Week 3
Genetics: BIO-2306

The sections this resource covers are the topics typically covered in the first 3 weeks of the semester. If you do not see the topics your particular section of class is learning this week, please take a look at other weekly resources listed on our website for additional topics throughout the semester.

We also invite you to look at the group tutoring chart on our website to see if this course has a group tutoring session offered this semester.

If you have any questions about these study guides, group tutoring sessions, private 30 minute tutoring appointments, the Baylor Tutoring YouTube channel or any tutoring services we offer, please visit our website www.baylor.edu/tutoring or call our drop in center during open business hours. M-Th 9am-8pm on class days 254-710-4135.

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

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

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

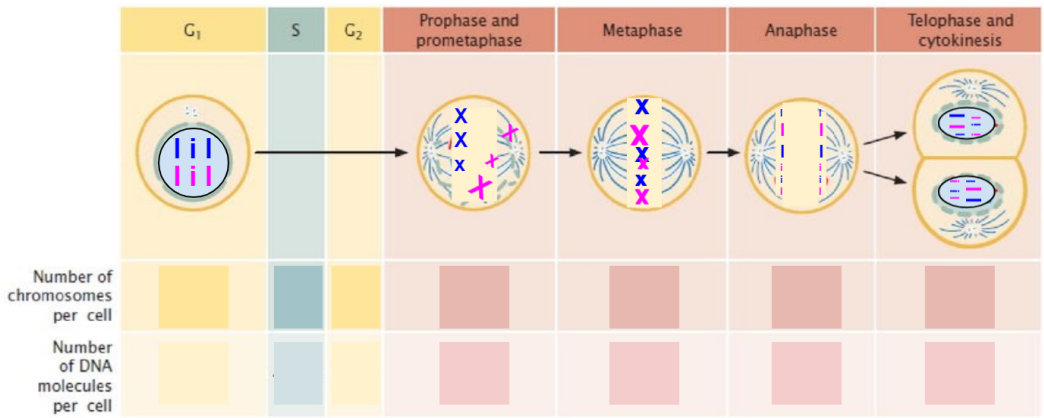
Ploidy: a constant (n) used to describe how many copies of the genome are present

- **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:

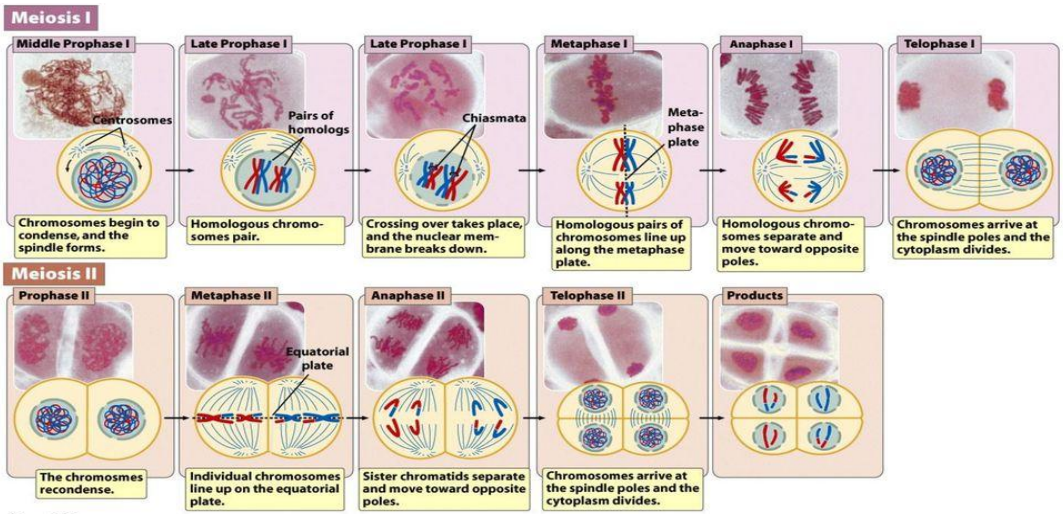
Random Alignment of homologs in metaphase 1

Crossing Over of homologs in prophase 1 (**chiatisma**)

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)



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

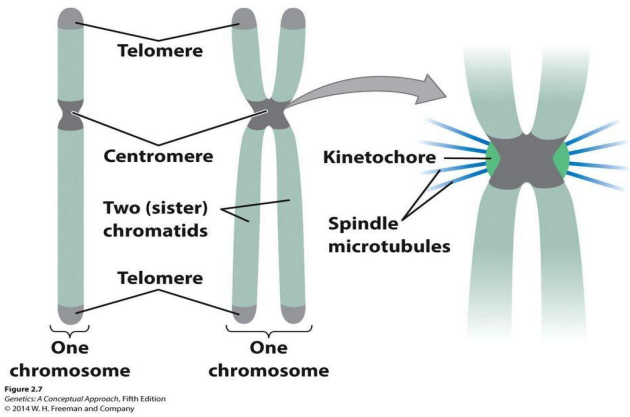
Model Organism: an organism which can be used to simply and feasibly replicate biological processes emergent in humans or other higher species

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



Eukaryotes vs. Prokaryotes: 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
  - **G_1:** Cellular growth and metabolism
    - **$G_{1\text{-}S}$ Checkpoint:** cell is either put into **G_0** or *commits* to undergoing division
    - **G_0:** a nondividing state which most body cells are in
  - **S :** *Synthesis* → DNA replicates, but chromosomal number is unchanged
  - **G_2:** Cell prepares structurally and biochemically to undergo mitosis
    - **$G_{2\text{-}M}$ Checkpoint:** ensure correct DNA replication; ensures cell is prepared to divide
- **M-Phase:** division of the nucleus
- **Cytokinesis:** division of the cytoplasm

Concept Check 1 *(answers at the end)* (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 $G_{2\text{-}M}$ checkpoint, what would be the expected outcome in mitosis?
2. What happens to a cell which is in a mature/resting tissue after G_1?

