Darwin’s Theory

Modern Synthesis

Pre-Darwinian Observations

- **Taxonomy**: a ‘familial’ resemblance among groups of animals
- The fossil record: animals have changed over time.
- **Embryology:** related groups of animals pass through similar developmental stages
- **Homology**: some structures seem to be modifications on a common theme (e.g., wings of bats and birds, fish fins, mammalian limbs)
- **Vestigial organs**: non-functional today, but perhaps functional in an ‘ancestral’ form
- **Biogeography:** very similar groups of animals sometimes have very discontinuous distributions (e.g., camels and llamas)
- **Animal husbandry**: selective breeding can dramatically change the appearance of animals.

Darwin’s Great Synthesis

- Darwin took the seven observations mentioned above and, along with Malthus’ description of population growth and Lyell’s theory of uniformitarianism, combined them with his vast knowledge of the natural world to make his theory of natural selection based on four simple but powerful postulates:

Postulates of Natural Selection (in modern language)

1. The individual organisms that make up a population vary in the traits they possess, such as their size or shape.
2. Some of the trait differences are passed on to offspring. This means that the variation in the trait has some genetic basis.
3. In most generations, more offspring are produced than can survive. As a result, only a subset of the offspring that are produced survive long enough to reproduce.
4. The subset of offspring that survive and reproduce is not a random sample of the population. Instead, individuals with certain traits are more likely to survive and reproduce or to produce the greatest number of offspring. The individuals with there traits are, in Darwin's words, "naturally selected"

Natural selection and the origin of species: an overview

- **Members of a species are never uniformly distributed across the entire range of the species.**
- **Therefore different *demes* (breeding subgroups) of the species are subject to slightly different environments and hence slightly different selection pressures**
- **Anything that diminishes the flow of genes from deme to deme (physical separation, behavioral differences, etc.) will allow natural selection to begin to operate differently on the demes**
- **Given enough time and enough separation the differences between the demes will become so great that they can no longer mate and produce viable, fertile offspring.**

Darwinian fitness

- Natural selection has often been described (as Darwin did) as "survival of the fittest"
  - Fitness, in Darwinian terms, means the ability to pass more of your genes on into the next generation than can your competitors.
  - Thus natural selection operates only on those characters that increase fitness.

This has some unpleasant consequences for those of us beyond child-bearing age!

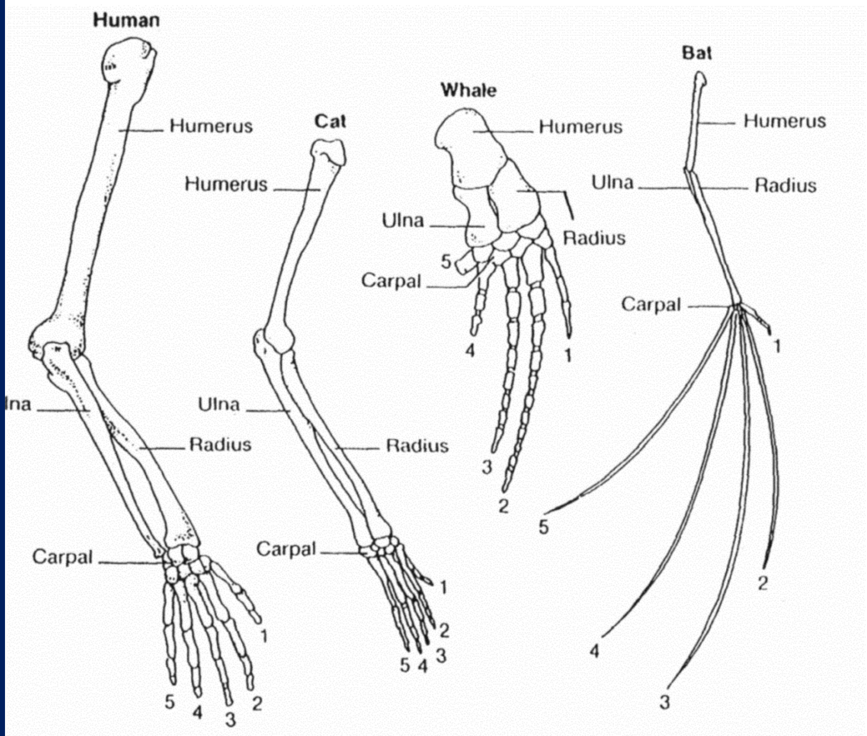
Some Examples of successful strategies

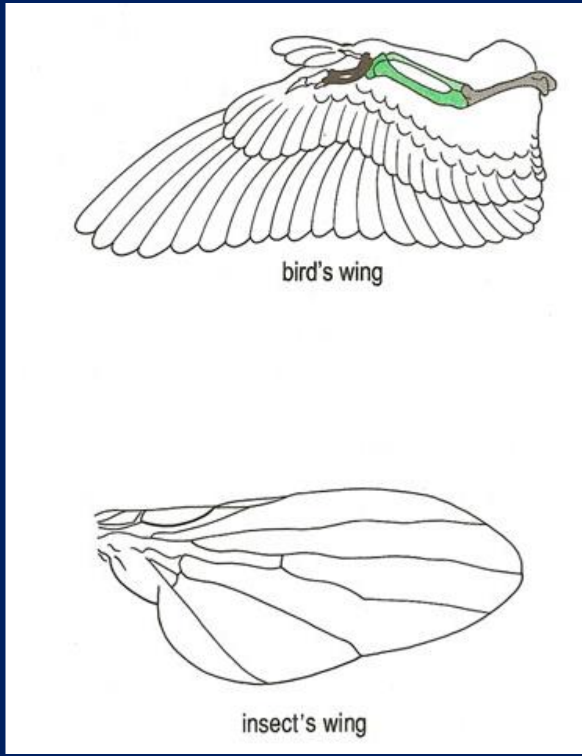
- Live short and have lots of offspring: e.g., insects, annual plants, many invasive species
- Live long and spend a lot of energy on parental care: e.g., us
- For males, be as promiscuous as you can get away with.
- For females, choose the most “fit” mate
- Disperse your gametes (or offspring) over the widest possible area: e.g., wind-pollination, burrs, Genghis Khan.

Does Evolution answer the questions?

- Taxonomy:
  - Organisms are related by descent from a common ancestor. The more remote the ancestor the more the difference (e.g., higher level taxa).
- Fossil Record:
  - Change over time is evidence of evolution. Extinction can be explained by natural selection (among other reasons).
- Homologies:
  - Descendants of common ancestors will share the same structure, although they may have been modified to different extents by natural selection.

Homology vs. Analogy





Homologous structures arise due to common ancestry. Analogous structures arise due to convergent evolution.

Does Evolution answer the questions?

- Embryology:
  - "ontogeny recapitulates phylogeny". Well, not totally, but close enough.
- Vestigial Organs:
  - Inherited from common ancestors, but diminished due to lack of positive selection pressure.
- Biogeography:
  - If there are camels in Africa and Asia and again in South America, there must be camels in North America. Paleontology shows fossil camels North America in the Tertiary (ca. 30MYA). Similarly, North America and Europe were connected by a land bridge.
- Animal husbandry:
  - A kind of natural selection.

