

# Bio Final Review

Daniel Filipski and Christopher Milan

May 30, 2018

## 1 General Information

For this, see the google doc.

## 2 Genetics

### 2.1 Sex-linked genes

These are genes located on the sex chromosomes. They will show different phenotype frequencies based on gender.

- (1) Gene A is on the X chromosome. A is the wild type, and  $\alpha$  is the diseased type.  
 $X^A X^\alpha \times X^A Y$ :

$$\begin{array}{c|c} X^A Y & X^\alpha Y \\ \hline X^A X^A & X^\alpha X^A \end{array}$$

### 2.2 Pedigrees

- = Unaffected Female
- = Affected Female
- = Unaffected Male
- = Affected Male

Connecting lines on pedigrees work just as they do on family trees. Relatively simple logic can be used to determine the genotypes of each member of the pedigree; however, some can be more difficult than others. My general method is to use the “method of staring” in the words of Mr. Letarte.

### 2.3 Genetic Disorders – Sickle Cell, Cystic Fibrosis, Huntington’s

#### Sickle Cell

- Red blood cells contain hemoglobin, which bind  $O_2$
- Hemoglobin is made up of two  $\alpha$ -globin and two  $\beta$ -globin polypeptides
- Mutation in  $\beta$ -globin makes it slightly less soluble
- When  $O_2$  is low, hemoglobin without  $O_2$  will start to clump and form long fibers that will change the shape of the red blood cell, which will then get stuck in capillaries
- If one is a heterozygote of this disease, they have an advantage against malarial
- Sickle Cell disease is recessive, because its effects are not great enough with only some of the  $\beta$ -globin broken.

#### Cystic Fibrosis

- In frame three base pair deletion in gene for CFTR
- CFTR is missing one amino acid (phenylalanine), which causes it to misfold and be destroyed

- CFTR is a channel in the epithelial cell membranes for  $\text{Cl}^-$
- Without CFTR, there is too much extracellular  $\text{Cl}^-$ , which makes the fluid outside the cell thicker
- Mucus clogs lungs and serves as a growth substance for *Pseudomonas aeruginosa*
- The allele for Cystic Fibrosis is recessive, as with one of the two CFTR, cells still have enough paths for  $\text{Cl}^-$

### **Huntington's Disease**

- Mutation is dominant, but the disease does not present itself until late 30's or early 40's
- Huntingtin gene expressed in nerve cells. Its developmental role in adults is unclear
- CAG (codes for glutamine)
- Wild type 6-35 repeats
- Diseased 36+ repeats
- Diseased protein forms aggregates in neurons, which lead to cell death.

## **2.4 Nondisjunction**

Nondisjunction Event – Failure to separate chromosomes

This is more common in meiosis I. Trisomy 21 causes down syndrome.

## **2.5 Recombinant DNA – Restriction Enzymes, Ligase, Electrophoresis, GFP, PCR, Selectable Markers, Screens, Plasmids, Transformations**

Recombinant DNA – Combination of two or more pieces of DNA to create an artificial construct.

Building Pieces of DNA:

1. Synthesize from scratch
2. Cut and Paste
  - Ligase is not specific and will join any two pieces of DNA.
  - Restriction enzymes originate from bacteria, where they served as a type of immune system.
  - Restriction System: methylase adds  $\text{CH}_3$
  - Restriction Enzyme cuts DNA if not regulated.

Eco RI:

- Sticky ends of *GAATTC* (a DNA reverse palindrome)
- Eco RI cuts between the G and the A

Plasmid:

- Mini chromosome in bacteria
- Must have an ori, a selectable marker
- Plasmids can be shared between cells. They also can be picked up from the environment, when they are there for whatever reason.
- An example of plasmids which can be shared between cells is antibiotic resistance.

DNA Sequencing:

- Denature DNA into single strands
- Add primer for only one strand

- Provide DNA polymerase and the four nucleotides, with a small fraction of the nucleotides modified so that DNA polymerase cannot extend from them (remove 3 hydroxyl)

GFP – Green Fluorescent Protein

GFP is used to see where or when certain promoters are expressed and to tag proteins.

PCR – Polymerase Chain Reaction

- Amplify a specific DNA Sequence
- NEED: Template DNA, Primers, DNA Polymerase, nucleotides (all four)
- Three steps
  1. Denature – Use heat to separate the helix
  2. Anneal – Lower the temperature, so the primers can bind
  3. Extension – Polymerase extends from the primers
  4. Repeat steps 1 - 3 around 30 or 40 times

## 2.6 Selective Breeding – Hybridization vs Inbreeding

Selective breeding – only allowing parents with certain characteristics to breed.

Hybridization:

- Crossing two organisms (typically plants) to get the best traits from both in a hybrid
- Can be different species.

Inbreeding:

- Continued breeding of individuals with similar characteristics
- Dramatically decreases genetic variation
- Increases prominence of some traits

## 3 Evolution

### 3.1 Darwin's Observations and Background Knowledge

Darwin made three key observations:

1. Species vary globally.
2. Species vary locally.
3. Species vary over time.

More specifically, there are the same types of animals in the same environments across the globe (ie. emu, ostrich, and reah) Also Darwin made the observation about the finches and stuff. That leads nicely into the next section, so read that.