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 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**:

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

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

Chi Square (X^2): a statistical test which assess if difference between observed and expected values is significant

$$X^2 = \Sigma \frac{(Observed - Expected)^2}{Expected}$$

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

Null Hypothesis (H_0): 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 $X^2 > CV$, $p < 0.05 \rightarrow$ significant difference; reject H_0

if $X^2 < CV$, $p > 0.05 \rightarrow$ insignificant difference; *FAIL TO REJECT* H_0

Additional Video Links:

https://www.youtube.com/watch?v=ODjNc3YgtNY&list=PLYjFOc4FIyilGI_jbf1vkMEofNQZDiZr2

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***)

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

Syndrome	Sex Chromosomes	Sexual Phenotype
n/a (*F*)	**XX**	**Female**
n/a (*M*)	**XY**	**Male**
Klinefelter	XXY	Male
Turner	XO	Female
Triple X	XXX	Female
XYY Male	XYY	Male

Sex Linked Gene: a gene located on a sex chromosome

X-Linked: mother to child **<u>or</u>** 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^w) and **red** (W^r) 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

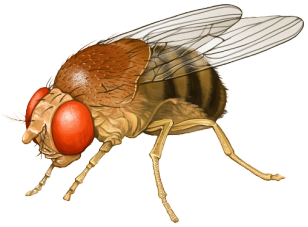
X^wY x $X^rX^?$

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

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Concept Check 2 *(answers at the end)*

1. Dwarfism in corgis is caused by a recessive allele (c) at the C^d 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^2 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?

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

Single Locus Factors

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
Genetics: A Conceptual Approach, Sixth Edition
© 2017 W. H. Freeman and Company

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=YJHGfbW55l0** (~7)
https://www.youtube.com/watch?v=higO3IZhdNY (~5)

Highlight: Pedigrees (6)

https://www.youtube.com/watch?v=Gd09V2AkZv4

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

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**)

CHECK YOUR LEARNING

Z-Concept Check 3 *(answers at the end)*

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

Genotype	Male	Female
H^uH^u	unhorned	horned
H^uh	unhorned	horned
hh	horned	unhorned

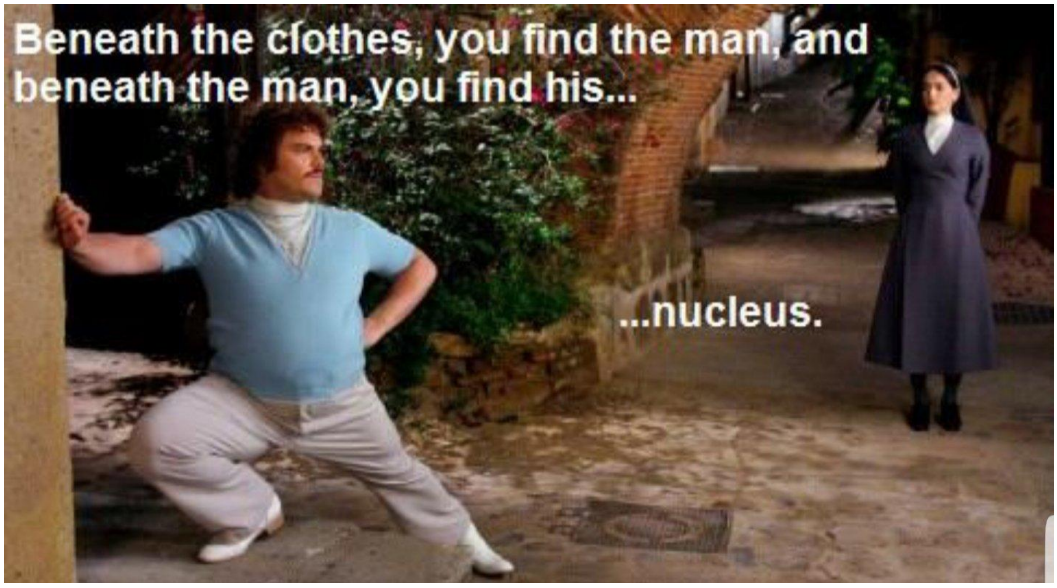
   a. What type of inheritance pattern is shown?
   b. What percent (%) of males would you expect to be horned if a horned male is crossed with a true breeding horned female?

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 ½(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!

You Try: Click the link to try these practice problems on google forms!
Formative Practice: https://forms.gle/26SKq7FnAikYuLgUA

[Link to Answers for Chromosome Counting]



CONGRATS: You made it to the end of the resource! Thanks for checking out these weekly resources! Don't forget to check out our website for group tutoring times, video tutorials and lots of other resources: www.baylor.edu/tutoring!
Answers to check your learning questions are below!

Answers:

Concept Check 1:

1. The *sister chromatids* will fail to separate during Anaphase
2. The cell enters nondividing G_0 phase

Concept Check 2:

1.
   a. 1:0 (tall:dwarf) → C^dC^d x cc; 1:1 (tall:dwarf) → C^dc x cc
   b. H_0: there is no significant difference between the observed and expected number of corgi phenotypes
      i. $\alpha = 0.05$ | DF: 1 | $X^2 = 0.925$ | CV = 3.84 | $0.5<p<0.75$
      ii. ***Fail to reject*** H_0; any variation is due to chance alone
2. p = 0.109

Drosophila X-ample: X-linked recessive; P female is X^rX^w → 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

Concept Check 3:

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