Lecture 13
BICD 100

- Class business
- Genetic variation exists: how to uncover?
  - microevolution: within species
- Hardy-Weinberg principle
  - determining if populations are in equilibrium
  - when H-W doesn't apply…
- Macroevolution
  - speciation
- Tracking evolutionary history of species

Class Business

- Only a few lectures left.
- We have already covered the syllabus!
- I've chosen topics
  to give you most useful experience

Upcoming Lectures

- 13 Evolution and Population Genetics
  - Klug chapter 23
- 14 Bacterial Genetics
  - Klug chapter 8
- 15 Viruses & Bacterial Gene Swapping
  - Klug chapter 8
- 16 Cancer Genetics
  - Klug chapter 16
- 17 To be announced

Upcoming Lectures

- The goal is to present to you
  useful, relevant information
  for your studies and life.

Lecture 13
Population & Evolutionary Genetics: Chapter 23 in Klug

- Class business
- Genetic variation exists: how to uncover?
  - microevolution: within species
- Hardy-Weinberg principle
  - determining if populations are in equilibrium
  - when H-W doesn't apply…
- Macroevolution
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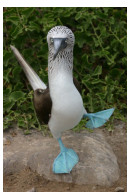
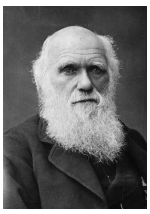
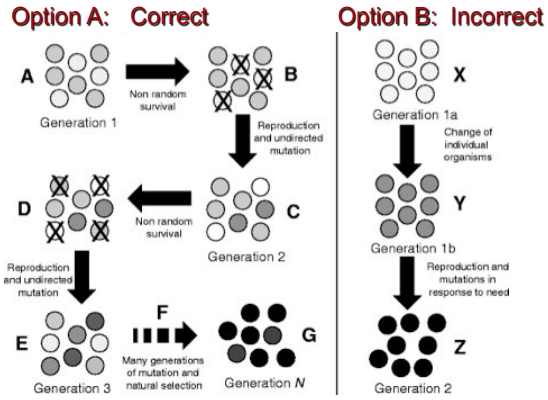


Evolution, Natural Selection & Population Genetics



- Darwin and the Galapagos Islands
- Natural selection is the mechanism for evolution
  - there is phenotypic variation
  - variation is heritable
  - there is competition among individuals and limiting resources
  - individuals with adaptive phenotypes will survive and have more offspring

How does evolution and natural selection work?



Evolution before our eyes

- Changes in influenza virus
  - new vaccine every year
- Bacteria evolve resistance to antibiotics

Defending the theory of evolution

- Hypothesis vs. theory
- The validity of a theory rests upon its ability to explain phenomena and predict outcomes. Theories may be s modified, based or Gravitational theor explain the nature theory explains the

Evangelical Scientists Refute Gravity With New 'Intelligent Falling' Theory

AUGUST 17, 2005 | ISSUE 41-33

KANSAS CITY, KS—As the debate over the teaching of evolution in public schools continues, a new controversy over the science curriculum arose Monday in this embattled Midwestern state. Scientists from the Evangelical Center For Faith-Based Reasoning are now asserting that the long-held "theory of gravity" is flawed, and they have responded to it with a new theory of Intelligent Falling.

Enlarge Image

Rev. Gabriel Burdett explains Intelligent Falling.

"Things fall not because they are acted upon by some gravitational force, but because a higher intelligence, 'God' if you will, is pushing them down," said Gabriel Burdett, who holds degrees in education, applied Scripture, and physics from Oral Roberts University.

Burdett added: "Gravity—which is taught to our children as a law—is founded on great gaps in understanding. The laws predict the mutual force between all bodies of mass, but they cannot explain that force. Isaac Newton himself said, 'I suspect that my theories may all depend upon a force for which philosophers have searched all of nature in vain.' Of course, he is alluding to a higher power."

Founded in 1987, the ECFR is the world's leading institution of evangelical physics, a branch of physics based on literal interpretation of the Bible.

