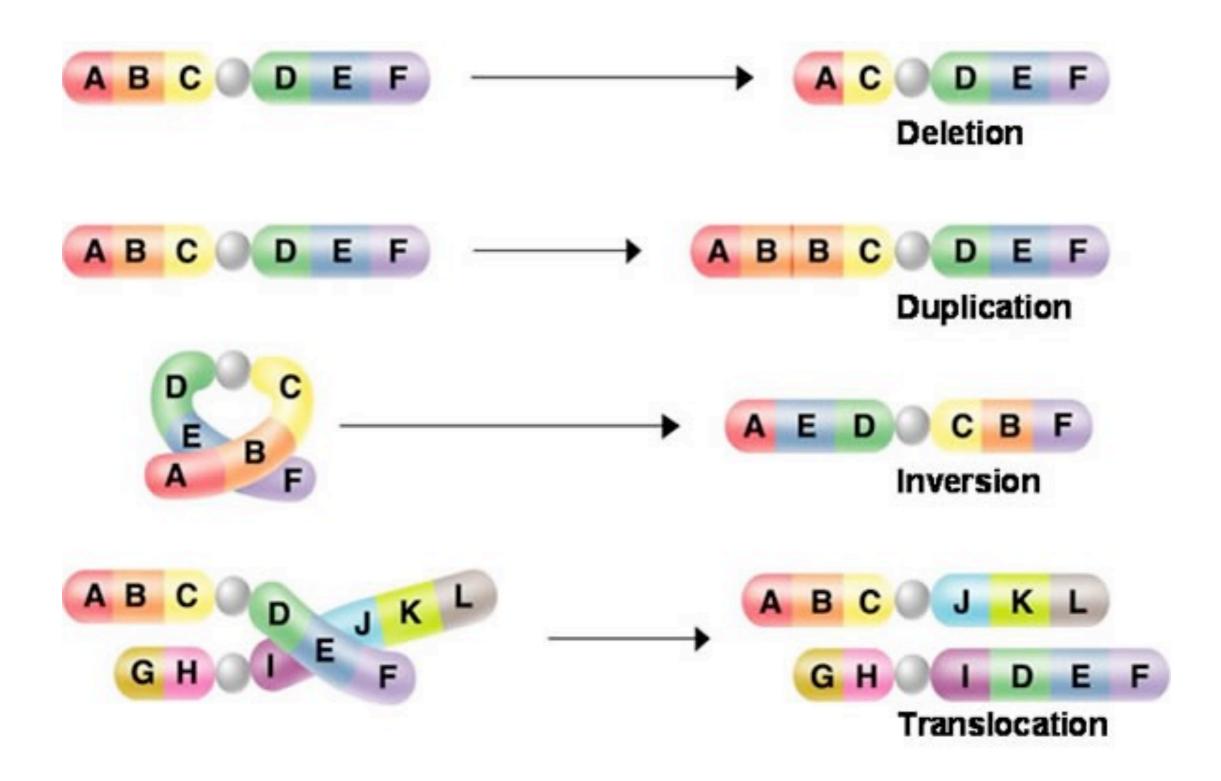


Figure 3: The deltaF508 deletion is the most common cause of cystic fibrosis. The isoleucine (IIe) at amino acid position 507 remains unchanged because both ATC and ATT code for isoleucine

#### Chromosomal Mutations



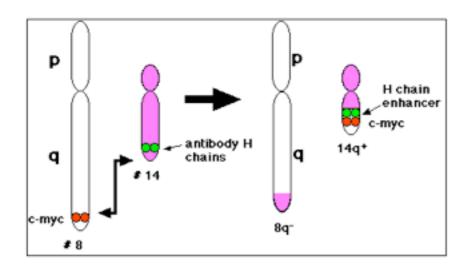
#### Chromosomal Mutations

#### **Burkitt's Lymphoma**

Burkitt's lymphoma is a solid tumor of <u>B lymphocytes</u>, the lymphocytes that the immune system uses to make <u>antibodies</u>. The genes for making antibodies are located on chromosomes **14** (the heavy [H] chains), **2** (kappa light chains), and **22** (lambda light chains). These genes are expressed only in B lymphocytes because only B cells have the necessary <u>transcription factors</u> for the <u>promoters</u> and <u>enhancers</u> needed to turn these antibody genes "on".

In most (approximately 90%) of the cases of Burkitt's lymphoma, a reciprocal <u>translocation</u> has moved the <u>proto-oncogene</u> c-myc from its normal position on chromosome 8 to a location close to the

enhancers of the antibody heavy chain genes on chromosome 14.



**Translocation** 



Seven-year-old
Nigerian boy with a several month history of jaw swelling which had been treated with antibiotics. The tumor was ulcerated and draining.

# Consider this...

- A long time a mutation occurred in mammals that enabled them to detect volatile chemicals (they could smell)
- Since then this gene has been duplicated and alter so many times that humans have about 1000 olfactory receptors, mice about 1300.
- The mutation rate in mice and humans is roughly equal
- Today about 60% of human olfactory genes are inactive due to mutation but mice have lost only 20%.
  - 1. Why did the original mutation (a mistake) become so numerous and common over time?
  - 2. What can we infer about olfaction between humans and mice today?

## Conservation Ecology

Main Idea: Small populations have low diversity and as a result are more vulnerable.



