Exam 3 Review

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Day 15

Terminology

Term	Definition
Gene	A segment of DNA that codes for a trait.
Genome	All your DNA.
Allele	The different variations of a gene.
Chromosome	“Chapter” in your DNA recipe book. A packet of DNA.
Homozygous	A **zygote** is an egg + sperm. Homozygous if the egg and sperm both brough the **same** allele.
Heterozygous	The egg and sperm brough **different** alleles to the table.
Genotype	What alleles you have.
Phenotype	How alleles express (what they look like).
Dominant/Recessive Inheritance	When one allele (dominant) completely masks the other allele (recessive).

Dominant alleles don’t take over the world because they are different frequencies. E.g. dwarfism, 6 fingers. Both of those are dominant but they are in low frequency. If you don’t have dwarfism or 6 fingers you are likely homozygous recessive.

Use the knowledge that for every gene, an individual possesses two alleles and that sometimes one allele

is dominant and one allele is recessive, to solve genetics problems.

click here for the section on problem solving.

What is a Punnett Square?

Hypothesize genetic explanations for inheritance patterns.

Analyze phenotypic and genotypic data and determine parent genotypes.

Predict offspring based on parental genotype and/or phenotype.

Use the terms homozygous, heterozygous, dominant, recessive, genotype, and phenotype correctly.

See the terminology table for Day 15 (today).

Describe human sex determination.

Day 16

Derive the Hardy-Weinberg equation.

Given a dominant allele R and a recessive allele r, we need two of either or each to define a geneotype. I.e. RR, Rr, rR, rr.

Assuming reproduction is random, let the probability of a dominant allele being drawn be p and the probability of a recessive allele being drawn be p. i.e. p and q are your **allele frequencies**. Then your **genotype frequencies** are described as such:

$$p + q = 1$$

$$\text{The probability of a getting a homozygous dominant individual is}$$
$$p * p = p^2$$

The probability of a getting a homozygous recessive individual is

$$q * q = q^2$$

The probability of a getting a heterozygous (and $\therefore$ dominant) individual is

$$pq \text{ or } qp = pq + qp = 2pq$$

The Hardy-Weinberg Equation describing the complete probability of all three possibilities (i.e. these values sum to 1)

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

Furthermore, your **phenotype frequencies** are described as:

Phenotype	Frequency
Dominant	$p^2 + 2pq$
Recessive	q^2

Recall: The phenotype is based off whether the trait shows or not. In other words, the dominant trait shows if you have the genotype RR, Rr, or rR. These are all separate genotypes, but they all result in the same *phenotype* (the display of the dominant trait).

Problem Solving Strategy

1. What have you been given?
2. What is being asked for?

The answer to both of these questions will be:

- A genotype?
- A phenotype?
- An allele?

Once you know the answer to these questions, use the following definitions and some basic algebra to solve the problem.

Expression / Equation	Definition
p	Dominant allele frequency.
q	Recessive allele frequency.
$p + q = 1$	The probability/frequency of dominant and recessive alleles sum to 1.
p^2	The frequency of a **homozygous dominant genotype**. e.g. RR.
q^2	The frequency of a **homozygous recessive genotype**. e.g. rr.

Expression / Equation	Definition
$2pq$	The frequency of a **heterozygous genotype**. e.g. Rr or rR.
$p^2 + 2pq + q^2 = 1$	The Hardy-Weinberg equation. States that the genotypic frequencies sum to 1.
$p^2 + 2pq$	The frequency of a dominant **phenotype**. e.g. if the creature has RR or Rr or rR, it will display a dominant trait and thus have a the dominant phenotype.
q^2	The frequency of a recessive **phenotype**. Notice that this is the same as the frequency of a *homozygous recessive genotype*, because the only way a recessive trait can show is if you have 2 recessive alleles making up the genotype.

Determine probabilities of each genotype and phenotype given the proportion of alleles in a population.

Determine allele frequencies given genotypes/phenotypes in a sample of individuals.

Determine the change in allele frequencies that will occur in a population over each generation.

Use the Hardy-Weinberg equation to determine allele frequencies in a population given a genotype and/or phenotype frequency and vice versa (assuming the population is in Hardy-Weinberg equilibrium).

Day 17

Terminology

Predict the outcome of the violation of each of the given assumptions of Hardy-Weinberg.