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Defending the theory of evolution

- Hypothesis vs. theory
- Ken Miller of Brown University defends evolution
  - Intelligent design on trial – Nova special:
    http://www.pbs.org/wgbh/nova/evolution/intelligent-design-trial.html

- Clip from the Trial
http://www.youtube.com/watch?v=zi8FfMBYCkk

In Defense of Evolution

Posted 10.01.07 | NOVA

Dr. Kenneth Miller is as familiar as anyone in the scientific community with the intelligent-design movement and its attempts to undermine the theory of evolution. A professor of biology at Brown University and coauthor (with Joe Levine) of the standard high-school textbook *Biology*, Miller testified at the Dover trial as an expert witness for the plaintiffs, the Dover parents who brought suit against their town's school board. Here, Miller, who stresses that he is also a man of faith, talks about why evolution matters, what flaws he sees in the intelligent-design argument, and why the Dover decision hardly means the end of the controversy.

The intelligent-design movement, says Ken Miller, "is basically designed to bring the supernatural into science. And that kind of introduction would destroy both science and religion." Enlarge

Photo credit: Courtesy Ken Miller

Natural selection with molecular info

1. Random mutations cause variations in DNA sequence.
2. The mutated alleles may be beneficial or detrimental (or neither) to the individual's survival.
3. Beneficial mutated alleles are more likely to be passed on to subsequent generations.
4. Over many generations, beneficial alleles increase because of the increased survival and reproduction of organisms carrying these alleles.

What natural selection does not do

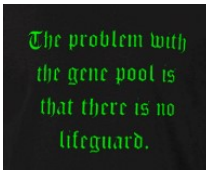
- select for perfect organisms

Natural selection does not grant organisms what they "need".

Selects for best-adapted individuals among diversity in population that ALREADY EXISTS

Genetic variation: lifeblood of selection and evolution

- Gene pool: genetic variation contained in all individuals of a population



Diversity can be hidden: how to measure it?

Most populations contain a high degree of heterozygosity

Phenotypes of recessive alleles not seen

Alleles can be masked by epistasis

Methods for measuring genetic variation

- Use artificial selection to uncover diversity
  - e.g. dog breeding uncovered diversity in the wild wolf population

Methods for measuring genetic variation

- Use artificial selection to uncover diversity
  - e.g. dog breeding uncovered diversity in the wild wolf population

Another example is Drosophila in the lab:
researchers have selected for eyeless mutants, body color mutants, courtship mutants, etc

Methods for measuring genetic variation

- variations in nucleotide sequence
  - cystic fibrosis gene: CFTR
    - 1500 mutations identified

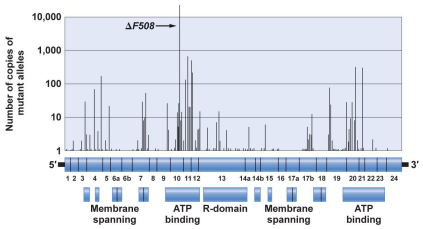


Figure 23-3

1 in 20 to 1 in 44 Europeans are heterozygous for these mutations

Most populations have an enormous reservoir of genetic diversity

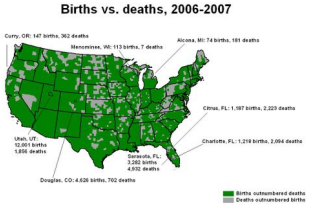
- Some is obvious, some needs to be uncovered

- What defines a species?

a group of organisms capable of interbreeding and producing fertile offspring of both sexes

Microevolution: diversity within a species

- Populations are dynamic: changes occur thru
  - birth, death, migration, contact w/others
  - allele and genotype frequencies change



- Microevolution: changes that do NOT result in reproductive isolation

Lecture 13
Population & Evolutionary Genetics: Chapter 23 in Klug

- Class business
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Hardy-Weinberg principle

- Relationship between relative proportion of alleles and frequencies of different genotypes

- Count the alleles
- Count the genotypes

Determine if population is in equilibrium from one generation to the next

Hardy-Weinberg principle

- Relationship between relative proportion of alleles and frequencies of different genotypes