Evolutionary Theory From Darwin to Today

Modern Synthesis

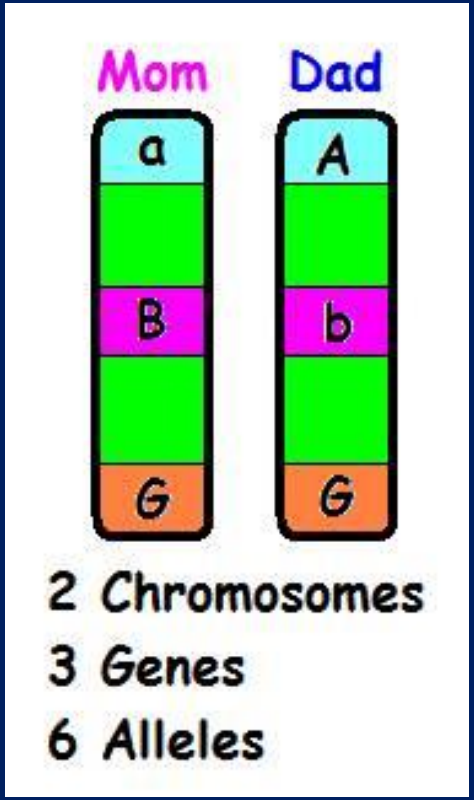
Evolution and Genetics

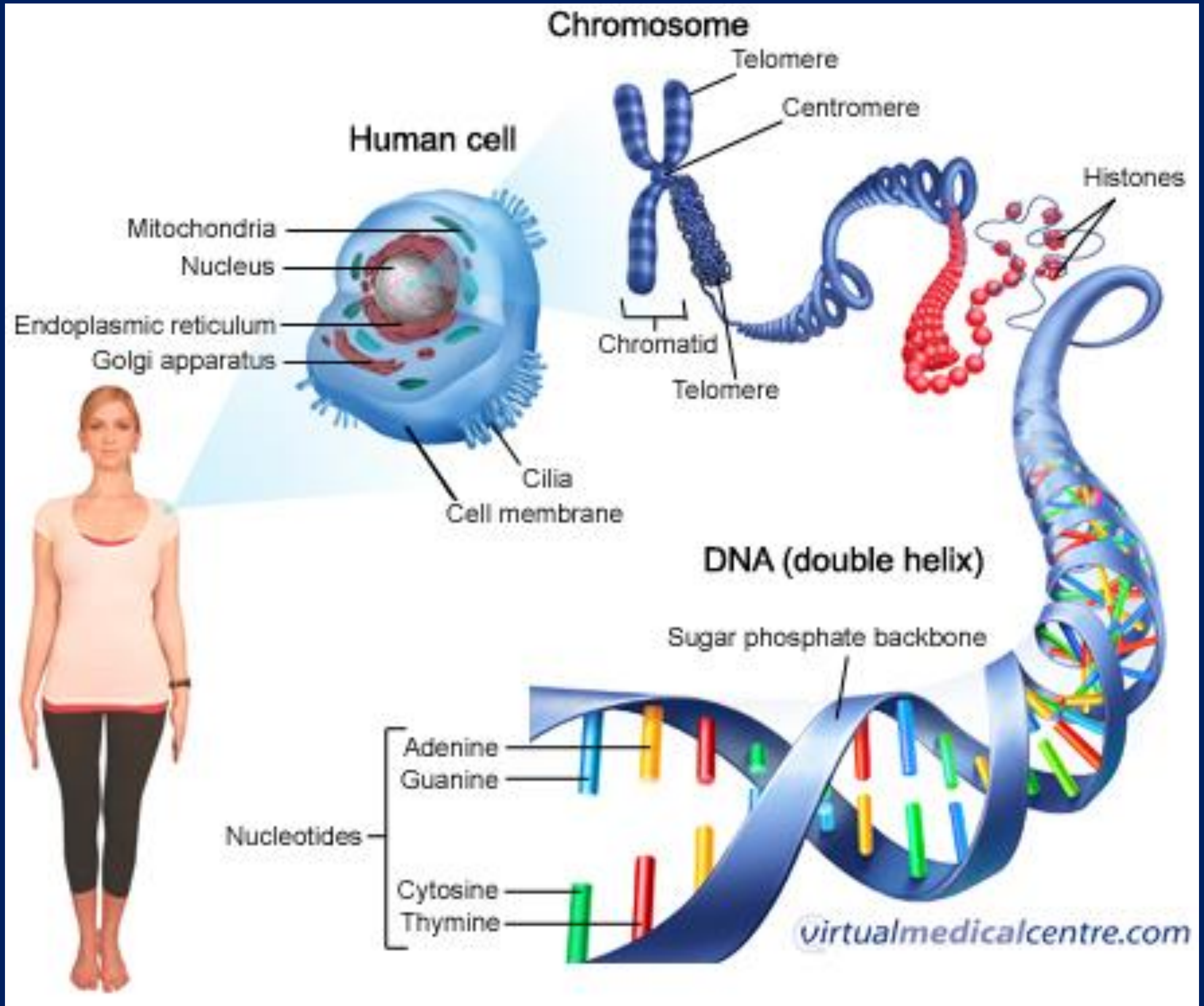
- The one thing Darwin missed out on was the fundamental mechanism underlying both the transmission of traits and their variation and change over time.
- He had the driving force (natural selection), but not the biological machinery it works on (genes).

Genes and alleles

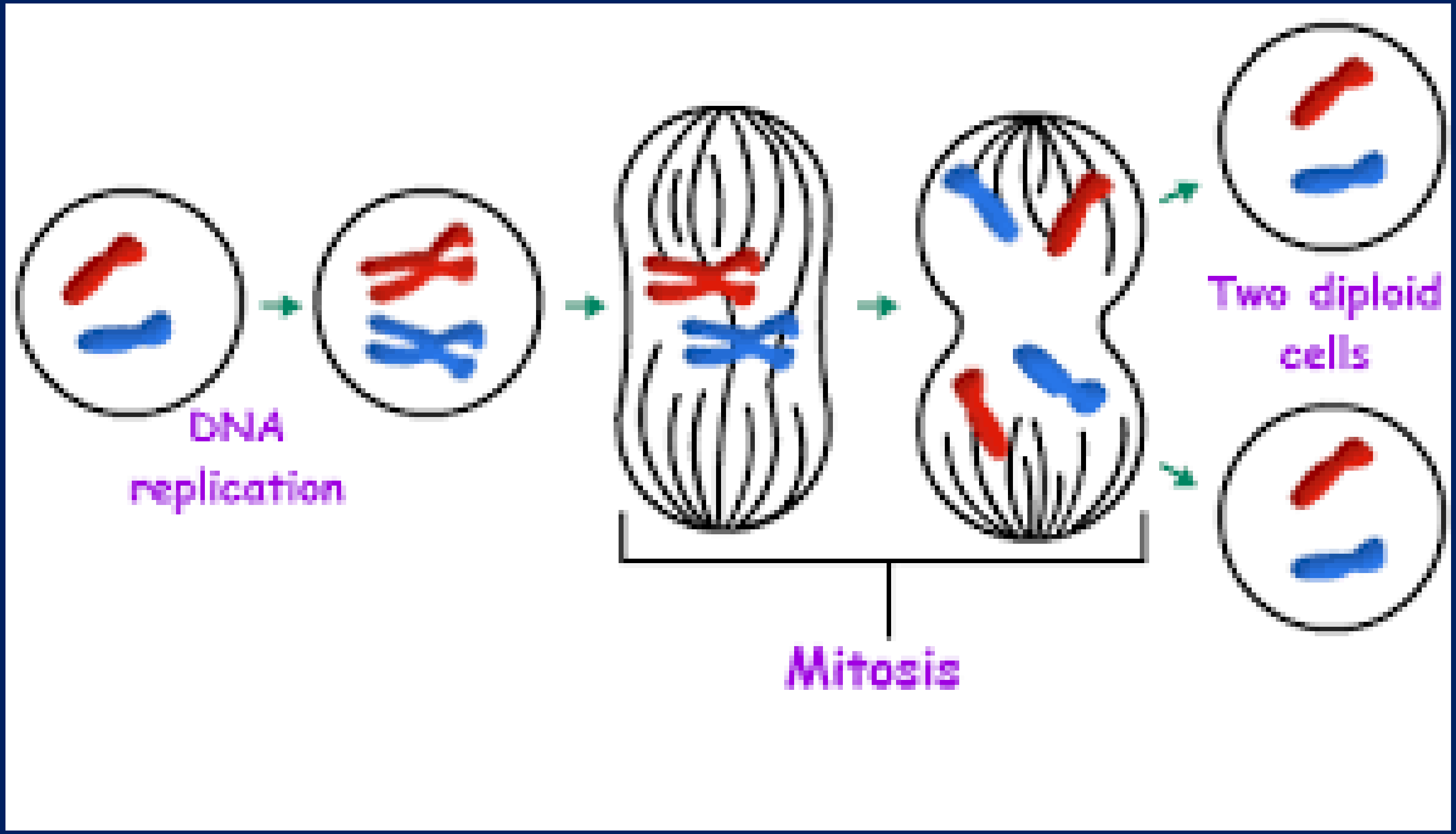
- **Genes** are the basic units of heredity.
- A given gene may have two or more forms, e.g., tongue-rolling (T,t); blood types(I^A,I^B,i). The specific forms are called *alleles.*
- Most organisms have two copies of each gene; one from each parent. If the alleles are the same, you are ***homozygous*** for that allele ; if they differ you are ***heterozygous*** for that allele.
- Your ***genotype*** is a list of your alleles; your ***phenotype*** is your actual appearance; the relationship between the two is very complex.
  - This relationship may be tight (e.g., insects) or loose (e.g. plants), allowing greater or less adaptation of the individual).

Genes and DNA





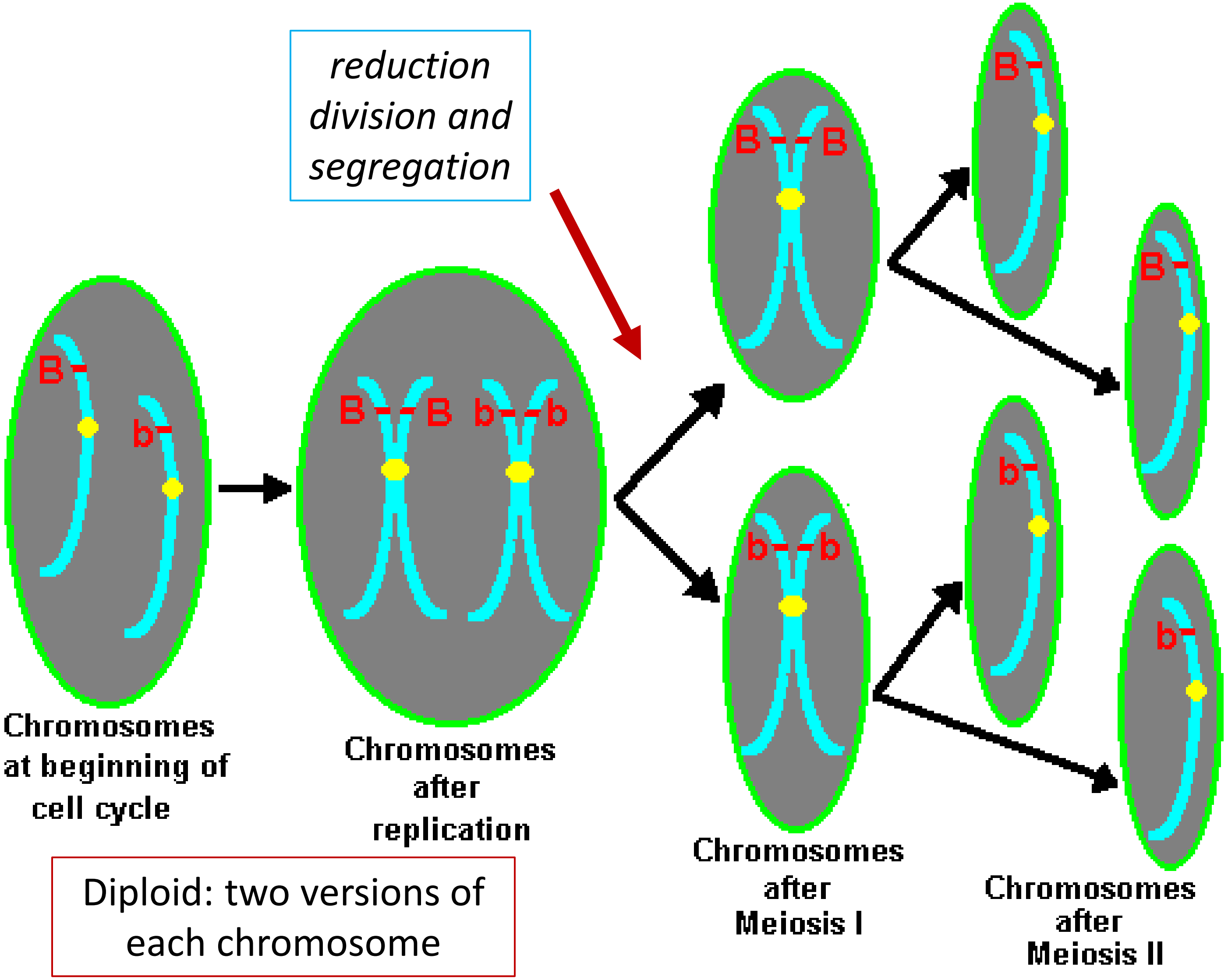
A Primer on Cell Division: Mitosis



Mitosis is the process of cell division designed to produce two identical daughter cells.

