Hello everybody! My name is Ben Fitch, and I am a Junior Cell and Molecular Biology major on the pre-med track and I am the master tutor for Genetics. Every week, I will be creating a ‘resource’ for this course over many of the primary topics which you are covering in class. These will act as supplementary resources to guide your studying and help provide extra practice with major concepts. If you ever have questions or concerns, feel free to contact me through the tutoring center or via my primary email address. This is, in my opinion, the most interesting course at Baylor so I’m so excited to jump into this new year with you!

Remember: the Tutoring Center offers free individual and group tutoring for this Genetics. Our Group Tutoring sessions will be Thursday from 5:15-6:15 PM at the Sid Rich basement, room 75! You can reserve a spot at https://baylor.edu/tutoring. I hope to see you there!

Keywords: Cell Cycle, Mitosis, Meiosis, Chromosome, Inheritance

Week One (01/17-01/21)

*Topic of the Week: Mitosis, Meiosis and Counting Chromosomes (2.2-2.3)*

*Homologous Chromosomes (homologs):* a matched pair of chromosomes from each parent

*Ploidy:* a constant (n) used to describe how many copies of the genome are present

- **Haploid (n):** cell only has one set of chromosomes
- **Diploid (2n):** cell with 2 sets of chromosomes (homologous)

*Mitosis:* the division of a parent cell into two identical daughter cells (2n → 2n)

- **Prophase:** chromosomes condense (spaghetti to breadstick), spindle poles form
- **Prometaphase:** nucleus degrades and spindles attach to kinetochores
- **Metaphase:** chromosomes align at the center of the cell
  
  **Checkpoint:** the spindle assembly is checked to ensure chromosomes will
  *spoiler alert* separate in anaphase

- **Anaphase:** *Separase* separates chromatids and move to poles; cell elongates

- **Telophase/Cytokinesis:** spindle/centrosomes degrade and nuclear membrane reforms; cell pinches and cleaves off into 2 daughters
*Please try to complete this chart; see the link on page 8 of the document and check your work!

**Meiosis:** The 2 divisions of a single diploid parent cell to 4 genetically different haploid daughters (2n→n)

**Sources of Variation:**
- **Random Alignment** of homologs in metaphase 1
- **Crossing Over** of homologs in prophase 1 (**chiasma**)

**Meiosis 1:** reductional division→ separates homologous pairs (2n → n)

**Shugoshin** prevents separase from lysing cohesins in sister chromatids

**Meiosis 2:** equational division→ divides chromatids as in mitosis (n → n)

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**Highlight #1: Overview of Genetics and Chromosomal Structure** (1; 2.1-2.2)

**Genetics:** the study of gene inheritance, structure and pathology
- **Transmission:** how traits are passed
- **Molecular:** the physical process of gene transmission and expression
- **Population:** changes and trends in genes over time within populations

**Model Organism:** an organism which can be used to simply and feasibly replicate biological processes emergent in humans or other higher species

**Viruses:** small genome wrapped in a protein coat capable of infecting cells

**Chromosomes:** **chromosomes** are bundles of DNA wrapped around proteins

- **Sister Chromatid:** 1 chromosome composed of 2 DNA strands joined at the centromere by **cohesin** proteins
- **Locus:** the specific point on a chromosome where a gene is located

All diagrams, tables and figures are the property of Benjamin A. Pierce; Genetics: A Conceptual Approach

Additional sources are the property of Paramount Pictures
Eukaryotes vs. Prokaryotes: [https://www.youtube.com/watch?v=RQ-SMCmWB1s](https://www.youtube.com/watch?v=RQ-SMCmWB1s)

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**Highlight #2: The Cell Cycle (2.2)**

**Cell Cycle:** the cycle of cellular growth and division

- **Interphase:** the part of the cell cycle dedicated to growth/repair, metabolism, and DNA replication
  - **G₁:** Cellular growth and metabolism
  - **G₁-S Checkpoint:** cell is either put into G₀ or commits to undergoing division
  - **G₀:** a nondividing state which most body cells are in
  - **S:** Synthesis → DNA replicates, but chromosomal number is unchanged
  - **G₂:** Cell prepares structurally and biochemically to undergo mitosis
  - **G₂M Checkpoint:** ensure correct DNA replication; ensures cell is prepared to divide