What does it mean to be in Hardy-Weinberg equilibrium?

The Hardy-Weinberg equation determines how alleles will be distributed using probability. When the HW equation is in equilibrium, it means that the population and its alleles and proportions of those alleles and phenotypes are **not changing**.

Predict which assumption has been violated given allelic frequencies of two populations.

For a population to be in Hardy-Weinberg equilibrium, there are a few requirements. i.e. we assume the following to be true when modeling a population after Hardy-Weinberg equilibrium.

1. No **natural selection** is occurring
2. No **gene flow** (immigration/emigration) is occurring
3. No **genetic drift** is occurring
4. Organisms are reproducing at random (no **non-random mating**)
5. No **mutations** are happening

Term	Definition
Natural Selection	There are differences in fitness.
Gene Flow	Immigration & emigration of a population or individuals in a population.
Genetic Drift	When some outside factor causes there to be a dramatic shift in allele and gene proportions. Two types: • Founder effect: A few of a population start over somewhere else • Bottleneck: All of a population are killed off but a few.
Non-random Mating	When mating is not random (wow this is helpful). i.e. ***sexual selection***.
Mutations	Alleles are changing.

The Hardy-Weinberg equation describes the situation where a population ***is not evolving***. It is our **null** for evolution. Thus if any of the 5 requirements are not met, then the population ***must be evolving***.

IMPORTANT NOTE: the definition table is *not* the list of assumptions. It defines terms in the assumptions, but the actual assumptions are the *absence* of the terms in the table. Just look at the numbered list above.

Apply the Hardy-Weinberg equation to real-world scenarios to hypothesize evolutionary causes

Case Study: Malaria

You don't need to know all this, it's just an example

- Kenya lowlands have high malaria presence, but low morbidity (death rates)
- Kenya highlands have just recently begun to experience high malaria presence and their morbidity rates are high.

Thoughts
From Heather: the people in the lowlands have been exposed to malaria for longer, so they have developed genetic resistances (through mutations). Maybe they're different genetically.

- Moormann et al., 2003, 2003 hypothesized that highland populations have less protective genes than lowland populations.
- Their test: look at frequency of a known malaria-resistant gene, *Hemoglobin S* (The gene for Sickle Cell Anemia).

If the lowland and highland populations are in Hardy-Weinberg equilibrium, then their genetics should be the same and not evolving.

So we took some datas and here we got the Hemoglobin A/S genotypes.

Genotype	Lowland area transmission	Highland area transmission
HbAA	254 (74%)	340 (97%)
HbAS	90 (26%)	12 (3%)
HbSS	0	0

The proportion of A in lowland:

$$\frac{254 * 2 + 90}{344 * 2} \approx .87$$

And in the highland:

$$\frac{352 * 2 + 12}{352 * 2} \approx .98$$

So if we've got one S you are more resistant. If you get two S's then you're totally immune apparently but a sad side effect is that most of them die in infancy. So here we see some Stabilizing Selection (away from either homozygous genotype and towards the heterozygous genotype).

Compare expected and actual frequencies to hypothesize evolutionary causes.

Day 18

Terminology

Term	Definition
Chromosome	A packet of DNA - 23 pairs
Gene	A Recipe for a trait
Allele	Variations of a gene
Homologous	Same genes, same order, but **NOT** necessarily the same alleles
Sister Chromatid	Duplicates of a chromosome

Genetic Equation

$$an = b$$

- a: the number of copies of a gene
- n: the number of unique chromosome types
- b: the total number of chromosomes

Draw the process of meiosis.

Meiosis is a type of cell division that reduces the number of chromosomes in the parent cell by half and produces four gamete cells. So it's turning diploids into a haploids.

Sperm production example

Jack has a $2n = 46$ structure (like all people). Since it's $2n$ he's got diploid cells.

- The cells that make sperm start out as diploids
- Before we go through meiosis the cell duplicates its chromosomes
- So we get these "ropes" that come out from the edges of the cell and get one of each chromosome.
    - All the homologous pairs line up as pairs in the **metaphase**.
    - Law of Independent Assortment
        - Every single gene is independent from one another. (mostly true)

Explain where and how the law of independent assortment and the law of segregation are fulfilled by meiosis.

Use the principles of meiosis to predict offspring outcomes.

Determine ploidy number (e.g., diploid and haploid) of a cell based on a figure or description of the cell.