1. Normally dispersed DNA in the nucleus replicates itself
2. The DNA condenses into chromosomes, as Dyads, identical strands linked at a centromere
3. The replication spindle forms
4. Centromeres attache to the spindle and line up on the equator
5. Sister chromosomes separate at their centromeres.
6. Spindle fibers shorten and pull each member of each dyad to the opposite pole.
7. The cell divides ("cytokinesis")
8. New nuclei form and the DNA again disperses.

Gamete Formation: Meiosis



Diploid: two versions of each chromosome

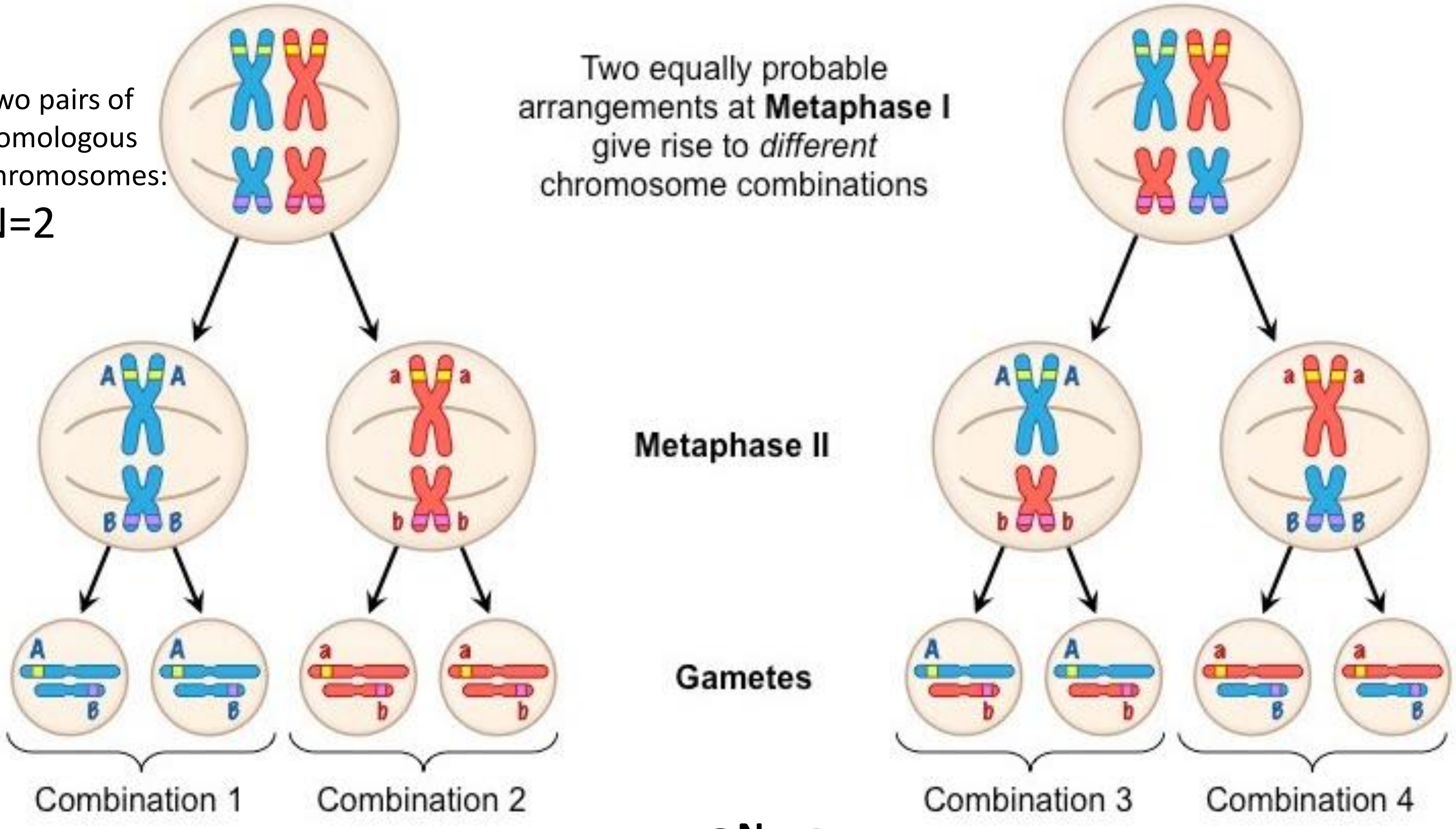
Haploid: one version of each chromosome

Upon mating, two haploid gametes unite to form one diploid zygote, now with shared information.

How Meiosis Produces Variability

Two pairs of homologous chromosomes:

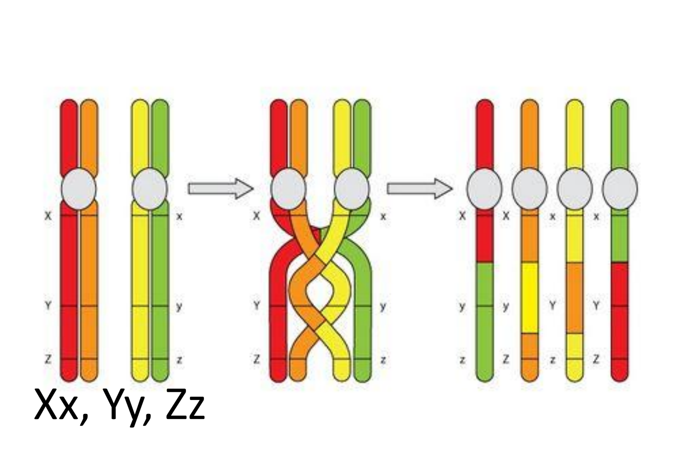
N=2

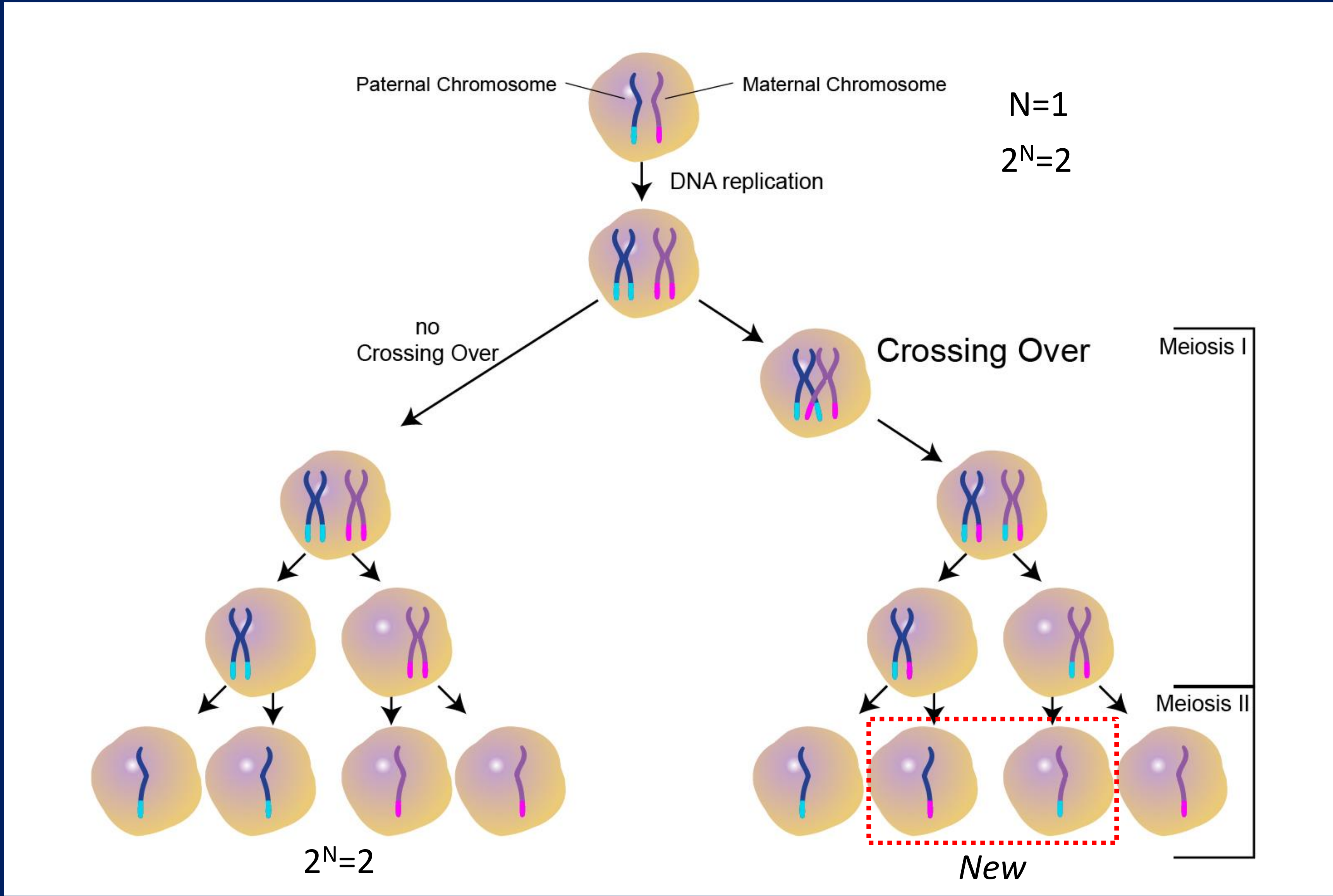


2^N=4

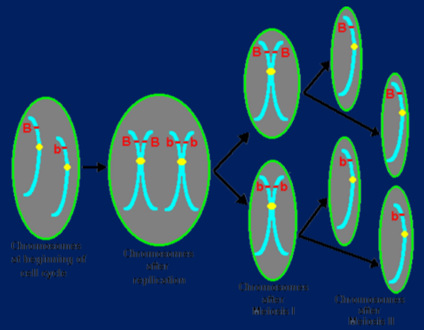
- For each pair of homologous chromosomes there exists two ways for them to line up at Metaphase I.
- This means that there are four possible daughter cells (2^N) for each homologous pair.
- Each daughter cell will produce two daughter cells after metaphase II. They will be haploid but identical, *unless crossover occurred at prophase I.*
- The number of different haploid gametes thus produced *without crossover* is 2^N.
- Humans have 23 homologous pairs.
- 2^{23}=8,388,608 possible gametes
- Fertilization in sexually reproducing species unites two haploid gametes to produce a diploid zygote.
- Thus some 16,777,216 unique zygotes are possible for two humans!!
  - *Your kids are not just "one in a million!"*

A wrinkle: Crossing Over ("Recombination")





Mendel's Rules



- Segregation:
  - Of every pair of alleles, one and only one will go to each daughter cell. This is a consequence of the segregation of the chromosomes in meiosis I.
- Independent Assortment:
  - The probability of a given allele of a gene appearing in a gamete is independent of the appearance of any other allele of any other gene[1].

[1]This rule is violated when the genes are on the same chromosome, depending on how far apart they are on the chromosome (i.e., probability of crossing over). The information may be used to "map" genes on chromosomes.

Muller's Ratchet

- Asexually reproducing organisms do not undergo meiosis and hence have no recombination (crossing over). Thus chromosomes are inherited as units, unchanged except for mutations.
- Once a deleterious mutation occurs in a chromosome it will be inherited in half of the offspring of that mating (unless there's a back mutation).
- Chromosomes will accumulate mutations.
- Each generation thus has more mutations (the ratchet)
- Eventually the organism will go extinct.

Genetics and Evolution: Population Genetics

- Population genetics is the study of how alleles are distributed in a population and how that distribution changes over time in response to evolution.

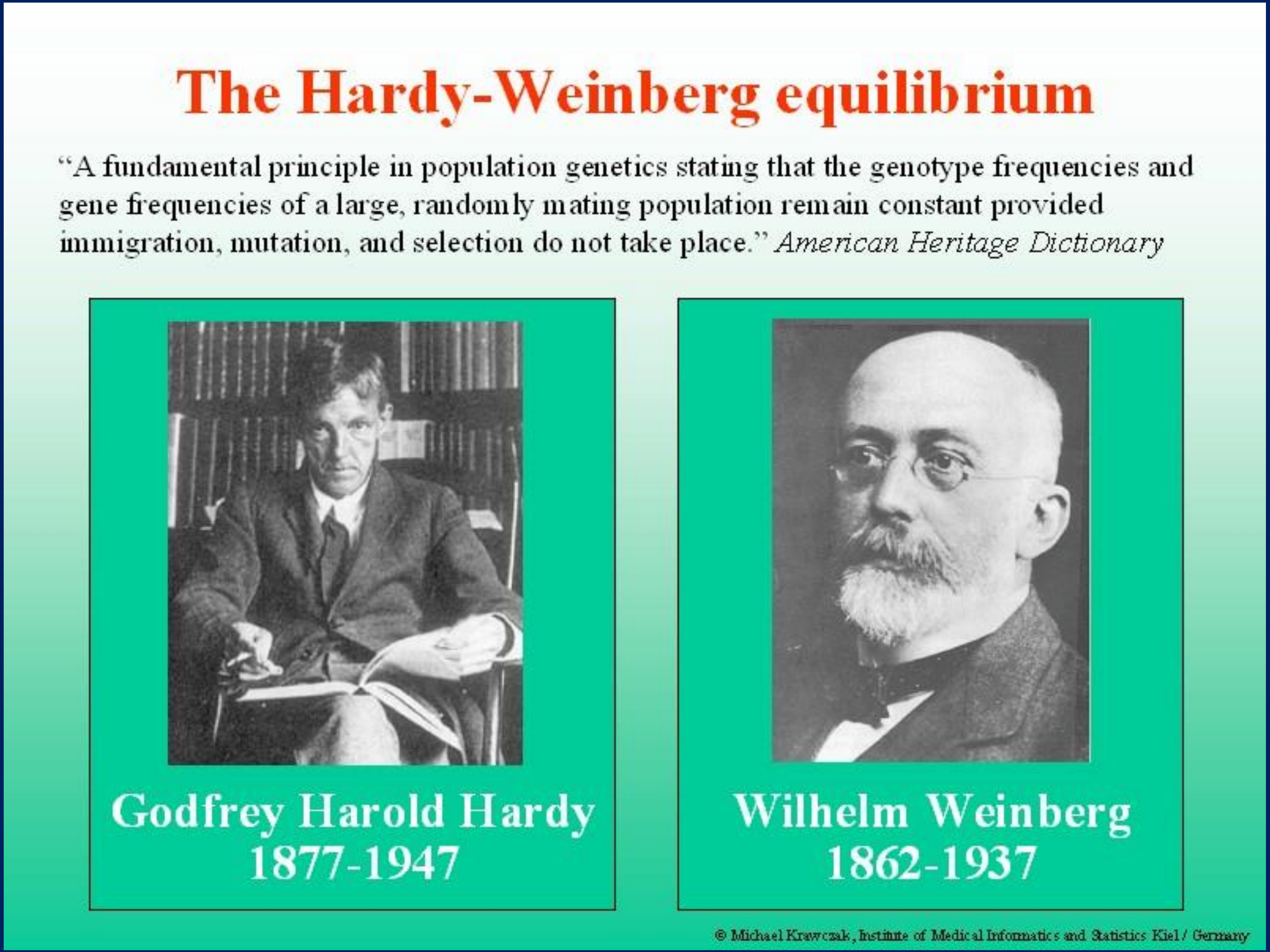
Population Genetics: allele frequency

- Simply put, allele frequency is the total number of a given allele in a population divided by the total number of alleles of that gene.
- For example, in every 100 Europeans there will be about 80 Rh- alleles (some individuals will have two Rh- alleles (*homozygotes*) while other have one Rh- and one Rh+ allele (*heterozygotes*))
- so the frequency of Rh-, F(Rh-), in Europeans is about 0.4

$$\frac{\text{80 Rh- alleles}}{\text{100 individuals x 2 Rh alleles per individual}} = \frac{80}{200} = 0.4$$

- Then since there are only two alleles for Rh, the frequency of the Rh+ allele, F(Rh+), must be 1-F(Rh-), or 0.6.

The Hardy-Weinberg Equilibrium



How it Works

- Suppose we have a gene whose alleles are Z and z, and F(Z)=0.8, so F(z)=0.2.
- Now let's mate the population randomly:

Occurance in the gene pool

	0.8Z	0.2z
0.8Z	0.64ZZ	0.16Zz
0.2z	0.16Zz	0.04zz

In 100 individuals of the offspring, F(Z)=(64x2+16+16)/200=160/200=0.8. The frequency is unchanged. This is Hardy-Weinberg equilibrium.

In More Abstract Terms:

- Let the frequency of one of the alleles (Z) represented by “p”. If there are two alleles for this trait, then the frequency of the other allele (z) must be 1-p, or “q”.
- Now let’s mate the population randomly:

	p	q
p	p^2	pq
q	pq	q^2

Frequency of homozygotes for one trait: p^2
Frequency of homozygotes for the other: q^2
Frequency of heterozygotes: 2pq
$P^2 + 2pq + q^2 = 1$

What about the next generation?

- **The ZZ individuals (p^2 of them) will contribute a p^2 proportion of the gametes as Z.**
- **The Zz individuals (2pq of them) will contribute a portion pq of the gametes as Z**
- **Thus the frequency of the Z gametes in the gamete pool is $p^2+pq = p(p+q)=p(1)=p$**
- **And similarly one finds that the frequency of z is q.**

Which is exactly what it was in the parental generation!!

In short, the HWE is a *"null hypothesis"* in that it will hold true *if and only if nothing interferes with the random exchange of genes*. The interesting cases are when the HWE proves untrue; then you need to explain <u>why</u>.

H-W Equilibrium and Evolution

- By definition, a population in Hardy-Weinberg equilibrium shows no changes in allele frequency.
- But by definition, evolution is a change in gene or allele frequency.
- Hence, a population in Hardy-Weinberg equilibrium is *not* evolving.

Thus, it’s the *exceptions* that are most interesting!!

- Evolution is technically defined as a change in the allele frequency of a population.