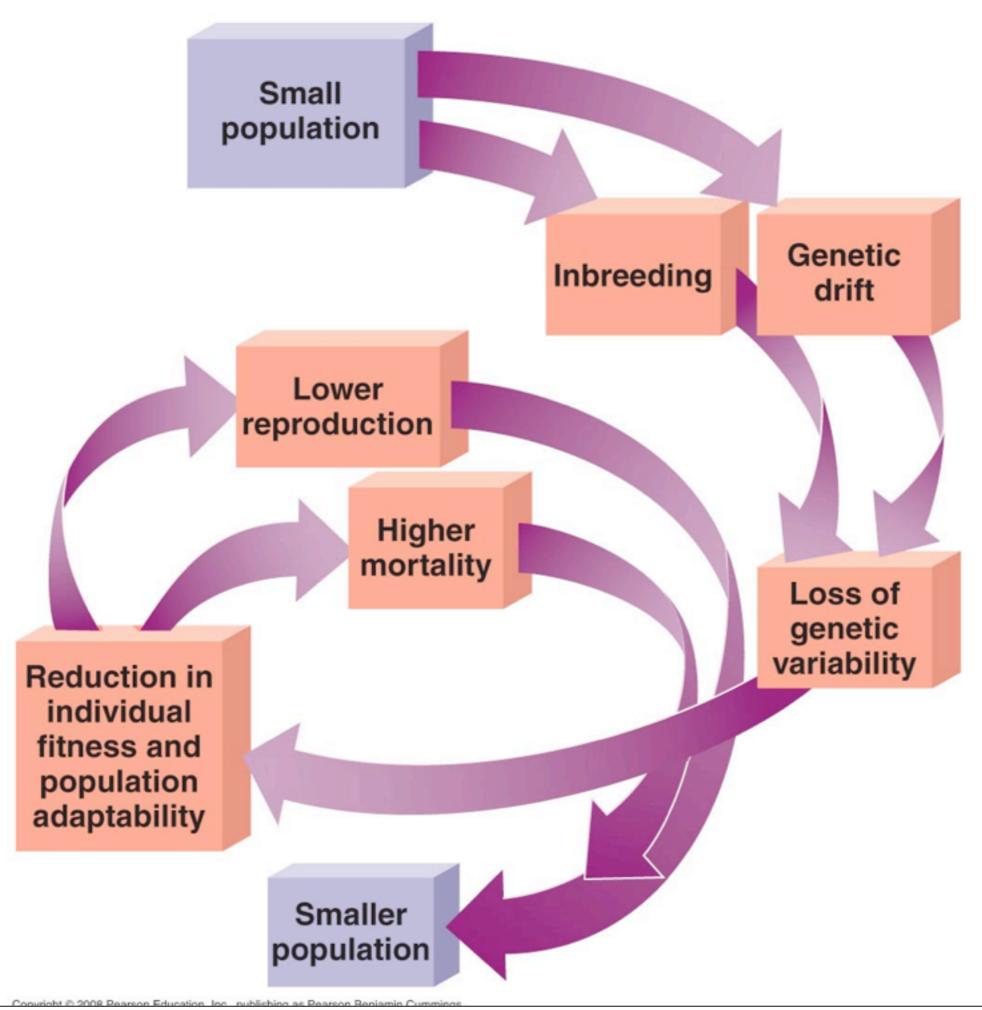
## POPULATION CONSERVATION FOCUSES ON POPULATION SIZE, GENETIC DIVERSITY AND CRITICAL HABITAT

## A. Small Population Approach

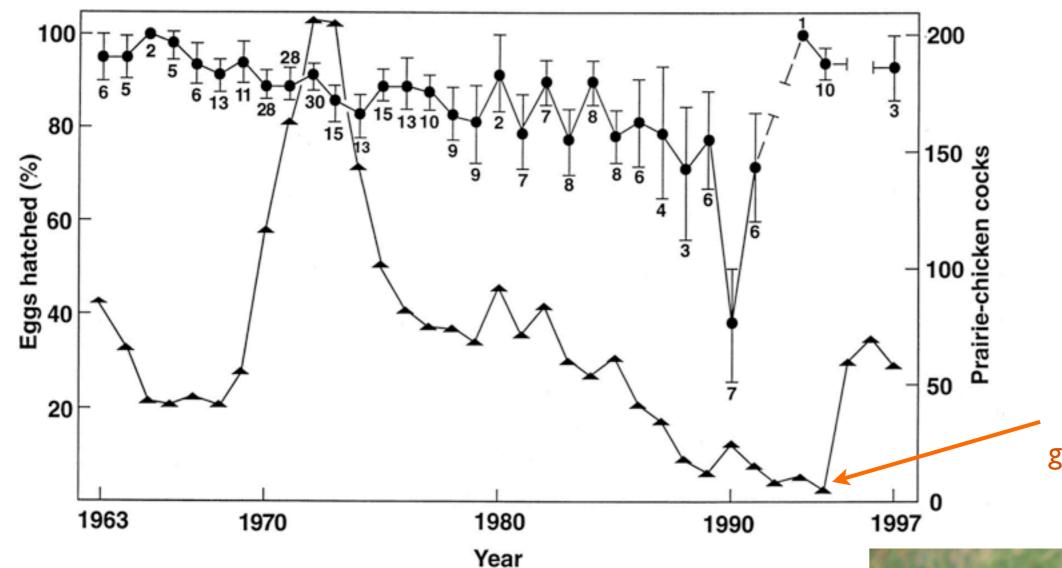
- Particularly vulnerable to over harvesting and habitat loss
- study processes that cause extinctions of small populations

## 1. The Extinction Vortex: Evolution Implications

 Small populations suffer from Inbreeding and Genetic Drift which result in a loss of genetic variation.



#### 2. Case Study: Greater Prairie Chicken



Transfusion of genetic variability

Imported 271 birds from a larger population elsewhere

#### 3. Minimum Viable Population Size

- Minimal population size at which a species is able to sustain its numbers..(MVP)
- Depends on the organism and other factors

#### 4. Effective Population Size

- Population size by itself can be misleading, Effective population size is based on breeding potential.
  - Consider a population of 1000 with only 5 females and a population of 100 with 40 females. See a difference?

How might the age of individuals in a population play a role?

\*Read Case Study on Grizzly Bears if you get a chance

Essential knowledge 1.A.1: Natural selection is a major mechanism of evolution.

d. Environments can be more or less stable or fluctuating, and this affects evolutionary rate and direction; different genetic variations can be selected in each generation.

## "The Rate of Speciation"

- The time it takes for geographical isolation to lead to reproductive isolation depends on a number a variables.
  - which genes mutate, the rate of mutations, extent of genetic drift, degree of environmental differences between the populations, generation times
- Intrinsic reproductive isolation leading to speciation may take thousands or millions of years or it may happen virtually "over night".