### 3.2 Evolutionary Theory – Adaptations, Fitness, Natural Selection, etc.

Natural Selection – the process by which individuals with greater fitness survive and leave behind more offspring. This occurs whenever more individuals are born than can survive, there is heritable variation, and that variation leads to varying levels of fitness.

Adaptation – any heritable characteristic that increase an organism's chance to survive and reproduce.

Fitness – how well an organism can survive in its environment.

### 3.3 Evidence for Evolution

- Fossil Record – evidence for common decent
- Carbon Dating – the Earth is around 4.6 billion years old
- Homologous Structures – Structures that are shared by related species and were inherited from a common ancestor
- Vestigial Structures – Structures that are inherited from common ancestors and no longer used
- Analogous Structures – Structures that share a common function but not a common structure.
- Embryology (The Study of Embryo Development) – The early stages of development of embryos look very similar for vertebrates
- Genetic Code – all living things use DNA as the genetic material, transcribe RNA from DNA, and translate proteins from RNA. Triplet code is nearly universal.
- Homologous Proteins and RNA – Proteins and RNA that share similar structure and function.

### 3.4 Population Genetics – Hardy-Weinberg Equation and Conditions that disrupt equilibrium, selection on polygenic traits

Hardy-Weinberg Principle – allele frequencies remain constant unless one or more factors cause them to change.

Single gene trait with two alleles. Define  $p$  and  $q$  as the allele frequencies, with  $p$  as the dominant allele and  $q$  as the recessive allele.

Hardy-Weinberg Equation:  $p^2 + 2pq + q^2 = 1$  or  $p + q = 1$

$p^2$	$qp$
$pq$	$q^2$

Example: Mouse Populations

50% AA [BLACKMOUSE] 50 Mice

40% Aa [BLACKMOUSE] 40 Mice

10% aa [WHITEMOUSE] 10 Mice

$P(AA) = p^2 \Rightarrow f(A)^2 = 0.49$

$P(Aa) = 2pq = 0.42$

$P(a) = q^2$

Disruptions of Equilibrium:

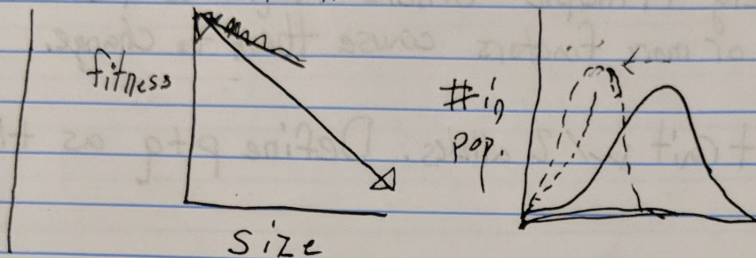
1. Natural Selection
2. Mutations
3. Migration (Immigration and Emigration)
4. Nonrandom Mating (Sexual Selection)
5. Small Population
  - Bottleneck Effect
  - Founder Effect

Polygenic selection on next page.

## Natural Selection on Polygenic Traits

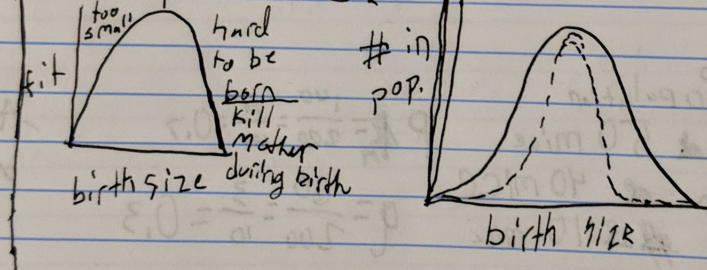
1. Directional - Curve shifts toward end with higher fitness

Ex: Size of Mice



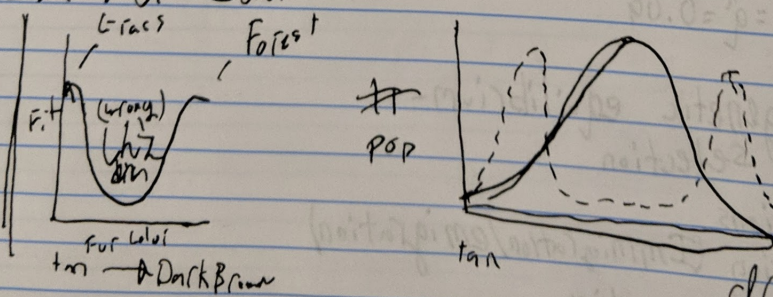
2. Stabilizing Selection - Individuals in the middle most fit, curve narrows

Ex: Pup Birth Size



3. Disruptive Selection - Extremes more fit than average, median selected against

Ex: Fur Color



## 3.5 Speciation

Species - Population that breeds together

Reproductive Isolation - Two populations can no longer interbreed and may evolve into two different species

Types of Isolation:

1. Geographic Isolation - Population physically separated
2. Behavioral Isolation - Differences in courtship rituals or methods
3. Temporal Isolation - Two populations do not mate at the same time

### 3.6 Molecular Clocks

Molecular Clock – Use mutation rates in DNA to estimate when two species diverged from a common ancestor

- Most mutations are neutral, which means they accumulate over time
- Some genes tolerate mutations better than others
- The greater the number of differences in the DNA sequences of two species, the more time has elapsed since sharing a common ancestor.