Justify why gametes should have half the number of chromosomes as the parent cell.

Determine all possible gametes that can result from a given parent.

Defining Sex

A persons sex is defined by whether they make eggs or sperm. The Y chromosome has something called SRY that makes the gonads fall.

Day 19

Inheritance Table

Type of Inheritance	Genotypes	Phenotypes	Picture
Complete Dominance	AA Aa aa	Dominant Dominant Recessive	
Codominance	AA AB BB	“A” Phenotype Both Phenotypes “B” Phenotype	
Incomplete Dominance	aa ab bb	“a” Phenotype Blended “b” Phenotype	

- For **incomplete dominance**, an ‘ab’ phenotype results in a blended phenotype rather than both showing contrastingly.
  - e.g. ‘aa’ is red, ‘bb’ is blue, then ‘ab’ results in purple

Blood Types

ABO

There are 3 alleles: `A`, `B`, `i`

- `AA`, `Ai`: A
- `BB`, `Bi`: B
- `ii`: O
- `AB`: AB

Rh Gene

This is the positive/negative thing

- `++`, `+-`: $\mathrm{Rh}+$
- `--`: $\mathrm{Rh}-$

Predict genotypes given phenotypes of family members, from word problems and pedigrees.

Solve problems about traits expressing complete dominance, codominance, and incomplete dominance.

Identify and predict genotypes for traits with more than two alleles.

Perform a monohybrid and dyhibrid cross, using Punnett squares and/or multiplication rules.

Assess the mode of inheritance for a given trait, using pedigrees or family history.

Day 20

Solve problems about traits expressing sex linkage.

Solve problems where traits are linked on the same chromosome.

Day 21

Explain the process of gamete formation in males and females.

Predict genetic results of in vitro fertilization using polar bodies.

Determine how to increase the odds of becoming pregnant.

Determine the parent in which nondisjunction occurred, and when during meiosis or mitosis nondisjunction occurred, given a genetic outcome of the daughter cells or offspring.

Day 22

Terminology

Term	Definition
Mitosis	Process used by the body to repair and grow cells. It's an exact replication of a cell. It creates an exact copy.
Interphase: G_1	Gap 1: Where the cell is doing it's job. This when a cell is being a cell. Like a heart cell is pumping the blood

Term	Definition
Interphase: S	Synthesis
Interphase: G_2	Gap 2: Doing it's job again
Checkpoints, What do they check?	Checks for errors in DNA, damage, health, do we need another cell?
G_0	"Retirement" of the cell. Still doing it's job but it's not gonna reproduce. Fancy term is *senesence*
Apoptosis	Cell death - on purpose. Cellsuicide. Celluicide.

Compare and contrast meiosis and mitosis.

While **meiosis** halves the number of chromosomes in the process of creating a new cell, **mitosis** duplicates a cell. So for **meiosis**:

$$2n \rightarrow n$$

But for **mitosis**

$$2n \rightarrow 2n$$

When thinking about whether an event is mitotic or meiotic, only look at the chromosome number.

Diagram the key steps in each phase of the cell cycle and in each phase of mitosis.

Predict a meiotic or mitotic event in a life cycle given the purpose.

Predict when and how cancer might occur in the life cycle of a cell.

Explain and contrast proto-oncogenes, tumor suppressor, and repair genes; predict the effects of a mutation in each.

Determine at which checkpoint a cancerous cell has experienced a mutation.

Predict the effect of lengthening telomeres.

Control Mechanisms

- Tumor suppressors - “off switches” - e.g. p53
- Proto-oncogenes - “on switch” - e.g. Cyclin
- Contact Inhibition
- Anchorage Dependence
- Telomeres
  - Every time you go through the S phase, the telomeres get a bit shorter
  - A cell can only go through mitosis a certain number of times.
  - Telomosomething enzyme lengthens telomeres in the balls and also in cancer cells

Benign vs. Malignant

These are two different types of growth.

- Benign expands but stays within its cell type.
- Malignant is expansion and inflitration of other cell types.

How to Treat Cancer

- Slash
  - you go in and physically remove it
- Burn
  - Burn it off with radiation. Cause a bunch of DNA damage so it goes through apoptosis
- Poison
  - Chemotherapy
  - Targets cells that are going through mitosis
  - So it kills cancer cells but also kills your other cells.