## Patterns in the Fossil Record

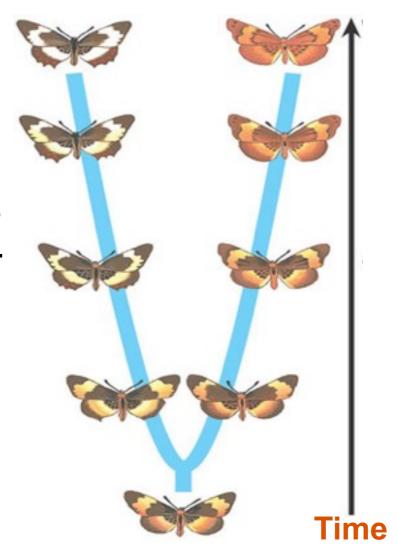
- The fossil record looks both static and dynamic.
- Some species appear in the fossil record virtually unchanged for over several strata (long time).
  - marine invertebrates
- While other species suddenly appear in the stratum, (short time)
  - punctuated equilibrium-Stephen Jay Gould
- And yet others appear to gradually change over time.
  - gradualism-Charles Darwin

## Speciation Rates

- The fossil record and other experimental data suggest that speciation may occur relatively slowly (40 million years) or very quickly (4,000 years).
- The interval of time between speciation events includes the elapsed time from before populations of the newly formed species start to diverge + the time it takes for speciation to occur once divergence begins.
  - On average speciation events occur at a rate of 6.5 million years and rarely less then 500,000 years.

 Darwin described speciation as a gradual change over time.

Gradualism model. Species descended from a common ancestor gradually diverge more and more in their morphology as they acquire unique adaptations.



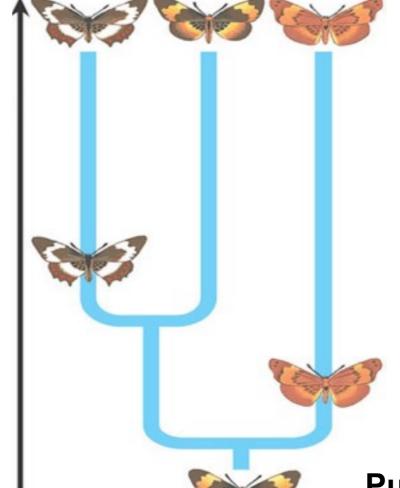
However a lot of fossil evidence looks very different from this.

 Contemporary biologists Stephen Jay Gould and Niles Eldridge proposed a different view of speciation.

Keep this in mind,
say a species
survives 5 million
years, but the
events that lead to
speciation occur
over 50,000 years,
this is only 1% of
its time on earth.

Since fossil formation is rare none of these fossil may exist only ones from the old and new species that lived for millions of years.

Time

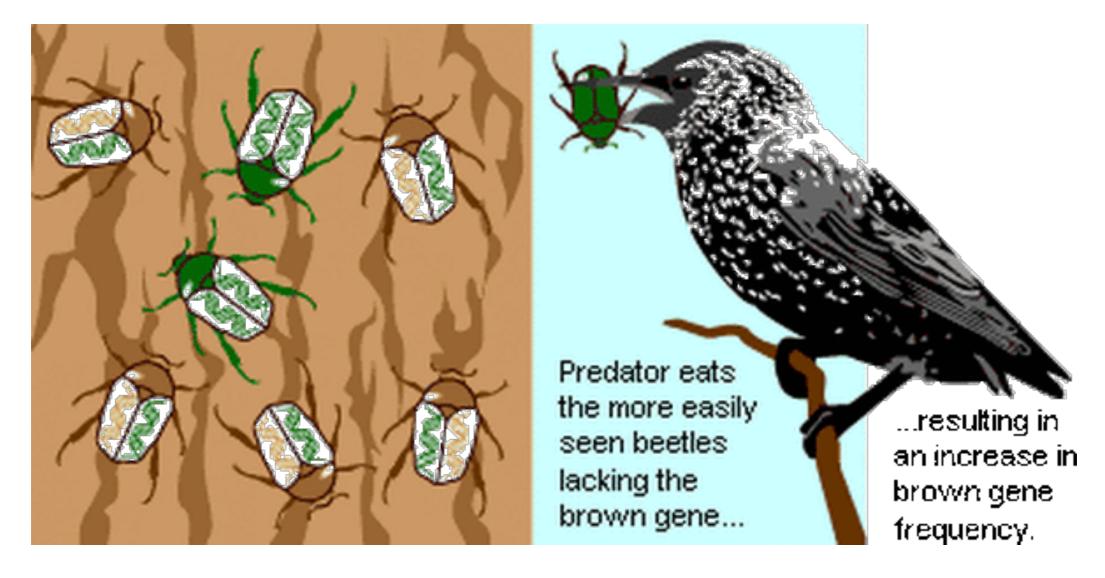


Many fossils appear similar for a long time, then exhibit abrupt changes in a short period of time followed by another period of stasis.

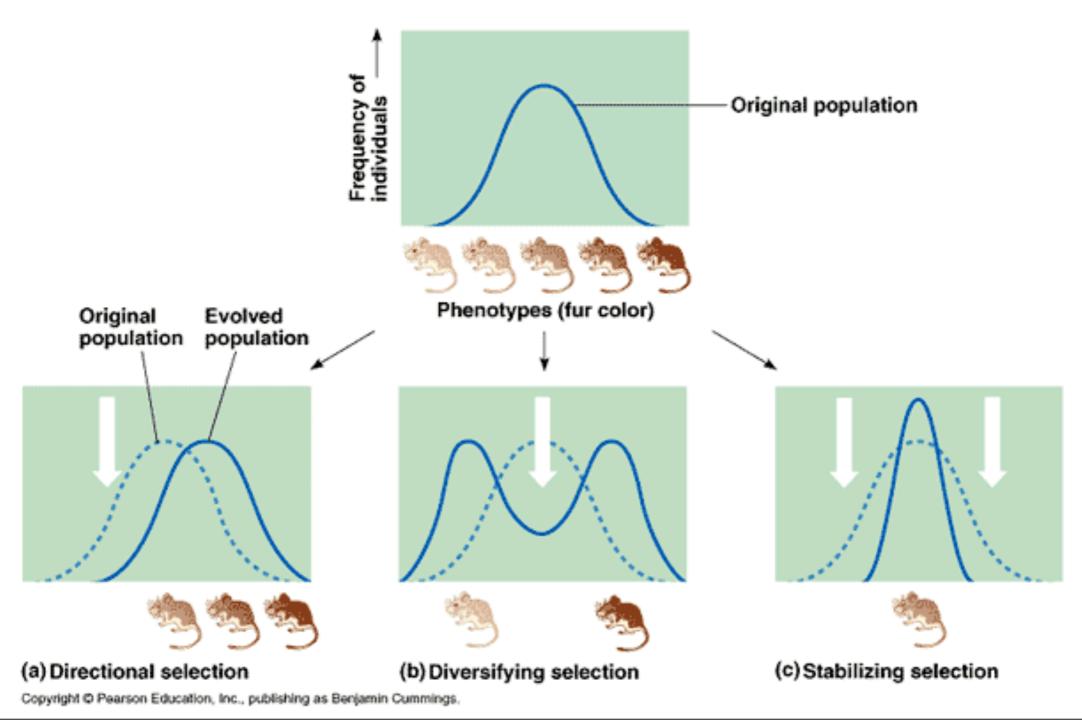
Punctuated equilibrium model.