### 3.7 Binomial Nomenclature and Linnaean Classification

Systematics – Science of naming and grouping organisms into taxa that have biological meaning.

Binomial Nomenclature – two word science names

Linnaean Classification System:

- Domain
- Kingdom
- Phylum
- Class
- Order
- Family
- Genus
- Species

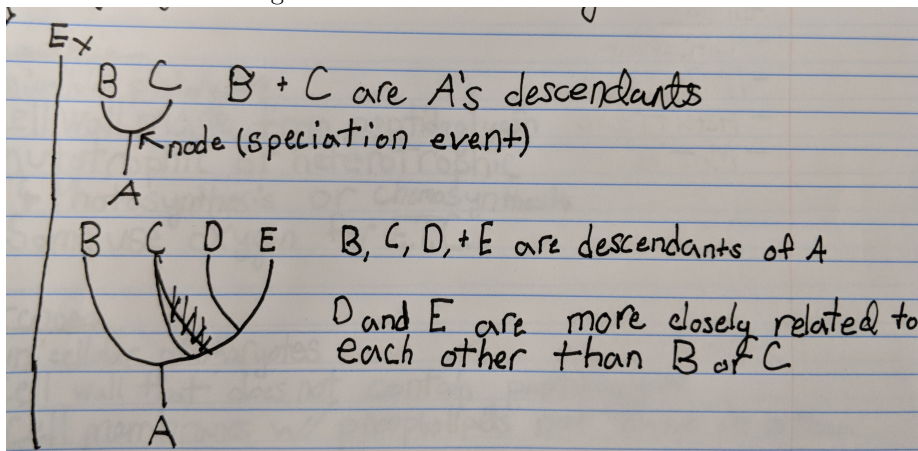
These can be remembered via the snazzy acronym **Katy Perry Claims That Orgasms Feel Good Sometimes**.

Domain	Kingdoms within
Bacteria	Eubacteria
Archaea	Archaea Bacteria
Eukarya	Protists, Fungi, Plants, Animals

### 3.8 Cladograms

Clade – a group of species that includes a single common ancestor and all descendants of that ancestor, living or dead

Monophyletic vs. Paraphyletic – paraphyletic groups are missing a common ancestor or contain multiple common ancestors or are missing descendants



Buildings Cladograms:

- derived characteristic – trait that arose in the most recent common ancestor of a lineage and was passed along to its descendants
- parsimony (Occam's Razor) – the simplest explanation is often correct.

### 3.9 Domains and Kingdoms

Bacteria:

- Unicellular Prokaryotes
- Cell wall made from peptidoglycan
- autotrophic (photosynthesis or chemosynthesis) or heterotrophic
- Some use oxygen for E.T.C.

Archaea:

- Unicellular Prokaryotes
- Cell wall that doesn't contain peptidoglycan
- Cell membranes with phospholipids not found in other domains
- Autotrophic (chemosynthesis) or heterotrophic
- Killed by O<sub>2</sub>

Protists

- Unicellular, Colonies and Multicellular (brown algae)
- At least five clades – one closely related to fungi, another to plants, and another to animals
- Some have cellulose cell walls
- Autotrophic (photosynthesis) or Heterotrophic

Fungi:

- Most multicellular, some unicellular
- Cell walls made from chitin
- Heterotrophs (detritivores or decomposers)

Plants:

- Most multicellular, some unicellular (green algae)
- Cell wall from cellulose
- Autotrophs (photosynthesis)
- nonmotile

Animals:

- Multicellular
- No cell wall
- Heterotrophs
- Motile

### 3.10 Fossil Record

- Most fossils preserved in sedimentary rock
- Made when organism falls in water and is buried in sand, silt, or mud. Then the sedimentary rock process happens. Shell, bones, and teeth remain if buried slowly. Soft tissue only remains if buried quickly.
- Dating Fossils:
  - Relative dating – use index fossils to establish a temporal sequence
  - Radiometric dating – use radioactive isotopes (they decay to get to stability)
    - \* Kinetics of Decay:
    - \*  $t = \text{time}$ ,  $\lambda = \text{rate constant}$ ,  $N_t = \text{number of nuclei at time } t$ ,  $N = \text{initial number of nuclei}$ ,  
 $t_{1/2} = \text{half life}$
    - \*  $\log_{10} N_t/N_o = -\lambda t$
    - \*  $t_{1/2} = 0.693/\lambda$

### 3.11 Origins of Life including Endosymbiont Theory

- All life came from one cell
- Prokaryote endosytosed another prokaryote
- Didn't break it down
- Mitochondria first
- Chloroplast
  - Evidence:
  - Small circular chromosome in Mitochondria and Chloroplasts
  - Double membrane surround those two
  - Custom ribosome
  - Reproduce separatly from the rest of the cell