Model makes 2 predictions:
1) frequencies do not change over time
2) with 2 alleles at a locus: *A* and *a*
after one generation of random mating, frequencies of genotypes *AA*:*Aa*:*aa* is $p^2 + 2pq + q^2 = 1$

p= frequency of *A*, q= frequency of *a*

Hardy-Weinberg principle

- calculating genotype freq. from allele freq.
- if you know frequency of allele A is 0.7, allele a is 0.3, can calculate the frequency of genotypes

applying allele freq to a Punnett square

Figure 23-4

		Sperm	
		p= fr(*A*) = 0.7	q= fr(*a*) = 0.3
Eggs	fr(*A*) = 0.7	fr(*AA*) = 0.7 × 0.7 = 0.49	fr(*Aa*) = 0.7 × 0.3 = 0.21
	fr(*a*) = 0.3	fr(*aA*) = 0.3 × 0.7 = 0.21	fr(*aa*) = 0.3 × 0.3 = 0.09

Hardy-Weinberg principle

- Makes several assumptions
  - no selection
  - no alleles are created or converted
  - no migration
  - population is infinitely large
  - population mates at random

Hardy-Weinberg allows us to see when these assumptions are NOT true, e.g. to see the effects of selection

Question: The Hardy–Weinberg law applies to populations with:

A) nonrandom mating.
B) selection acting on genotypes.
C) regular influx of immigrants.
D) a very large population size.
E) All of the above.

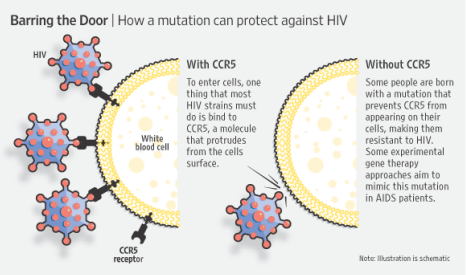
Question: The Hardy–Weinberg law applies to populations with:

Answer:
D. a very large population size.

Explanation:
It also applies to populations with random mating and no mutation, no migration, and no selection. It suggests that there will be no allele or genotype frequency changes in the population if these conditions are met.

Applying Hardy-Weinberg to human populations

- e.g. CCR5 gene and HIV/AIDS



Applying Hardy-Weinberg to human populations

- e.g. CCR5 gene and HIV/AIDS
  - mutation in CCR5 called Δ32
    - Δ32/ Δ32 homozygotes: resistant to HIV
    - Δ32/+ heterozygotes: partially resistant
    - +/+ susceptible to HIV

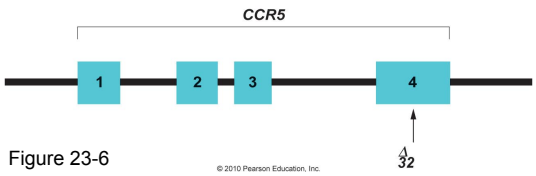
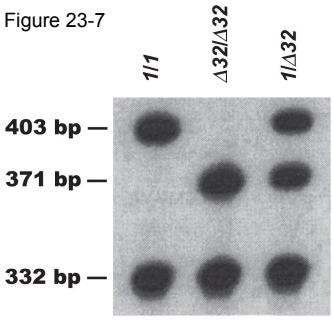


Figure 23-6

Measuring frequency of Δ32 CCR5 allele

Figure 23-7



PCR amplification of CCR5, followed by restriction digest and then gel electrophoresis

Testing for Hardy-Weinberg equilibrium with CCR5-Δ32 and CCR5-1 alleles

- First count alleles from genotyping data
  - calculate p=0.89 and q=0.11

TABLE 23.2 Methods of Determining Allele Frequencies from Data on Genotypes

A. Counting Alleles

Genotype	*1/1*	*1/Δ32*	*Δ32/32*	Total
Number of individuals	79	20	1	100
Number of *1* alleles	158	20	0	178
Number of *Δ32* alleles	0	20	2	22
Total number of alleles	158	40	2	200

Frequency of *CCR51* in sample: 178/200 = 0.89 = 89%
Frequency of *CCR5-Δ32* in sample: 22/200 = 0.11 = 11%

can also count from genotypes (see Table 23.2B)

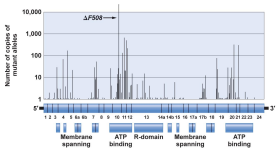
Testing for Hardy-Weinberg equilibrium with CCR5-Δ32 and CCR5-1 alleles