Forces that affect the HW equilibrium

Mutation

Founder principle

Gene flow

Immigration

Genetic drift

Kin selection

Natural selection

emigration

altruism

Forces affecting allele frequencies

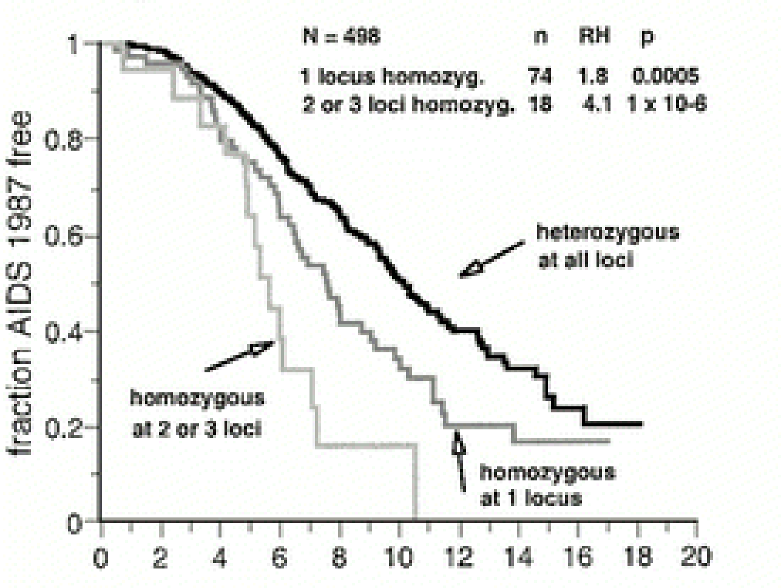
- **Natural Selection**:
  - **One phenotype has some advantage over another. This means that one phenotype will produce more viable, fertile offspring.**
  - **Natural selection could occur at any point from acquisition of mates to survivability to sexual maturity of the offspring.**
  - **Forces of natural selection include all those factors present in the environment, physical as well as biological (inter- and intraspecies competition).**

Forces (cont'd)

Heterozygote advantage:

- In humans, heterozygosity for sickle cell anemia acts to protect against sleeping sickness. In areas infested with tsetse flies this is an advantage and will increase the number of deleterious (recessive) alleles.

- Heterozygosity at the MHC loci seems to prolong succumbing to HIV AIDS:



Time since seroconversion (yrs)

Forces (cont'd)

- Kin Selection
  - Selection force expressed in helping the reproductive success of your kin because they share your alleles. Powerful force in colonial organisms such as ants and bees. Also in pack animals such as wolves who give up mating privileges to protect the alpha male and female. Depends in part on the degree of shared alleles (e.g., parents and offspring).
- Altruism
  - Like kin selection in that an individual lowers its own reproductive fitness to enhance that of others; but in this case the sharing of alleles is less strong. Examples: buffalo herds; birds who put off reproducing to assist older birds in raising young.

Forces (cont’d)

- Inbreeding:
  - Most if not all members of a group carry recessive deleterious alleles (usually loss-of-function alleles) which are masked by a dominant allele. Inbreeding increases the risk of homozygosity of these recessive alleles, causing the deleterious trait to be expressed with consequent loss of the function of that gene.
  - The opposite is hybrid vigor.

Evolutionary Theory From Darwin to Today

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Marist College - Center for Lifetime Study

CLS

1992 - 2012

An example of interbreeding effect: mortality rates of children

		Children of 1st cousins	Children of nonrelatives
Deaths	Period	%	%
Children under 20 (U.S.)	18th-19th Century	17.0	12.0
Children under 10 (U.S.)	1920-1956	8.1	2.4
At or before birth (France)	1919-1950	9.3	3.9
Children under 1 (Japan)	1948-1954	5.8	3.5
Children 1 to 8 (Japan)	1948-1954	4.6	1.5

Inbreeding in humans: degrees of relatedness

Relationship	Average % DNA Shared	Range
Identical Twin	100%	N/A
Parent / Child	50%*	N/A
Full Sibling	50%	38% - 61%
Grandparent / Grandchild Aunt/Uncle Niece/Nephew Half Sibling	25%	17% - 34%
1st Cousin Great-grandparent Great-grandchild Great Uncle/Aunt Great Nephew/Niece	12.50%	4% - 23%
1st Cousin once removed Half first cousin	6.25%	2% - 11.5%
2nd Cousin	3.13%	2% - 6%
2nd Cousin once removed Half first cousin	1.50%	0.6% - 2.5%
Half second cousin		
3rd Cousin	0.78%	0% - 2.2%
4th Cousin	0.20%	0% - 0.8%
5th Cousin to Distant Cousin	0.05%	Variable

*47% in father-son relationships

Random (non-directional) forces affecting allele frequencies

- Migration: movement into or out of the population
  - Immigration: influx into a population of individuals from a different subgroup having different allele frequencies.
  - Emigration: exodus of a *non-random* or *non-representative* sampling of individuals, perhaps based on some genetic trait (explorers??) A corollary is the *Founder Effect*: the founders of a new subgroup are not a random sample of the original population.
- Genetic Drift: any change in the allele frequencies due purely to chance
  - E.g., wiping out of a random sample of the population because of a storm, meteor strike, etc.
  - In the case of genetic drift, survival is not based on genotype.
  - Genetic drift is more pronounced in small populations.
  - The "bottleneck effect" is the genetic drift equivalent of the founder principle.

Forces Affecting Allele Frequency: A summary

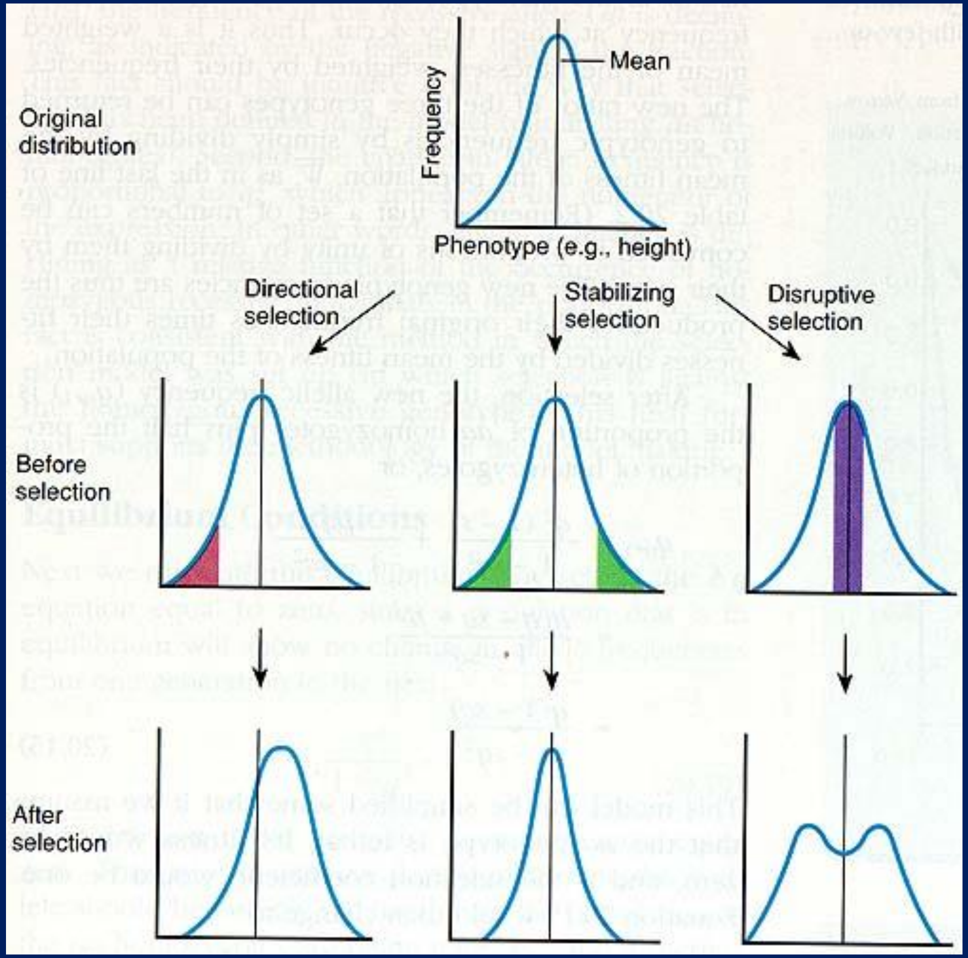
- Mutation
  - the ultimate source of all new alleles
- Natural Selection
  - Environmental (including other organisms of the same or different species)
- Genetic Drift
  - Immigration/ emigration
  - Founder Principle
  - Gene Flow

Of these three, only Natural Selection is directional. The others are random.

Allele Frequencies: Blood Type MN

Location	Percent of Population			Allele Frequency	
	MM	MN	NN	p	q
Greenland	83.5	15.6	0.9	0.92	0.08
Iceland	31.2	51.5	17.3	0.57	0.43
Australia	2.4	30.4	67.2	0.18	0.82
Egypt	27.8	48.9	23.3	0.52	0.48
Germany	29.7	50.7	19.6	0.55	0.45
China	33.2	48.6	13.2	0.58	0.42
Nigeria	30.4	49.5	20.4	0.55	0.45

Patterns of Natural selection



Coevolution: mutual selection and the "Red Queen Hypothesis"

(Lewis Carroll's Red Queen: "It takes all the running you can do, to keep in the same place")

- Predator/Prey: an arms race
  - Prey: camouflage, speed, wariness, herding, etc.
  - Predator: speed, intelligence, packs, etc.
- Parasite/host (incl. bacteria and viruses)
  - Host: immune response
  - Parasite: cell-surface proteins, "hiding" inside cells

- Plant-animal interactions
  - Plant
    - attract pollinators: skunk cabbage, orchids, etc.
    - Employ animals as seed carriers: berries, burrs, etc.
    - Repel consumers: toxins, spines, etc.
  - Animal
    - Adaptations for getting nectar, etc.
    - Resistance to toxins

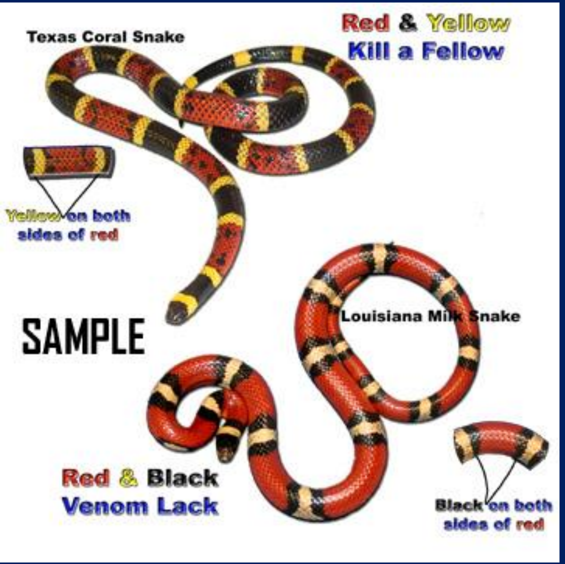
A unique form of coevolution: sexual selection

- Occurs when individuals in the population differ in their ability to attract mates.
  - Acts more strongly on males due to the amount of parental investment females make in the offspring. Leads to sexual dimorphism, most pronounced in species where the male has little role in raising the offspring. ("Sperm is cheap; eggs are expensive.")