A new species changes most as it buds from a parent species and then changes little for the rest of its existence.

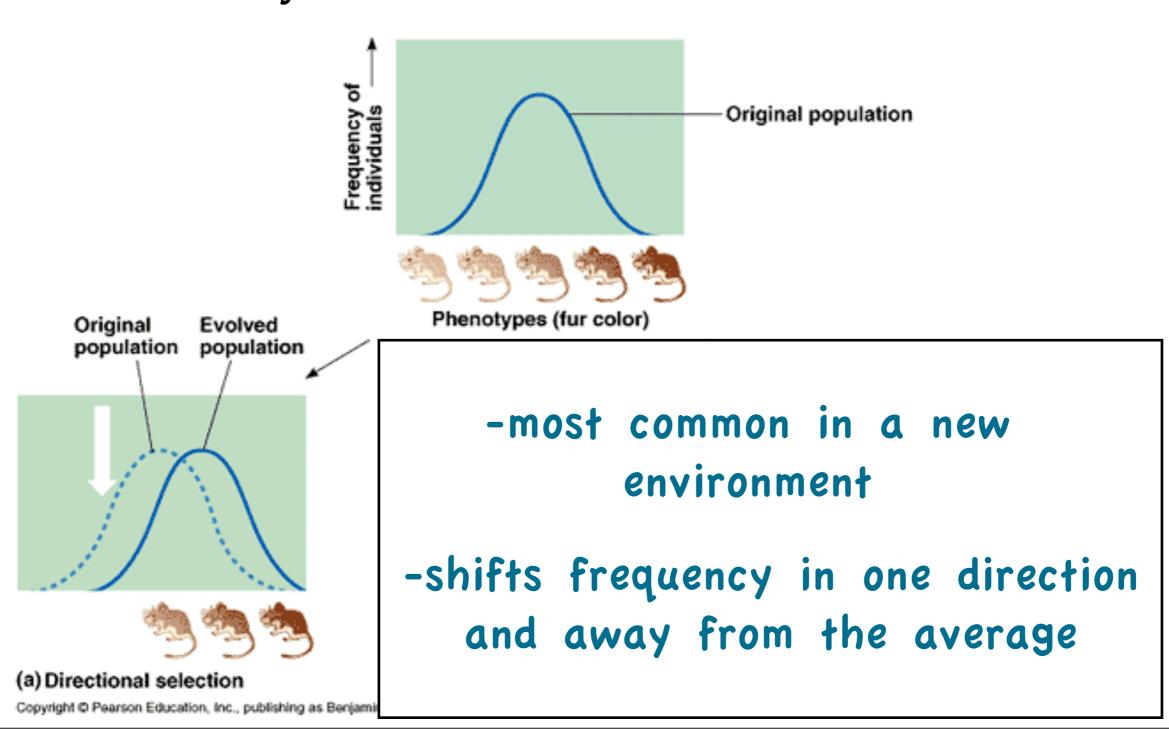
- Natural Selection- differential reproductive success.
  - Certain favorable traits become more frequent, non favorable traits become less frequent and the population becomes better suited to its environment over time.



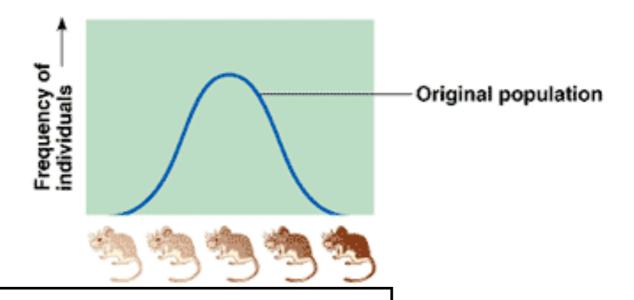
 Natural Selection- can alter frequency distribution of traits in 3 ways.



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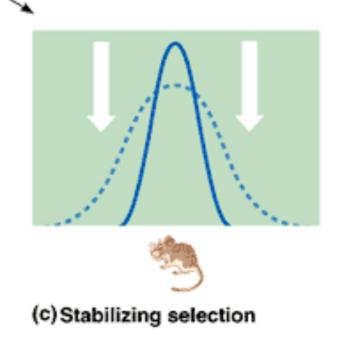


 Natural Selection- can alter frequency distribution of traits in 3 ways.