- Next, genotype 283 individuals
  - 223 are 1/1; 57 are Δ32/1; 3 are Δ32/Δ32
    - calculate CCR5-Δ32 allele frequency
      - [57 + 2(3)]/566 = 0.11
    - calculate CCR5-1 allele frequency
      - [57 + 2(223)]/566 = 0.89
- Use Hardy-Weinberg equation for genotypes
  - expected frequency of 1/1 is $(0.89)^2 = 0.792$
  - expected frequency of Δ32 /Δ32 is $(0.11)^2 = 0.012$
  - expected frequency of 1/Δ32 is $2(0.89)(0.11) = 0.196$
- No evidence for selection
  - can use chi squared test to quantitate

May take several generations to see effects of selection

Calculating heterozygote frequency

Figure 23-3



- Cystic fibrosis
  - mutations in CFTR
  - autosomal recessive disease
  - 0.0004 people are affected (Europeans)
    - frequency of recessive allele: q= 0.02
    - frequency of wt allele: p= 1-0.02 = 0.98
  - If in Hardy-Weinberg equilibrium
    - frequency of heterozygotes = 2pq = 0.04
      - 4% of people are carriers of this allele

Question: If a population in Hardy–Weinberg conditions has an ***aa*** genotype frequency of 0.16, what is the frequency of the ***a*** allele?

A) 0.4
B) 0.16
C) 0.32
D) 0.64
E) 0.8

Question: If a population in Hardy–Weinberg conditions has an ***aa*** genotype frequency of 0.16, what is the frequency of the ***a*** allele?

Answer:
A. 0.4

Explanation:
0.16 would be the value of q^2, and the value of q (the allele frequency) would be its square root, 0.4.

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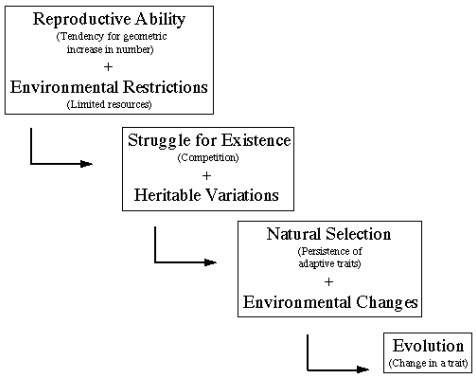
A population in Hardy-Weinberg equilibrium shows no change

- When the Hardy-Weinberg equilibrium fails to apply
  - natural selection
  - mutation
  - genetic drift
  - nonrandom mating

Natural selection as driving force for evolution

- there is phenotypic variation
- variation is heritable
- there is competition among individuals and limiting resources
- individuals with adaptive phenotypes will survive and have more offspring

Natural selection and evolution

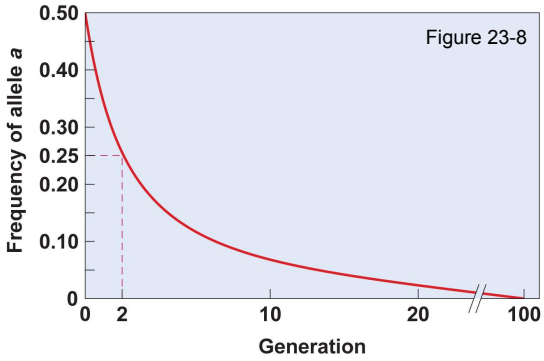


Natural selection can change allele frequencies in next generation

- Use Hardy-Weinberg to calculate this
- e.g. 100 individuals with freq p = 0.5 *A* alleles and q= 0.5 *a* alleles
  - frequencies of genotypes *AA:Aa:aa* is $p^2 + 2pq + q^2 = 1$
  - therefore, 25 *AA*, 50 *Aa*, 25 *aa*
- different survival/reproduction rates for each:
  - 100% for *AA* (25), 90% for *Aa*, (45) 80% for *aa* (20)
  - new gene pool: 2(25) + 2(45) + 2(20) = 180 gametes
  - freq of *A*: 25(2) + 45(1) = 95 *A* alleles; 95/180 = p = 0.53
  - freq of *a*: 20(2) + 45(1) = 85 *a* alleles: 85/180 = q= 0.47

Allele frequencies have changed.