Coevolution: predation and camouflage

- **Aposematic coloration**
  - **I can hurt you!!**
  - **I taste bad.**

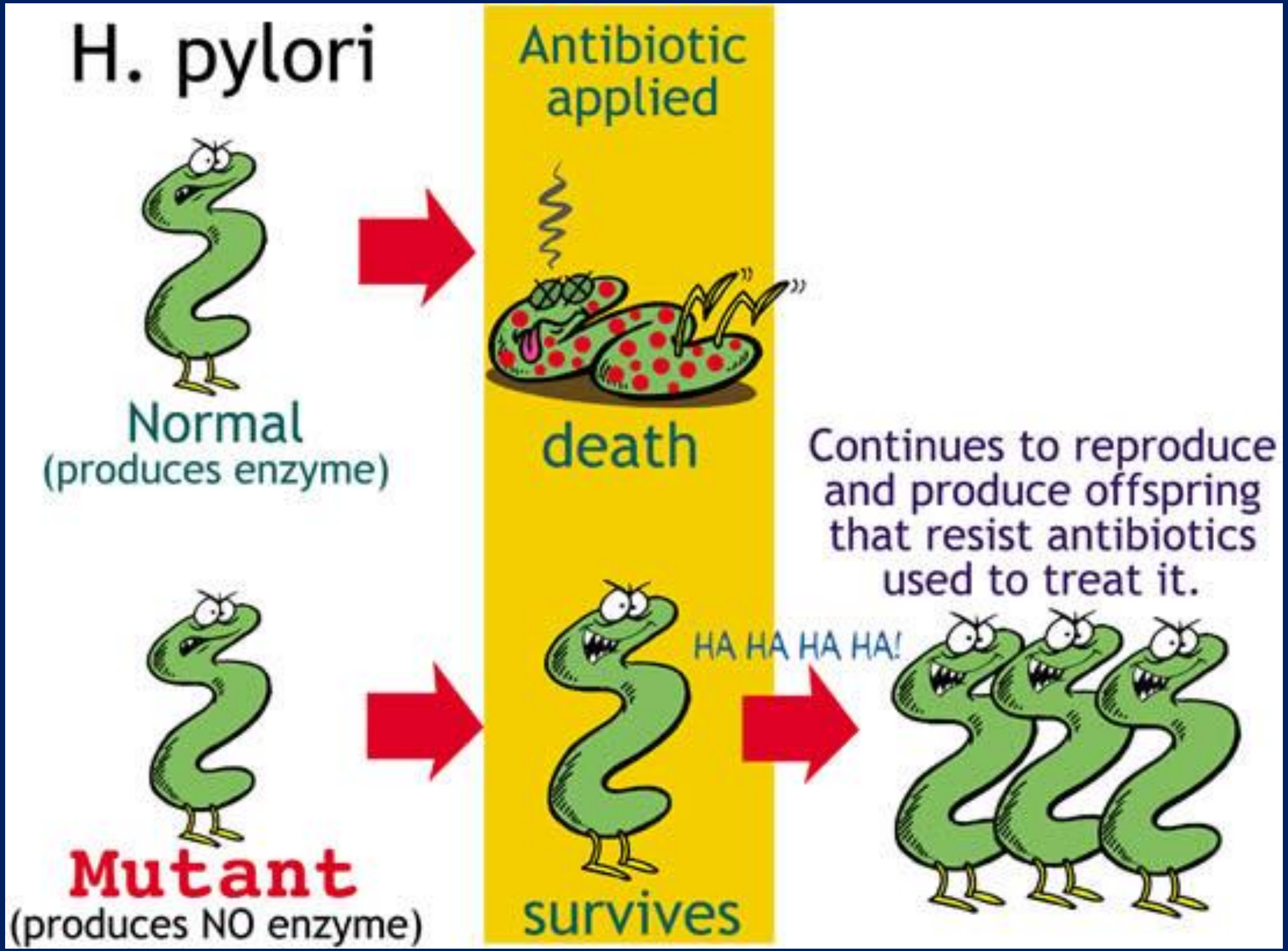
- **Batesian mimicry**
  - **I look *just like* that one that tasted so bad or hurt you!!**



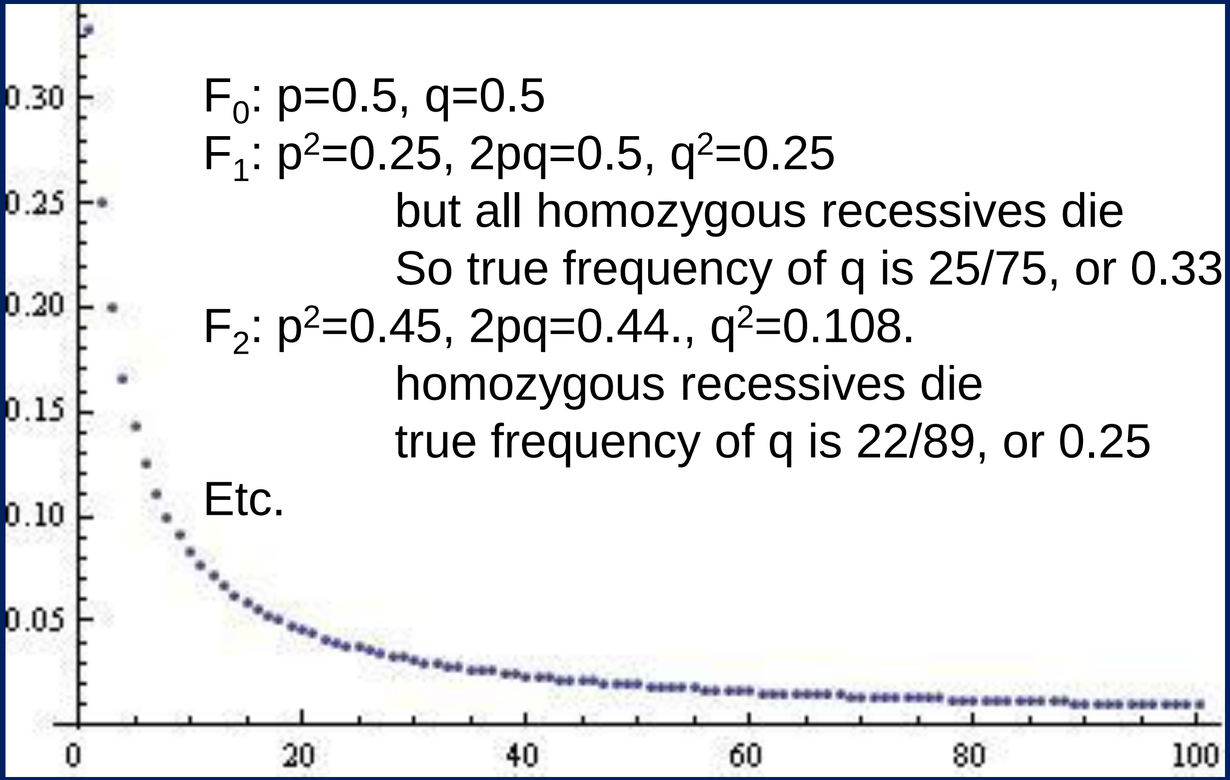
Coevolution (cont'd)

- Commensalism and Symbiosis (not parasitism)
  - Two (or more) different species living in a mutually beneficial relationship
  - Can be heavily mutually dependent leading to strong selection pressure
    - Human gut bacteria
    - Zooxanthelae of corals
  - Can also be less heavily mutually dependent
    - Cleaner shrimp in coral reefs
  - Can also be one-way, with selection pressure primarily on one organsim
    - Remora and sharks

“Natural” selection and antibiotics



Protection of recessive allele



F_0: p=0.5, q=0.5

F_1: p^2=0.25, 2pq=0.5, q^2=0.25

but all homozygous recessives die

So true frequency of q is 25/75, or 0.33

F_2: p^2=0.45, 2pq=0.44., q^2=0.108.

homozygous recessives die

true frequency of q is 22/89, or 0.25

Etc.

F(a)

Generations

Genetics and Inheritance

Genotype to phenotype connections

- Genotype-environment interactions.
  - The final phenotypic expression of a genotype is often determined by environmental factors. An example might be height in humans.
- Gene-gene interactions (epistasis).
  - The phenotypic expression of a gene is often influenced by the presence of other genes. An example might be albinism in humans. Sometimes called "penetrance".
- Incomplete dominance.
  - The phenotype of Aa might produce a different phenotype than AA (or aa), if each allele codes for a different protein, for example.