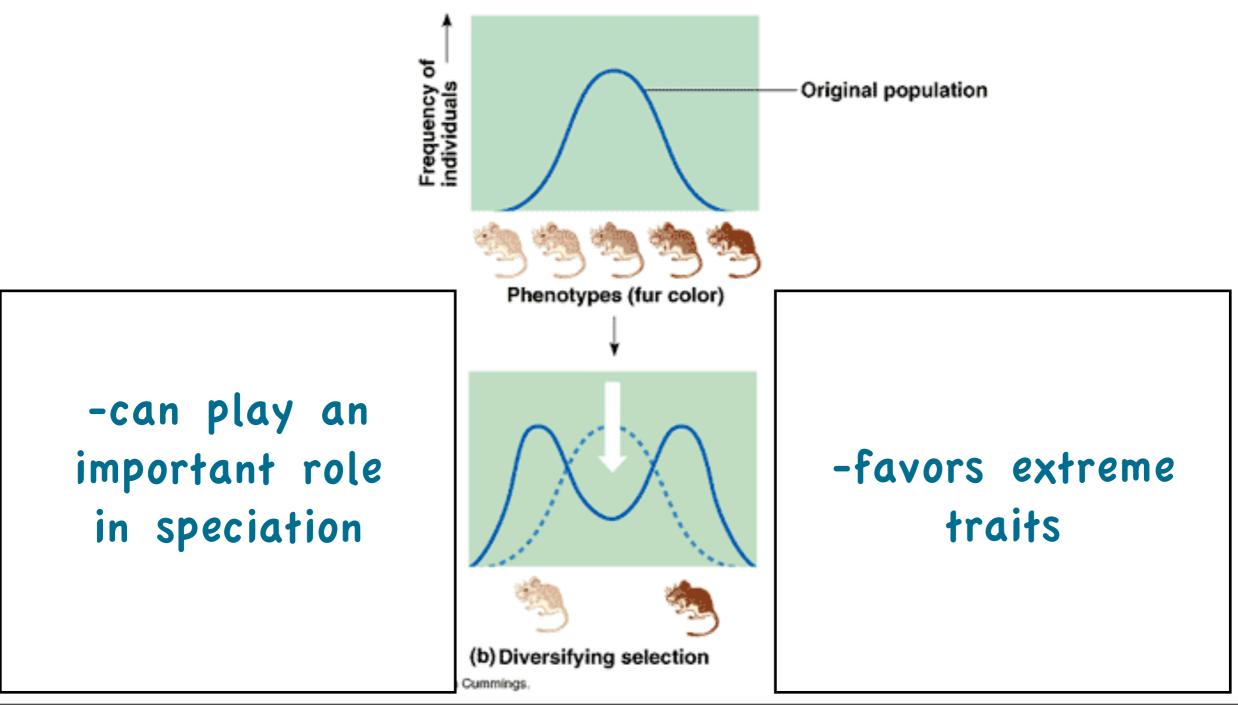


-reduces variation

-selects the extremes, moderate traits are favored



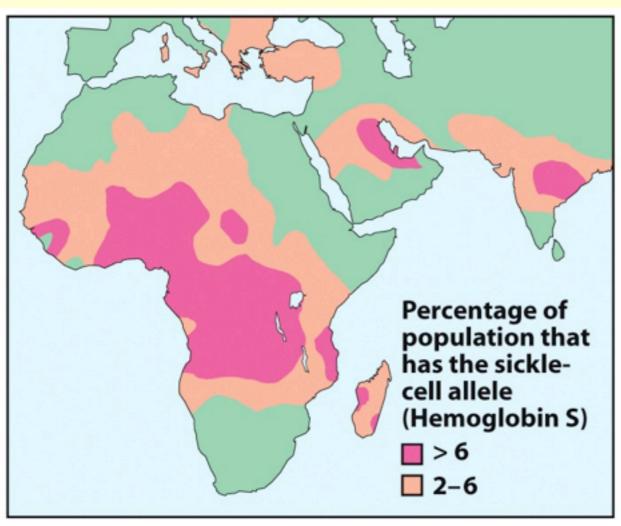
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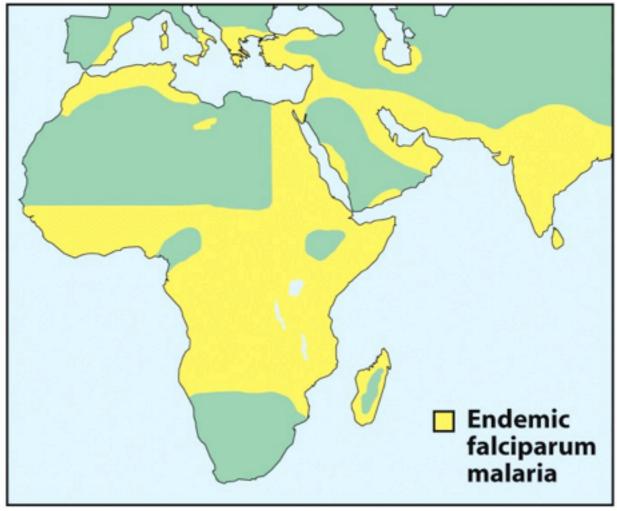


Essential knowledge 1.A.1: Natural selection is a major mechanism of evolution.

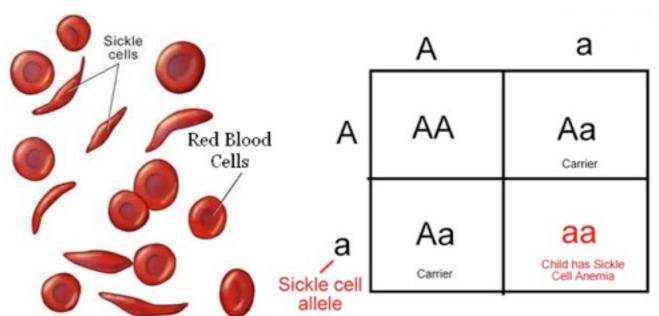
e. An adaptation is a genetic variation that is favored by selection and is manifested as a trait that provides an advantage to an organism in a particular environment.

## Sickle Cell Trait & Malaria

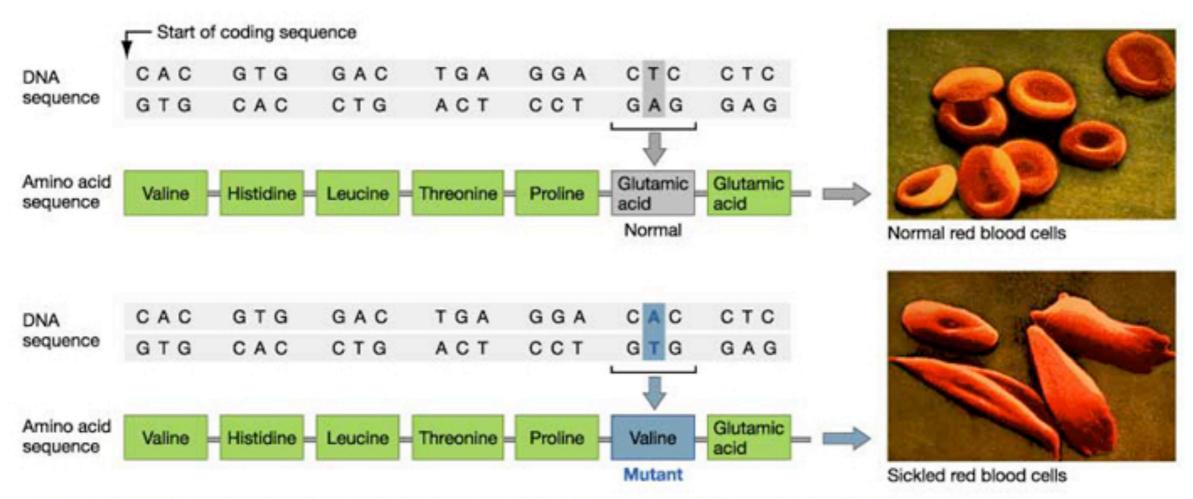








## Sickle Cell Trait & Malaria



The change in amino acid sequence causes hemoglobin molecules to crystallize when oxygen levels in the blood are low. As a result, red blood cells sickle and get stuck in small blood vessels.