Change in frequency of a lethal allele



Different types of selection

- Phenotype is the result of the combined influence of genotype and environment
- Directional selection
- Stabilizing selection
- Disruptive selection

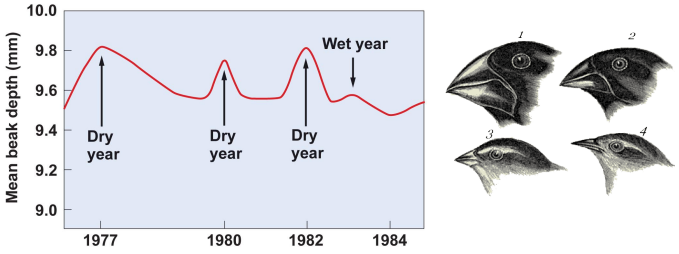
Different types of selection

- Phenotype is the result of the combined influence of genotype and environment
- Directional selection
  - phenotypes at one end of the spectrum are selected for or against

Different types of selection

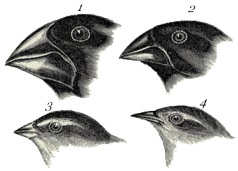
- Directional selection: Galapagos finches

Figure 23-10



In dry years select for bigger beaks

Evolution on the Galapagos Islands continues!



- In 1982 a larger species of finch moved into one of the Galapagos Islands
  - *G. magnirostris* is bigger than *G. fortis*
    - Can crack open & eat seeds 3X faster
- In 2003 & 2004 drought occurred reducing food supply: *G. fortris* w/large beaks died
- In 2006, reduction in the beak size of *G. fortris* was observed
  - can take advantage of new food sources
  - response to competition
- Example of evolution taking place before our eyes…

piquero de patas azules

Las Islas Galápagos

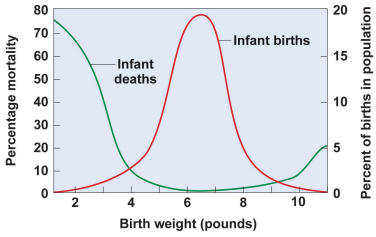
blue-footed boobies

Galápagos Islands

Different types of selection

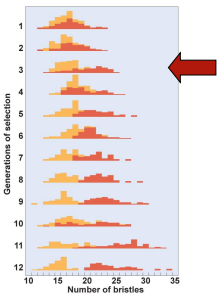
- Stabilizing selection
  - favor intermediate phenotypes
- Example: birth weight

Figure 23-11



Different types of selection

- Disruptive selection:
  - select against the intermediate phenotypes



Example: artificial selection for Drosophila bristle #

opposite of stabilizing selection

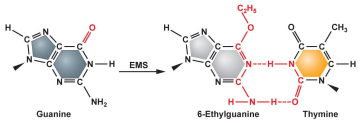
Figure 23-12

A population in Hardy-Weinberg equilibrium shows no change

- When the Hardy-Weinberg equilibrium fails to apply
  - natural selection
  - mutation
  - migration
  - genetic drift
  - nonrandom mating

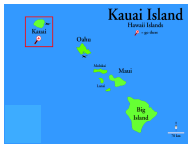
If mutation rate changes, H-W may no longer apply

- Mutations create new alleles in the gene pool
  - these can be selected for, selected against, or be neutral
- If *A* → *a* mutation rate changes, then alleles *A* and *a* may no longer be in H-W equilibrium



If migration and genetic drift occur, H-W may not longer apply

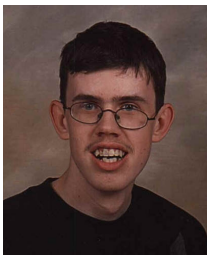
- Migration can alter allele frequencies



e.g. species found on both Kauai and Oahu, but with different allele frequencies at a locus

- Genetic Drift causes random changes in allele frequency in small populations
  - random fluctuations in allele frequency

If non-random mating occurs, H-W may no longer apply



Would you rather mate with this man?

Or this man?

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Changes leading to speciation

- Macroevolution: reproductive isolation

TABLE 23.4 Reproductive Isolating Mechanisms

Prezygotic Mechanisms **Prevent fertilization and zygote formation**
1. **Geographic or ecological:** The populations live in the same regions but occupy different habitats.
2. **Seasonal or temporal:** The populations live in the same regions but are sexually mature at different times.
3. **Behavioral** (only in animals): The populations are isolated by different and incompatible behavior before mating.
4. **Mechanical:** Cross-fertilization is prevented or restricted by differences in reproductive structures (genitalia in animals, flowers in plants).
5. **Physiological:** Gametes fail to survive in alien reproductive tracts.
Postzygotic Mechanisms **Fertilization takes place and hybrid zygotes are formed, but these are nonviable or give rise to weak or sterile hybrids.**
1. **Hybrid nonviability or weakness.**
2. **Developmental hybrid sterility:** Hybrids are sterile because gonads develop abnormally or meiosis breaks down before completion.
3. **Segregational hybrid sterility:** Hybrids are sterile because of abnormal segregation into gametes of whole chromosomes, chromosome segments, or combinations of genes.
4. **F_2 breakdown:** F_1 hybrids are normal, vigorous, and fertile, but the F_2 contains many weak or sterile individuals.