Additive effects

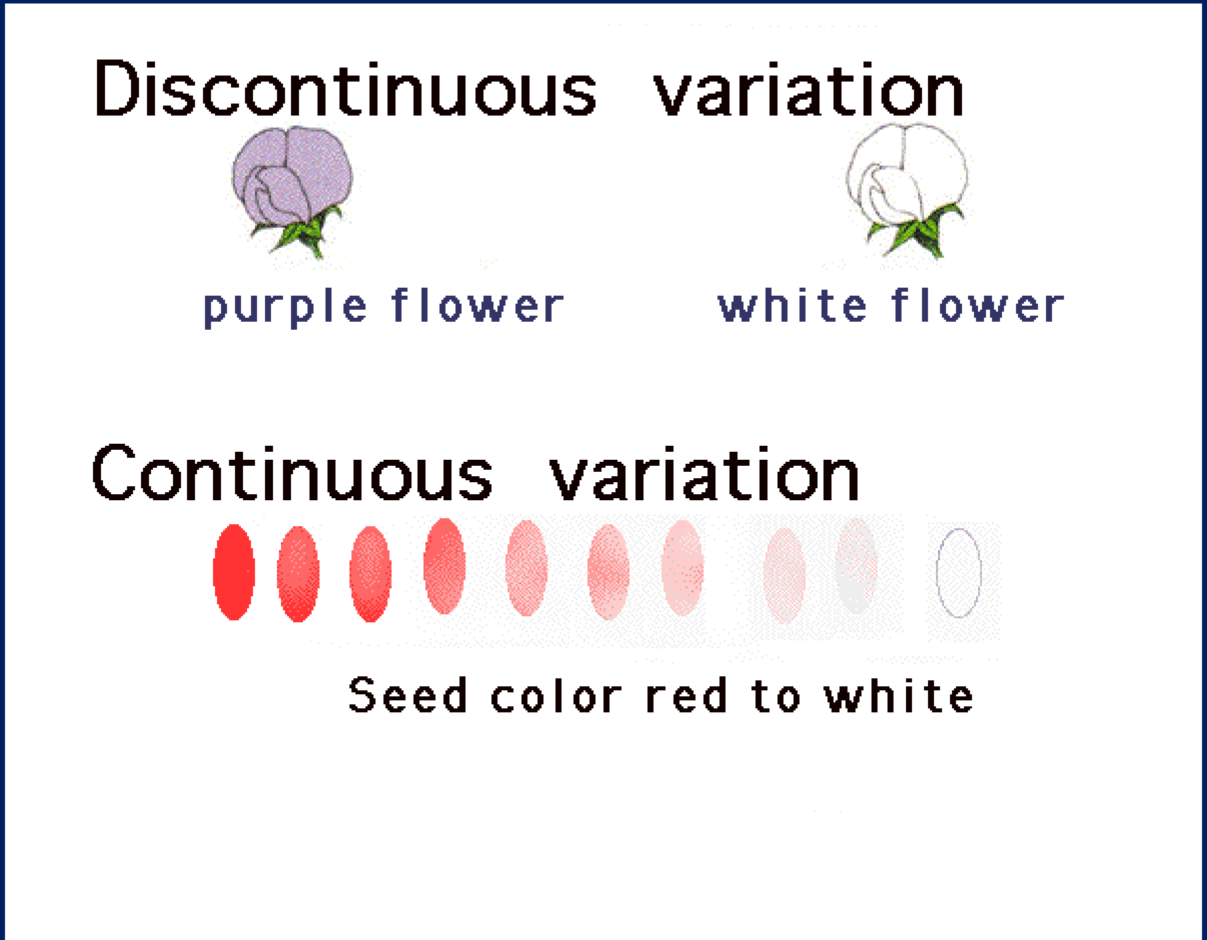
- If a phenotypic trait is controlled by multiple alleles (for example, height or skin color in humans), then the actual phenotype is a sum of the contributions of all of the alleles of all of the genes.
- Epistasis and dominance reduce the additive effect in that they produce fewer phenotypes for a given range of genotypes.

sperm

eggs

Gametes	ABC	ABc	AbC	Abc	aBC	aBc	abC	abc
ABC	6	5	5	4	5	4	4	3
ABc	5	4	4	3	4	3	3	2
AbC	5	4	4	3	4	3	3	2
Abc	4	3	3	2	3	2	2	1
aBC	5	4	4	3	4	3	3	2
aBc	4	3	3	2	3	2	2	1
abC	4	3	3	2	3	2	2	1
abc	3	2	2	1	2	1	1	0

Additive effects vs dominance



- Natural selection works ONLY on the phenotype which is exposed to the environment, NOT on the genes themselves!!

Polymorphism and multiple alleles

- There are only two alleles for a given gene present in diploid cells.
- However some genes have more than two possible alleles (multiple allelism).
- If the different combinations produce different phenotypes the population will be polymorphic.

Polymorphism:more than two possible alleles

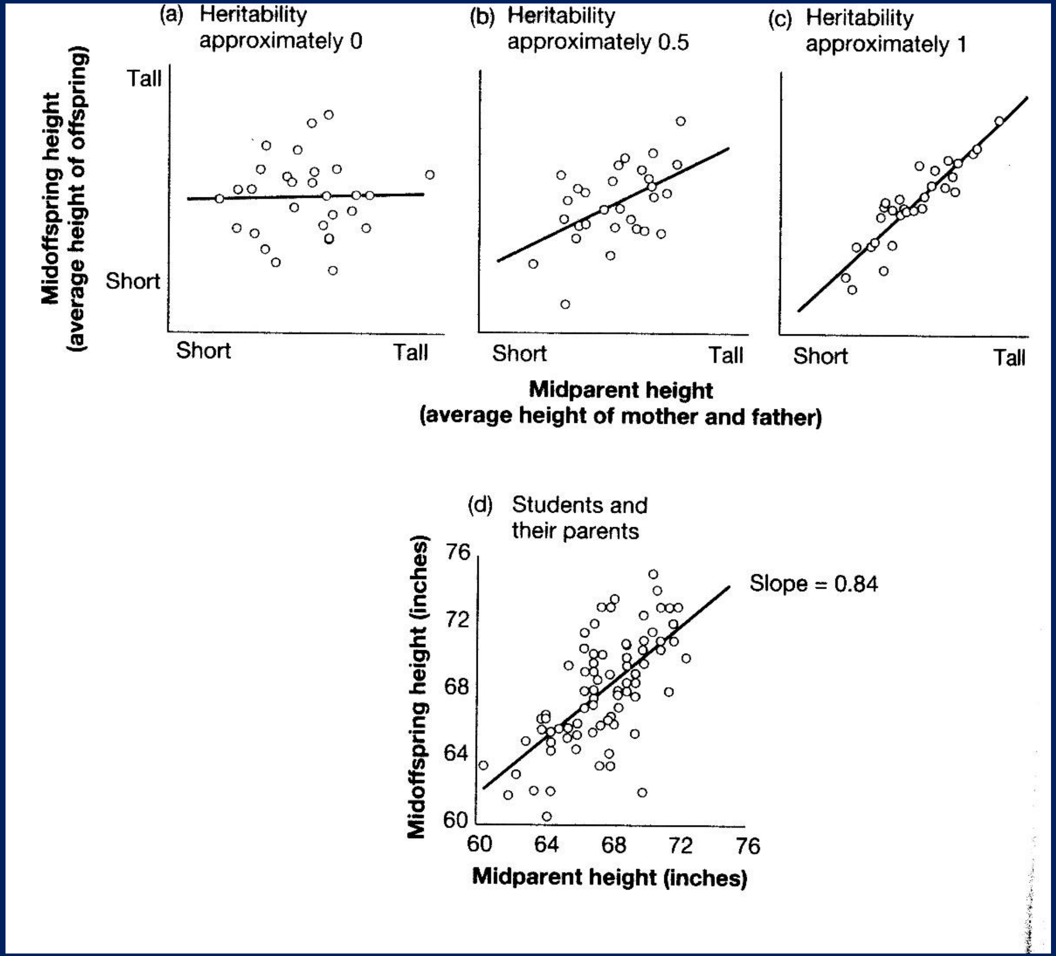
Landsteiner Blood Types

Here, there are THREE alleles in the population:
I^A, I^B, and i.
Each individual in the population will have only *two* of the alleles
Which two alleles an individual has will determine their phenotype.
Because of codominance (I^A and I^B), four phenotypes are produced.

	I^A	I^B	i
I^A	I^AI^A	I^AI^B	I^Ai
I^B	I^AI^B	I^BI^B	I^Bi
i	I^Ai	I^Bi	ii

A B AB O

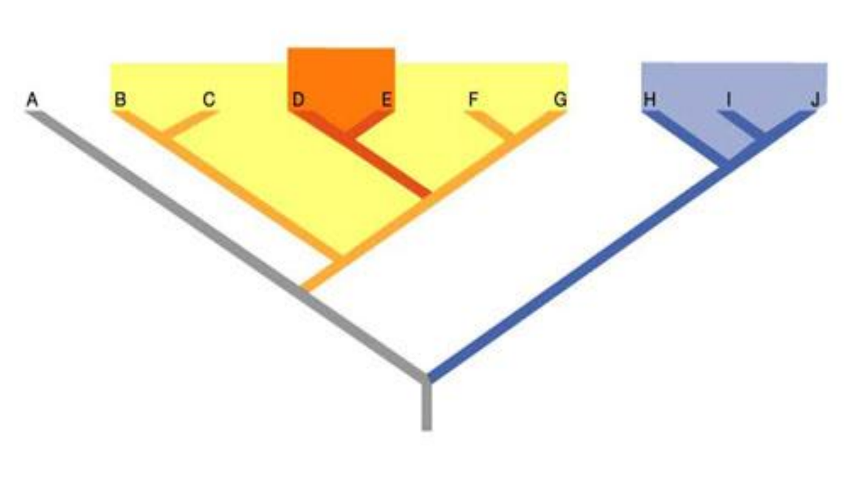
Heritability as the slope of a regression line



- Natural selection works ONLY on heritable characteristics!!