This is a "substitution" mutation notice the thymine was switched with alanine.

The normal beta subunit consists of 438 nucleotides and 146 amino acids.

A change in 1 nucleotide, changes 1 amino acid resulting in sickle cell disease

# Pocket Mice Video

http://www.hhmi.org/biointeractive/making-fittest-natural-selection-and-adaptation

Essential knowledge 1.A.1: Natural selection is a major mechanism of evolution.

f. In addition to natural selection, chance and random events can influence the evolutionary process, especially for small populations.

## Changing Gene Pools

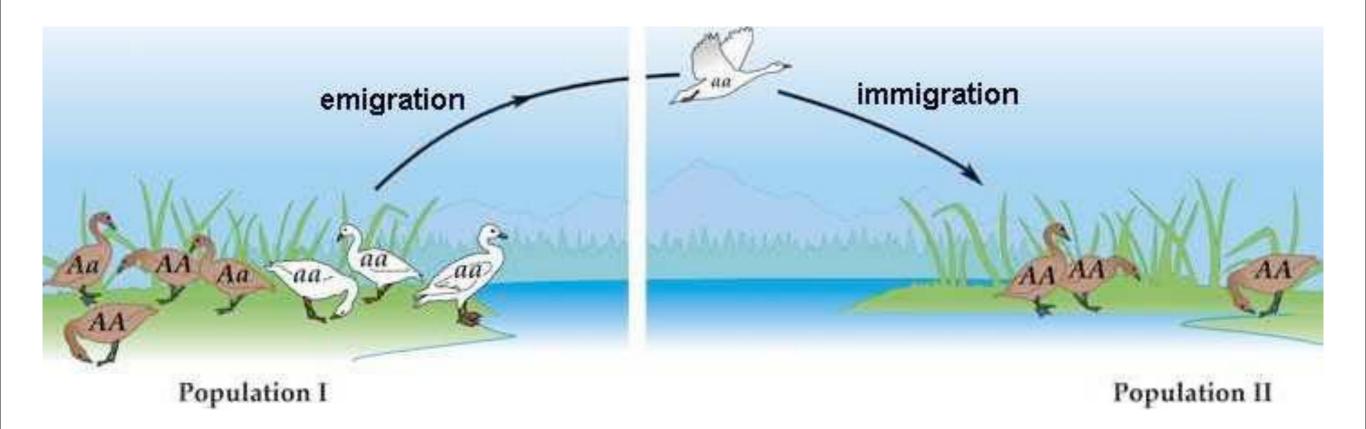
- A deviation in any of the 5 Hardy-Weinberg conditions will change allele/gene frequencies.
- However three processes in particular change gene pools or cause evolution more than anything else.
  - Gene Flow
  - Genetic Drift
  - Natural Selection

The smaller the population the greater there effect

Important Note...Natural selection is the only one that results in a population becoming more well adapted to its environment over time.

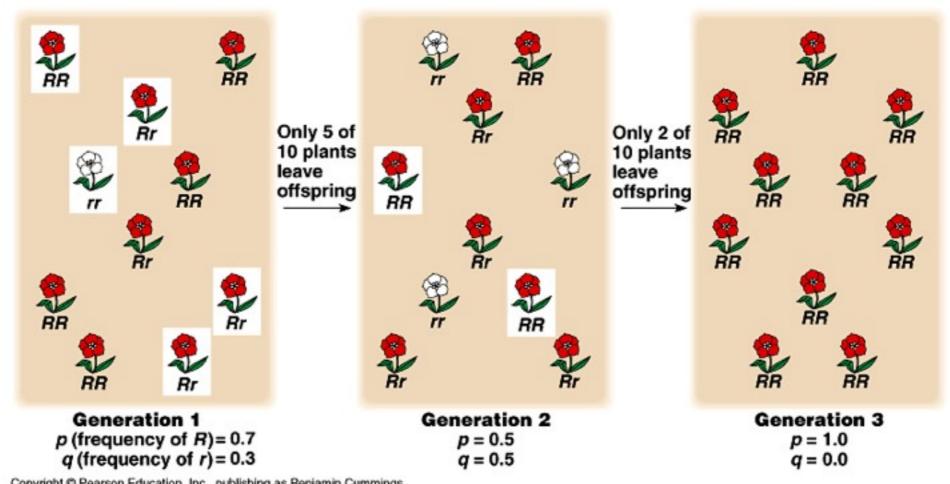
## Gene Flow

- Gene Flow- a population can gain or lose alleles/genes as a result of individual or gamete movement.
- Gene flow often reduces differences between populations, it can "melt" two populations into one.



## Genetic Drift

- Genetic Drift-random and unpredictable fluctuations in allele/gene frequencies.
  - Genetic Drift will have a greater effect on smaller populations
  - Genetic Drift tends to reduce variation
  - Genetic Drift can "fix" (100% frequency) harmful alleles