- **M-Phase:** division of the nucleus
- **Cytokinesis:** division of the cytoplasm

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**Week 1 Concept Check** (for practice with counting chromosomes, see Formative Practice)

1. A cell loses function in its gene for separase. Assuming that this does not affect passage through the G₂M checkpoint, what would be the expected outcome in mitosis?
2. What happens to a cell which is in a mature/resting tissue after G₁?

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**Week Two (01/24-01/28)**

**Topic of the Week: Math and Stats in Genetic Crosses (3.2-3.4)**

**Crosses:** do you best to memorize the common patterns in all Mendelian crosses

- **Monohybrid Cross:** a cross at a single locus (Law of Segregation)
- **Dihybrid Cross:** a cross at two loci (Law of Independent Assortment)
- **Testcross:** a cross between a homozygous recessive and an unknown genotype

**Addition Rules:** rules for adding probabilities (Keyword “OR”)

**Multiplication Rules:** rules for multiplying probabilities (Keyword “AND”)

**Conditional Probability:** the probability of an event happening depending on another

**Binomial Expansion:** the probability (P) that an event (x) with a probability p will occur s times and the alternate event (y) with probability q will happen t times:

\[ P = \frac{n!}{s!t!} (p^s \times q^t) \]

* n is the number of times an event occurs and “!” is the factorial, and is typed on the calculator as [value]!

**Chi Square (X²):** a statistical test which assess if difference between observed and expected values is significant
\[ X^2 = \sum \frac{(\text{Observed} - \text{Expected})^2}{\text{Expected}} \]

*note: degrees of freedom (DF) is n-1 (number of samples -1)

**Null Hypothesis (H₀):** states that the difference between O and E is due to chance alone

**α-Value = 0.05:** states that you are 95% (1.0-0.05 = 0.95) confident in your significance

**Critical Value:** value on \( X^2 \) table that matches with the p value at a given DF

**Rule of thumb:** if \( X^2 > \text{CV}, p < 0.05 \rightarrow \text{significant difference; reject } H_0 \)

if \( X^2 < \text{CV}, p > 0.05 \rightarrow \text{insignificant difference; FAIL TO REJECT } H_0 \)

**Additional Video Links:**
[https://www.youtube.com/watch?v=ODjNc3YgtNY&list=PLYjFOc4FIyilGl_jbf1vkMEofNQZDiZr2](https://www.youtube.com/watch?v=ODjNc3YgtNY&list=PLYjFOc4FIyilGl_jbf1vkMEofNQZDiZr2)

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**Highlight #1: Principles of Mendelian Genetics (3.1-3.2)**

**Mendelian Inheritance:** the general pattern of heredity discovered by Gregor Mendel

**Law of Segregation:** each individual has 2 copies of an allele which code for a trait; these two alleles are separated (Anaphase 1) of gamete formation

**Law of Independent Assortment:** in a cross involving more than two genes, the alleles segregate independently of each other (unless they are linked)

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**Highlight #2: Sex Determination and Sex-Linkage (4.1-4.3)**

**Chromosomal Sex Determination:** generally, most studied organisms display the X-Y system for sex determination, though several others exist

**X-Chromosome:** present in males and females; has dose-dependent* sex determining genes and autosomal genes

**Y-Chromosome:** only present in males; controls expression of masculinizing genes

**SRY Gene:** the gene which causes testes to form and increases testosterone

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Sex Chromosomes</th>
<th>Sexual Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>n/a (F)</td>
<td>XX</td>
<td>Female</td>
</tr>
<tr>
<td>n/a (M)</td>
<td>XY</td>
<td>Male</td>
</tr>
<tr>
<td>Klinefelter</td>
<td>XXY</td>
<td>Male</td>
</tr>
<tr>
<td>Turner</td>
<td>XO</td>
<td>Female</td>
</tr>
<tr>
<td>Triple X</td>
<td>XXX</td>
<td>Female</td>
</tr>
<tr>
<td>XYY Male</td>
<td>XYY</td>
<td>Male</td>
</tr>
</tbody>
</table>