Speciation

- What is a species?
  - —Biological species: members of separate biological species cannot mate and/or produce viable, fertile offspring (e.g., horses and donkeys); reproductive isolation.
  - Morphospecies: morphologically dissimilar groups. Useful for asexual organisms, but subject to mistakes.
  - Phylogenetic species: a group ("clade") consisting of the common ancestor and all its descendants and *only* those descendants.



Speciation
(a.k.a. The Origin of Species)

- All populations when spread across a habitat area ("range") clump into subgroups called "demes" due to the patchiness of the habitat.
- Organisms (plants, animals, fungi, etc.) tend to exchange gametes more commonly within a deme than between demes.
- Thus, over time, gene frequencies differ from deme to deme, even within the same species, due to the forces of natural selection, genetic drift, etc. .

This leads to phenotypic variation across the range of the species.

Example: a "cline" such as with N. American wolves

Speciation (cont’d)

- The process of differentiation proceeds so long as and at a rate influenced by the degree of restriction of “gene flow” (mating) between demes.
- If it proceeds to the point where individuals from the separate demes can no longer interbreed and produce viable, fertile offspring a new species is born.

- Evolution works only on individuals. That is, the unit of natural selection is the individual, not the species or any higher taxon.

Speciation: isolating mechanisms

- Allopatric speciation: physical separation of demes
  - Continental drift; sea-level change (e.g., Bering Strait bridge); glaciation; migration over inhospitable intervening terrain (e.g., Galapagos finches; Australia; lakes; elevation); etc.
- Sympatric speciation: without physical separation but partially restricted gene flow between demes
  - e.g., patchy distribution over the range; sex selection; diurnal cycles; fertility cycles; prey/host preferences; etc.

The bottom line

- Once the restriction of gene flow allows different demes to so change that some biological mechanism prevents them from mating to produce viable, fertile offspring even should they meet and mingle, they are now separate species.
- Henceforth the forces of natural selection (as well as genetic drift, migration, etc.) will act independently in the two species, and they will drift further and further apart morphologically, behaviorally, etc.

Speciation: Restriction Hierarchy

Two sexually reproducing populations become separate biological species when they can no longer produce viable, fertile offspring.

- Partners don't meet
  - Behavioral (Circadian rhythms)
  - Physical (insect plant preference; host preference)
- Mates meet but don't exchange gametes
  - Behavioral (mate preference, mating rituals)
  - Physical isolation (incompatible reproductive organs)
- Partners can mate but no zygote forms
  - incompatible cell surface proteins, poor sperm survival
- Zygote forms but embryo aborts
  - spontaneous abortion; incompatible genes; chromosome number, etc.
- Embryo goes to term but dies before maturity
- Offspring mature but are infertile.
  - Poor survival, unable to mate with either parent group.

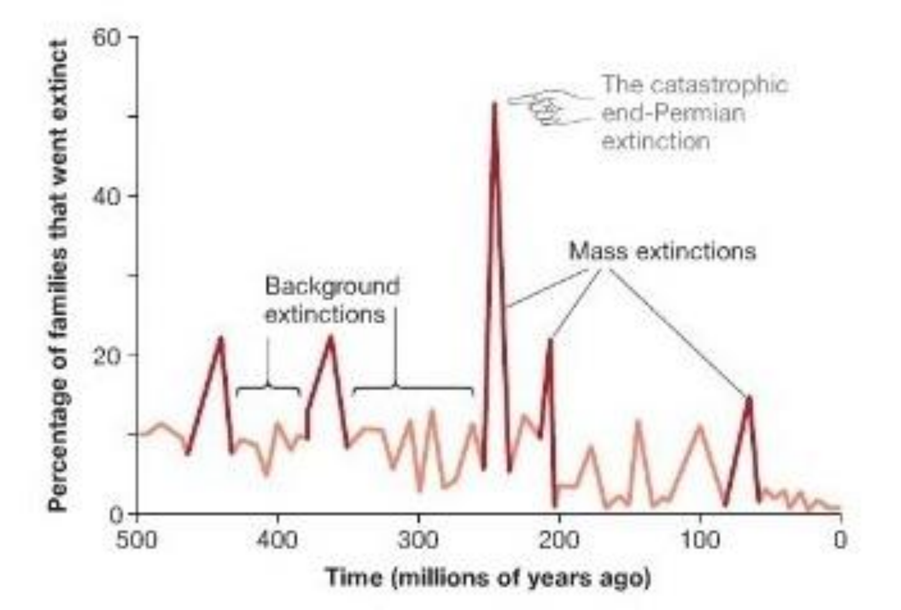
Adaptive Radiation

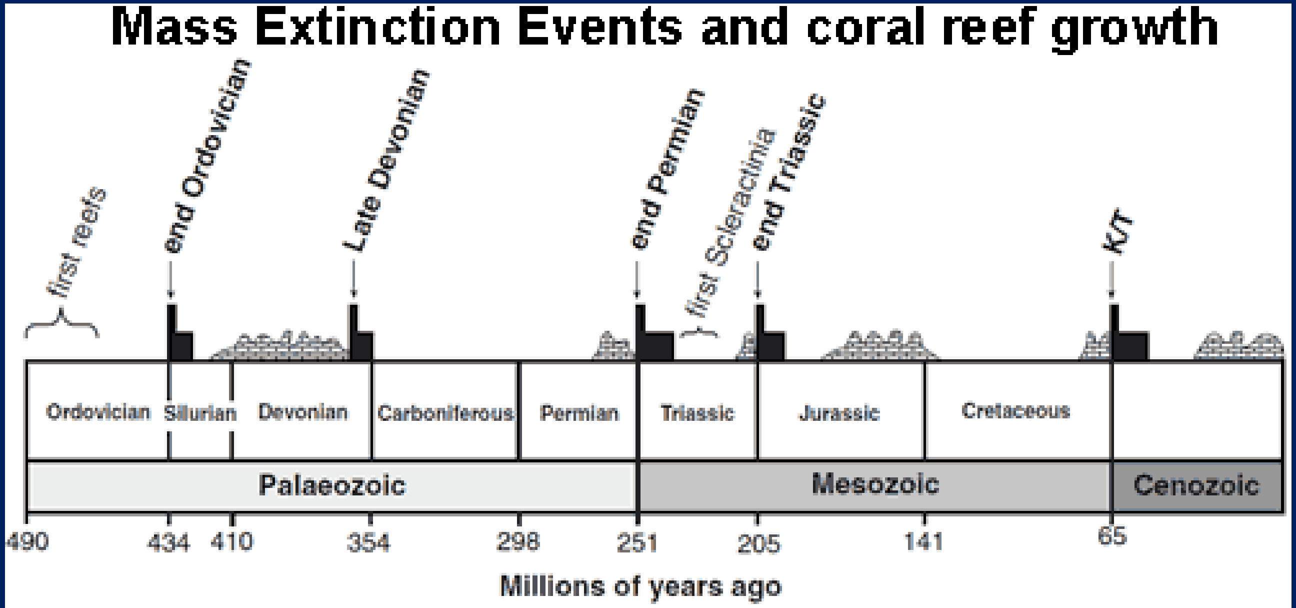
- ***Adaptive radiation*** is an evolutionary term to describe the (usually) rapid evolution in a species and/or speciation that occurs upon entering a new environment in which there are many unoccupied *niches*.

  This might occur after a major upheaval such as the melting of glaciers, an asteroid strike, etc. which creates new niches or removes competitor species from old niches.

  Another way it can occur is by a species acquiring the ability to enter a hitherto unoccupied or sparsely occupied environment. Examples would be plants or animals moving from sea to land, or animals (insects, reptiles, mammals) developing wings.

Mass Extinctions lead to adaptive radiation





Two levels of evolution

- **Microevolution:** "All events and processes that occur at or below the level of the species, such as the variability of populations. Adaptive changes in populations, geographic variation and speciation[1]."
- **Macroevolution:** "Processes that occur above the species level, particularly the origin of new higher taxa, the invasion of new adaptive zones, and, correlated with it, often the acquisition of evolutionary novelties such as the wings of birds or the terrestrial adaptations of the tetrapods or warm-bloodedness in birds and mammals[1]."

[1]Ernst Mayr, "What Evolution Is". Perseus Books, 2001

Two Theories on the Pattern of Evolution: Data

- Observation: The fossil record shows long periods in which essentially the same flora and fauna persisted with only minor changes, interspersed with relatively short intervals in which major changes such as extinctions and the arising of whole new taxa.

Two interpretations of the data

- **Gradualism** (Ernst Mayr): evolution is necessarily gradual in large groups, where gene flow rapidly dilutes out small changes; evolution primarily occurs in smaller satellite groups whose rarity biases the fossil record. More rapid evolution only occurs with drastic changes such as climatological shifts, asteroid strikes, etc. that close and open many different ecological niches.
- **Punctuated Equilibrium** (Steven Jay Gould): once adapted, species are stable for long periods, slowly accumulating small, virtually unnoticeable changes. However, at some critical tipping point, the species will undergo very rapid evolution and speciation.

Rates of evolution

- Fact: Some groups and species of organisms seem to show little or no change over many millennia while others change rapidly. Yet environments have changed greatly over this period, so it can’t be some “perfect adaptation”.
  - North American and Asian skunk cabbage groups have been separated for 6-8 million years yet are morphologically indistinguishable and can readily cross-pollinate.
  - Horseshoe crabs: unchanged since the Triassic (200 MYA)
  - Lampshell: unchanged since the Silurian (400 MYA)
  - Gingko tree: unchanged since the Jurassic (180 MYA)
  - Horsetail (Equisetum): unchanged since the mid-Permian (250 MYA)
- Why? Structure of the genome (balance)? Phyletic history (exoskeleton vs. endoskeleton)?