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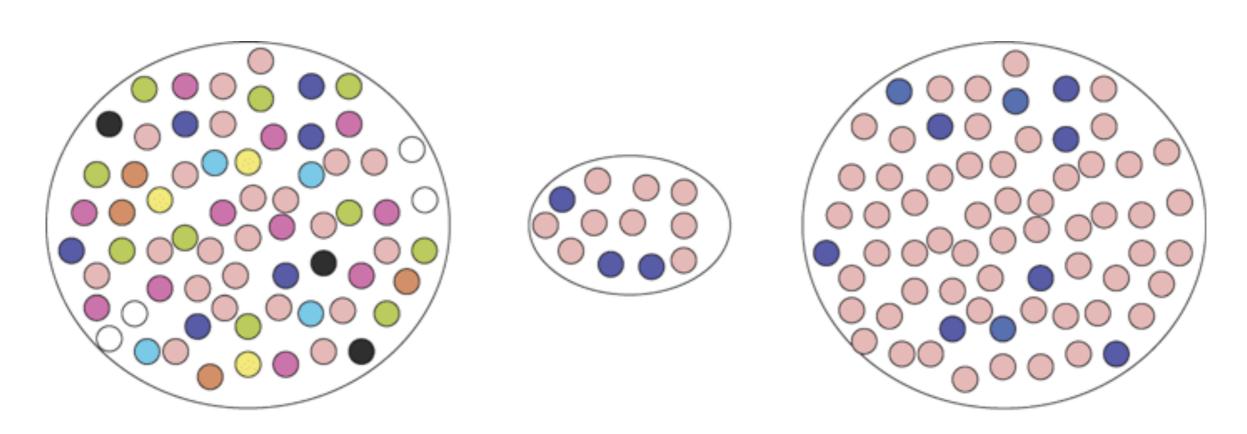
#### Generation I p (frequency of A) = 0.6 q (frequency of a) = 0.4 AA Because of Generation II chance events, p = 0.8only the highlighted q = 0.2moths produce offspring... AAGeneration III p = 1.0q = 0.0AA AA ..causing the AA frequency of the A allele to reach 100% in two generations. AA

#### Genetic Drift

- There are two events that increase the effect of genetic drift.
  - A bottleneck effect
  - A founder effect

## The Bottleneck Effect

- Bottleneck Effect- can occur when there is large reduction in population size, the survivors may not reflect the composition of the original population.
  - can have a lasting effect on populations



## The Founder Effect

 Founder Effect- when a small group leaves a larger population, establishes a new population that is not representative of the original.

original population

new population Essential knowledge 1.A.1: Natural selection is a major mechanism of evolution.

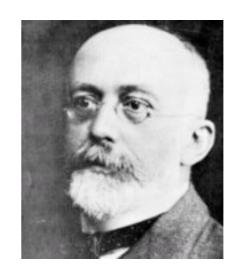
g. Conditions for a population or an allele to be in Hardy-Weinberg equilibrium are: (1) a large population size, (2) absence of migration, (3) no net mutations, (4) random mating and (5) absence of selection. These conditions are seldom met.

## Non-Evolving Populations

- A population not evolving would have to have ALL allele/gene frequencies remain constant over time in order for the gene pool itself to remain constant.
  - Keep in mind populations have hundreds to thousands of alleles/genes.
  - Is it likely or even possible that all allele/gene frequencies remain unchanged over time?...NO
    - Never the less we can still ask...Are there conditions where a populations' allele(s)/gene(s) would NOT change?

## Godfrey Hardy & Wilhelm Weinberg





- A British mathematician and German physician asked this same question and their work tells us...
  - Yes it is possible for allele/gene frequencies to remain unchanged in a population, Yes it is possible that a population does not evolve over time.
  - However it is only possible provided the population meets certain criteria.

## Hardy-Weinberg Equilibrium

- This principle describes a hypothetical non-evolving population
- In order for a population to maintain the Hardy-Weinberg equilibrium (not change) they must meet all 5 of the following conditions:

Very large population	No genetic drift can occur.
No emigration of immigration	No gene flow can occur.
No mutations	No new alleles can be added to the gene pool.
Random mating	No sexual selection can occur.
No natural selection	All traits must equally aid in survival.

# How likely is it that a population meets these conditions?

## Hardy-Weinberg Equilibrium

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No natural selection	All traits must equally aid in survival.

How likely is it that a population meets these conditions?

Not at all likely, close to impossible!

Essential knowledge 1.A.1: Natural selection is a major mechanism of evolution.

h. Mathematical approaches are used to calculate changes in allele frequency, providing evidence for the occurrence of evolution in a population.

To foster student understanding of this concept, instructors can choose an illustrative example such as:

Graphical analysis of allele frequencies in a population Application of the Hardy Weinberg Equation

## Evolution...Measured?

- Can we determine objectively whether evolution is taking place in a population?
  - YES
- Can we quantify the rate or degree of evolution in a population?
  - YES
- Can a population not evolve?
  - YES

## Population Genetics

- Recall: a *population* is a group of organisms of the same species, in the same general area that interbreed successfully.
- A gene pool represents all genes (alleles), at every locus in all members of a population.
- Recall: <u>evolution</u> is a change in gene (allele) frequencies in a population over time.

## Population Genetics

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# Measuring Allele Frequency

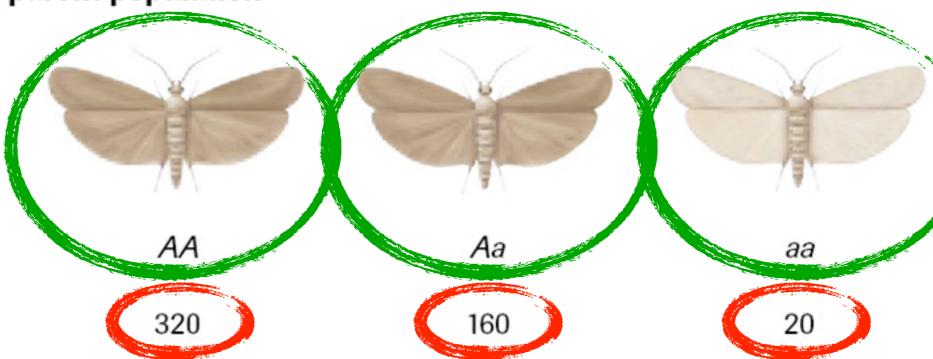
- To determine that evolution has occurred we must know the allele(s) frequency in a population at a point in time and compare it to the allele(s) frequency at a later time.
- This implies/requires that we can measure allele(s) frequencies...but HOW?
  - First we must the know the total number of individuals in a population
  - Next we must the know the frequency of a trait in a population.
  - Lastly we need to which alleles control the trait we are examining

which alleles control the trait Genetic structure of parent population

phenotypes

genotypes

number of moths (total = 500)