*All diagrams, tables and figures are the property of Benjamin A. Pierce; Genetics: A Conceptual Approach*
*Additional sources are the property of Paramount Pictures*

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Sex Linked Gene: a gene located on a sex chromosome

X-Linked: mother to child or father to child (dominant or recessive)

Y-Linked: father to son only

Hemizygous: since males only carry one copy of the X-chromosome (or the Y), they are considered hemizygous (single allele carriers)

Supplementary Videos: https://www.youtube.com/watch?v=YXVyIDaUOD8 (~2min)
https://www.youtube.com/watch?v=h2xufrHWG3E (~6min)

Drosophila Eye Color (X-ample)

Eye color inheritance in Drosophila (fruit flies) is sex linked. White (X<sup>w</sup>) and red (W<sup>r</sup>) is a common cross. A testcross of a white eyed male and an unknown female with red eyes is shown below. Red-eyed F1 males were crossed with white eyed F1 females. The following progeny was found below. Based on this, what method (dominant or recessive) affects white eye inheritance and what is the original female’s genotype?

P
X<sup>w</sup>Y x X<sup>r</sup>X<sup>? </sup>

F1
♂→ 1:1 of red : white ♀→ 1:1 of red : white

F2
♂→ all white ♀→ all red

Lyon Hypothesis: in all individuals with more than 1 X-chromosome, all but 1 will be inactivated (at random)

Barr Body: the remnant of an inactivated X chromosome

*note: some sex determining genes are not inactivated, so the ‘feminizing’ effect depends on X-chromosome dosage and whether or not there is an SRY gene

**Week 2 Concept Check**

1. Dwarfism in corgis is caused by a recessive allele (c) at the C<sup>d</sup> locus. In a testcross with a regular corgi of unknown genetic origin and a dwarf corgi (cc),
   a. What are possible expected progeny ratios?
   b. Imagine, in this situation, the cross yields a litter of 27 puppies. 16 are regular height and 11 are dwarves. Is this difference significant? (include an X<sup>2</sup> value and a probability in your answer)

2. Kelvin and Jade have 8 kids. What is the probability that 6 will be female and 2 will be male?

**Week Three (01/31-02/04)**

*Topic of the Week: Extensions and Modifications of Basic Principles (5)*

Single Locus Factors

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*Additional sources are the property of Paramount Pictures*
Incomplete Dominance: when the phenotype of a cross fall in between that of the two parents
Codominance: the phenotype is represented by expression of both alleles (ex. Roan cow)
Multiple Alleles: each individual only possesses 2 alleles at a locus, but more than two possible alleles exist at the locus. Additionally, this may display codominance (ex. Human ABO blood type)

![Figure 5.11](image)

Penetrance: the proportion of people who both possess a genotype and express the genotype
Incomplete Penetrance: the genotype does not always produce the expected phenotype

Gene Interaction
Epistasis: when a gene at one locus covers the effect of a separate gene at a different locus
  Recessive Epistasis: 9:3:4 → (ex. labs)
  Dominant Epistasis: 12:3:1 → (ex. squash)
  Duplicate-Recessive Epistasis: 9:7 → (ex. snails)

Sex Effects
Sex Influenced: autosomal genes which differ in their expression in males and females (one allele may be dominant in males and recessive in females, or vice versa!)
Sex Limited: a trait which is expressed in one sex, but has zero penetrance in the other sex (ex. Precocious puberty)
Genetic Maternal Effect: the phenotype of the offspring is determined by the genotype of the mother
Genomic Imprinting: certain genes may be expressed differently based on if they came from the mother or the father’s set of chromosomes

More Videos: [https://www.youtube.com/watch?v=YJHGfbW5510](https://www.youtube.com/watch?v=YJHGfbW5510) (~7)
[https://www.youtube.com/watch?v=higO3lZhdNY](https://www.youtube.com/watch?v=higO3lZhdNY) (~5)