Source: From G. Ledyard Stebbins, *Processes of Organic Evolution,* 3rd ed., copyright 1977, p. 143. Reprinted by permission of Prentice Hall, Upper Saddle River, NJ.

Genetic changes can be used to track evolution

TABLE 23.5 Amino Acid Differences and Minimal Mutational Distances between Cytochrome *c* in Humans and Other Organisms

Organism	(a) Amino Acid Differences	(b) Minimal Mutational Distance
Human	0	0
Chimpanzee	0	0
Rhesus monkey	1	1
Rabbit	9	12
Pig	10	13
Dog	10	13
Horse	12	17
Penguin	11	18
Moth	24	36
Yeast	38	56

Source: From W.M. Fitch and E. Margoliash, Construction of phylogenetic trees, *Science* 155: 279–284, January 20, 1967. Copyright 1967 by the American Association for the Advancement of Science.

Genetic changes can be used to track evolution

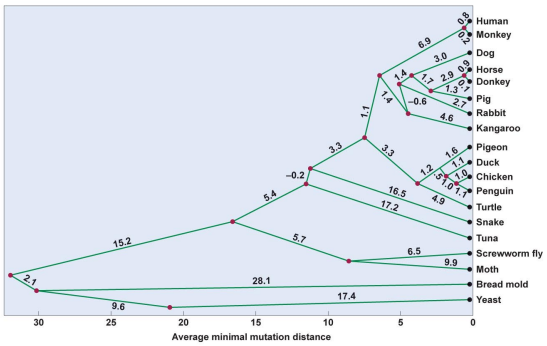

Figure 23-21

Neanderthals, humans, genomics…

- 30,000 to 300,000 years ago Neanderthals co-existed with Homo sapiens in Europe
  - then they died off
    - did we kill them? were they not well-adapted? did we interbreed with them?
- Are Neanderthals our ancestors?

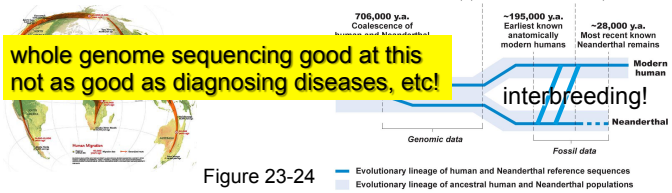

Figure 23-24

Neanderthals, humans, genomics…

- 30,000 to 300,000 years ago Neanderthals co-existed with Homo sapiens in Europe
  - then they died off
    - did we kill them? were they not well-adapted? did we interbreed with them?
- Are Neanderthals our ancestors?
  - Mitochondrial DNA analysis in 1997 suggested we diverged ~300,000 years ago
    - Nuclear DNA analysis in 2006 suggested similar conclusion
  - Nuclear DNA analysis with more data published in May 2010 reached different conclusion: we DID interbreed!
    - 1-4% of genes in non-Africans came from Neanderthals

Evolution of our understanding about human evolution!

Lecture 13
Population & Evolutionary Genetics:

- Genetic variation exists: how to uncover?
  - microevolution: diversity within species
- Hardy-Weinberg principle/equation
  - determining if populations are in equilibrium
  - if you know allele frequencies, you can solve for genotype frequencies, and if you know genotype frequencies, you can solve for allele frequencies. YOU WILL NEED TO BE ABLE TO DO THIS
- Macroevolution
  - speciation
- Tracking evolutionary history of species

- Next Lectures

- 14 Bacterial Genetics
  - Klug chapter 8
- 15 Viruses & Bacterial Gene Swapping
  - Klug chapter 8
- 16 Cancer Genetics
  - Klug chapter 16
- 17 To be announced

  and based on most useful and interesting topics