population size frequency of a trait in a population

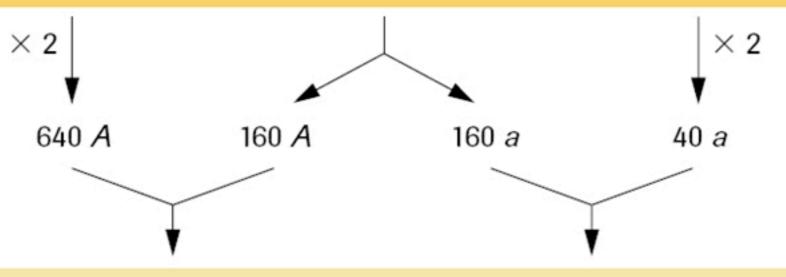
genotype frequencies

$$\frac{320}{500} = 0.64 \text{ AA}$$
  $\frac{160}{500} = 0.32 \text{ Aa}$   $\frac{20}{500} = 0.04 \text{ aa}$ 

$$\frac{160}{500} = 0.32 \, Aa$$

$$\frac{20}{500} = 0.04 \ aa$$

number of alleles in gene pool (total = 1000)



allele frequencies

**Bingo!** 

$$\frac{800}{1000} = 0.8 A$$

$$p = \text{frequency of } A = 0.8$$
  $q = \text{frequency of } a = 0.2$ 

$$\frac{200}{1000} = 0.2 a$$

$$q = \text{frequency of } a = 0.2$$

# Measuring Allele Frequency

- To determine that the moth population is evolving we simply recalculate the allele(s) frequency in the population at a later time.
  - If we find that the frequency of A is no longer 0.8 or 80% and a is no longer 0.2 or 20% then the population has evolved because its allele frequencies have changed.
  - If we find that the frequency of A remains 0.8 or 80% and a remains 0.2 or 20% then the population <u>has not evolved</u> because its <u>allele frequencies have not changed</u>.

- This principle describes a hypothetical non-evolving population
- When gametes are donated randomly and matings are also random allele frequencies will not change and we can calculate genotypic frequencies from allele frequencies:
  - We can summarize the union of gametes in an algebraic equation

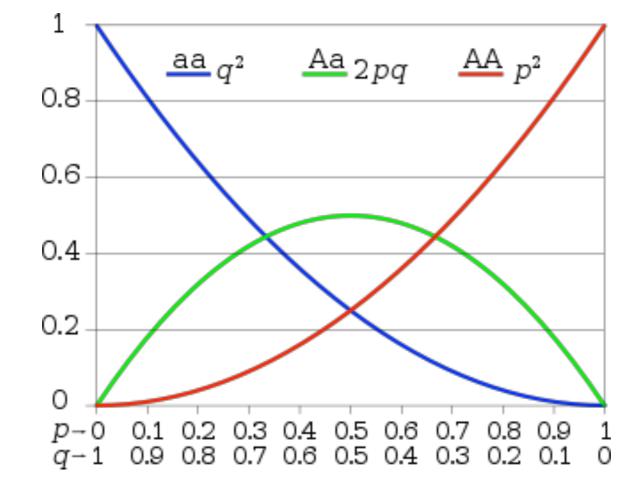
$$(p+q)(p+q) = p^2 + 2pq + q^2 = 1$$

Thus a locus with two alleles, the genotypes will appear in the proportions.

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

Here is graphic representation of allele and genotypic

frequencies.



#### Hardy-Weinberg Health Applications

#### Hardy-Weinberg proportions in a geometric view

• The Hardy-Weinberg equation can be used to estimate the percentage of the population that carriers an allele for a disease (among other things as well)

fraction of ova with A: p oick an ovum at random fraction of ova with B: q

fraction fraction of sperm of sperm with A: p with B: q Homozygotes from Heterozygotes from B ovum A ovum and A sperm and A sperm pq Homozygotes Heterozygotes from from B ovum B ovum and A sperm and B sperm рq

pick a sperm at random

#### Solving Hardy-Weinberg Problems

 Every Hardy-Weinberg problem is essentially the same, you will be given one (or more) of the 5 variables below, you will then use simple algebra to solve for the unknown.

#### **Know This**

#### p = dominant allele

q = recessive allele

 $p^2 = AA$  genotype

2pq = Aa genotype

 $q^2 = aa genotype$ 

#### Follow these steps

First- determine the unknown (which of the variables on the left do need to solve for)

Next- determine the known (which of the variables on the left do we have)

Last- using the equations on the right and simple algebra solve for the unknown

#### **Use These**

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

$$p + q = 1$$

Let's try a tough problem together.

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There is a hypothetical population with 100 individuals. There is a trait with two alleles, B and b. 12 people are normal homozygous dominant (BB), 78 people are carriers of a disease (Bb) and 10 people have the disease (bb). Is this population evolving?

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Before we can predict what a non-evolving population looks, we need to know what the actual population looks like.

Start by finding alleles frequencies "p" and "q".

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Population with 100 individuals will have 200 alleles.

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12 people are normal homozygous dominant (BB) so (12)(2)= 24 B's

78 people are carriers of a disease (Bb) so (78)(1) = 78 B's and 78 b's

10 people have the disease (bb) so (10)(2) = 20 b's

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Now 24B + 78B = 102 total B's out of 200 or 102/200 = 0.51 or 51% B's "P" And 78b + 20b = 98 total b's out of 200 or 98/200 = 0.49 or 49% b's "Q"

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Remember "p" = dominant allele (B)

Remember "q" = dominant allele (b)

And since there are only two alleles p + q = 1

OK Now we have p and q let's plug them into the Hardy-Weinberg equation and see what a non-evolving population would look like.

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 $(0.51)^{2} + 2(0.51)(0.49) + (0.49)^{2} = 1$ 
 $0.26BB$ 
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Now that we know what a non-evolving population looks like. We have to compare our "predicted" non-evolving population with our "actual" population

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Predicted	Non-Evolving	Actual	Po	pulation

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 12 BB

$$(0.50)(100) = 50 \text{ Bb}$$
 78 Bb

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Bb

# Learning Objectives

- LO 1.1 The student is able to convert a data set from a table of numbers that reflect a change in the genetic makeup of a population over time and to apply mathematical methods and conceptual understandings to investigate the cause(s) and effect(s) of this change. [See SP 1.5, 2.2]
- LO 1.2 The student is able to evaluate evidence provided by data to qualitatively and/or quantitatively investigate the role of natural selection in evolution. [See SP 2.2, 5.3]
- LO 1.3 The student is able to apply mathematical methods to data from a real or simulated population to predict what will happen to the population in the future. [See SP 2.2]