Highlight: Pedigrees (6)
[https://www.youtube.com/watch?v=3Gd09V2AkZv4](https://www.youtube.com/watch?v=3Gd09V2AkZv4)

Autosomal Recessive: Equal proportions in males and females; can skip generations/be ‘hidden’ by carriers
Consanguinity: inbreeding/cross between cousins
Autosomal Dominant: Every affected individual must have an affected parent; Won’t skip generations

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X-Linked Recessive: Unequal proportion of males and females affected (more in males); may skip generations

Rule of Thumb:
- When a daughter is affected, the father is affected
- An affected son’s mother has the trait, or is a carrier (heterozygote)

X-Linked Dominant: Every affected individual must have an affected parent; Won’t skip generations

Rule of Thumb:
- Every affected male’s daughter has the trait
- Sons: inherit from mom only
- Daughters: inherit from mother or father

Y-Linked trait: Passed from father to son; doesn’t skip generations (males only)

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Week 3 Concept Check

1. Unicorn horns differ in expression between males and females. The trait follows this pattern

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Male</th>
<th>Female</th>
</tr>
</thead>
<tbody>
<tr>
<td>H&lt;Hu</td>
<td>unhorned</td>
<td>horned</td>
</tr>
<tr>
<td>H&lt;hu</td>
<td>unhorned</td>
<td>horned</td>
</tr>
<tr>
<td>hh</td>
<td>horned</td>
<td>unhorned</td>
</tr>
</tbody>
</table>

a. What type of inheritance pattern is shown?
b. What percent (%) of males would you expect to be horned if a horned male is crossed with a true breeding horned female?

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THINGS YOU MAY STRUGGLE WITH:

1. A sister chromatid is only one chromosome, but is two molecules of DNA. Until separase cleaves the cohesins in Anaphase, chromosome count is $\frac{1}{2}$ (number of DNA). At anaphase, chromatids split into two separate chromosomes, so number of chromosomes = numbers of DNA.

2. You DON’T need to use punnett squares! You can use addition and multiplication rules to solve problems quicker and easier.

3. A dihybrid cross can be set up a two separate monohybrid crosses; Mendel’s law of independent assortment show that alleles at different loci are inherited separately, so we can set up two monohybrid crosses and use the multiplication rule to find the probability of each happening together.

4. Genomic imprinting, sex influence, sex limited and genetic maternally effected traits are NOT sex linked! These depend on epigenetics, expressivity and penetrance, not the location of an allele on the X or Y chromosome!

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You Try: Click the link to try these practice problems on google forms!
Formative Practice Week 3: https://forms.gle/26SKq7FnAikYuLgUA
Link to Answers for Chromosome Counting

CONGRATS: You made it to the end of the resource! Again, group tutoring will be every Thursday from 5:15-6:30 PM. You can reserve a spot at https://baylor.edu/tutoring. I hope to see you there!

Answers:

Week 1:
1. The sister chromatids will fail to separate during Anaphase
2. The cell enters nondividing G₀ phase

Week 2:
1.
   a. 1:0 (tall:dwarf) → CᶜCᶜ x cc; 1:1 (tall:dwarf) → Cᶜc x cc
   b. H₀: there is no significant difference between the observed and expected number of corgi phenotypes
      i. α = 0.05 | DF: 1 | X² = 0.925 | CV = 3.84 | 0.5<p<0.75
      ii. Fail to reject H₀; any variation is due to chance alone
2. p = 0.109

Drosophila X-ample: X-linked recessive; P female is XX⁺ → that is the only way that there could be F₁ white-eyed males because the P male would not pass his X-chromosome to male offspring

Week 3:
1.
   a. Sex influenced trait
      i. Male: h dominant; Female: H⁺ dominant
   b. H⁺H⁺ x hh (female x male) → progeny?
      i. Female progeny → H⁺h 1
      ii. Male progeny → H⁺h 1
         1. 100% of males are hornless

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