7.013 Recitation 4 – Spring 2018

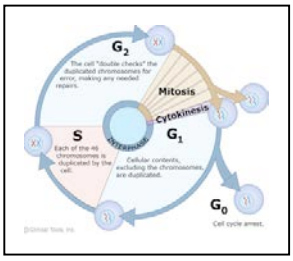
*(**Note:** The recitation summary should NOT be regarded as the substitute for lectures)*

Summary of Lectures 5 (2/16) and 6 (2/20):

Note: Some commonly used terms in genetics along with their definition are given below.

Terms	Definitions
Gene	(abstractly) a fundamental unit of inheritance; (chemically) a sequence of DNA that encodes an RNA or protein.
Allele	Different forms or variants of a gene
Homozygous	Presence of identical alleles at one loci between homologous chromosomes
Heterozygous	Presence of different alleles at one loci between homologous chromosomes
Genotype	Set of alleles; genetic make-up
Phenotype	Physical appearance, trait or characteristic imparted by the genotype
Pure- or true- breeding	Contains homozygous alleles at the gene of interest
Trait	One or more detectable variants in a genetic character
Dominant	Phenotype that is expressed in a heterozygote
Recessive	Phenotype that is NOT expressed in heterozygote

Cell cycle: The cell cycle is the process by which one cell becomes two identical cells. The cell cycle is the chain of events that occur in a dividing cell. Cells preparing to undergo cell division must first copy each of their double-stranded DNA molecules (or chromosomes) by DNA replication. The duplicated homologs remain attached to each other by cohesin protein. The cell cycle consists of four stages – G1, S (DNA synthesis/ replication), G2, and M (mitosis). G1 phase is when the cells are preparing to replicate their DNA, which occurs in S phase. G2 is when the cells are preparing to divide, which occurs in M phase. Of these, the G1 phase shows variable length in different cell types. Hartwell & Nurse received the Nobel Prize in 2001 for their contribution. Please see the link below.



Link: http://www.nobelprize.org/nobel_prizes/medicine/laureates/2001/illpres/index.html

Cell division: Mitosis is the division of a diploid somatic cell (2n) to form two daughter cells (2n) that are similar to the parental somatic cell. Meiosis has two divisions; meiosis 1 looks different from mitosis, but meiosis II looks similar to mitosis. In Meiosis I a parent cell (2n) duplicates its genome (4n) and then divides to form two daughter cells (2n), this involves the separation of duplicated homologs. In meiosis II, each daughter cell (2n) divides to form two daughter cells (n), this involves the separation of chromatids. So the end result of meiosis is the production of four haploid gamete cells/ reproductive cells such as eggs or sperm, each of which has half the number of chromosomes (n) as a diploid somatic cell (2n).

Link: http://www.pbs.org/wgbh/nova/body/how-cells-divide.html

Non-disjunction and its effect: It is important to note that failure of separation of duplicated chromosomes (non-disjunction) during cell division may result in daughter cells with abnormal number

of chromosomes. If non-disjunction occurs during Mitosis, it results in both daughter cells having an abnormal number of chromosomes (n+1 for one daughter cell and n-1 for the other), which may be transferred to subsequent daughter cells and can result in diseases such as cancer. However non-disjunction in mitosis is never passed on to the offspring of the next generation. In comparison, non-disjunction either during Meiosis -1 or Meiosis-II can result in gametes with abnormal number of chromosomes and is the cause of genetic disorders such as Downs syndrome, Edwards syndrome, Turner's syndrome, Kleinfelter's syndrome etc.

Recombination and crossing over: Homologous recombination is the reciprocal exchange of DNA between two homologous chromosomes that occurs only during metaphase of meiosis I. During homologous recombination, two homologs (e.g. the chromosome #7 inherited from dad and the chromosome #7 inherited from mom) break at the same place and switch ends with each other. Homologous recombination results in a change in which alleles are on the same chromosomes together (e.g. a cell with "AB" on the maternal homolog and "ab" on the paternal homolog can give rise to gametes containing "Ab" or "aB" chromosomes). The two recombinants ("Ab" and "aB") will always be created in equal frequencies because one of each kind is created by a single recombination event.

Mendel's laws; Monohybrid cross: Here you cross/ mate two true-breeding (i.e. homozygous for the alleles of a gene(s)) organisms that differ by a single characteristic that is regulated by one gene, such as plants that produce round versus wrinkled seeds. The two organisms you cross are called the P or F_O (parental) generation. For example, a cross between two plants, which are both homozygous and differ from each other with respect to one gene, can be represented as AA X aa where "A" and "a" are the two alleles of the same gene. The offspring that represent the F1 (filial) generation will all receive one allele from each parent and will have the "Aa" genotype, which makes them heterozygous. These F1 offspring will display whichever of the two phenotypes is dominant i.e. such as round seeds. If you cross two F1 plants together (Aa x Aa), you create the F2 generation. The ratios of different genotypes (AA:2Aa:aa) and phenotypes (Round 3: wrinkled 1) within the F2 generation can be derived either by using the basic probability (the sum rule and the product rule) or by using a Punnett Square.

Mendel's laws; Dihybrid cross: A dihybrid cross is when you cross/ mate two true-breeding organisms that differ by two characteristics each being controlled by a single gene, such as pea plants that produce round versus wrinkled seeds (controlled by Gene R, that can exist as allele R or r) and green or yellow peas (regulated by Gene Y that exists as allele Y or allele y). For example, a cross between two pea plant P1 (genotype: RRYY) and P2 (genotype: rryy) can result in F1 offspring (genotype: RrYy), which are heterozygous for the alleles of both genes and which display the dominant phenotypes (in this case round and green). If you cross two F1 organisms together (RrYy x RrYy), you create the F2 generation with the phenotypes: Round/ Yellow (9): Round/ Green (3): Wrinkled/Yellow (3): wrinkled/ Green (1). The ratios of different genotypes and phenotypes within the F2 generation can be derived either by using the probability rule or by using a Punnett Square.

Test cross: This is used to determine whether an individual showing a dominant trait is homozygous or heterozygous for the allele(s) of a gene(s). The individual in question is crossed with an individual that is homozygous for the recessive trait- this is easy to identify, because all individuals with the recessive phenotype are homozygous for that trait. If the individual in question is homozygous then all the offspring resulting from the test cross will be heterozygous and will exhibit the dominant trait. In comparison, if the individual in question is heterozygous, then the back cross will produce offspring showing the dominant trait and offspring showing the recessive trait in the ratio of 1:1.

Linked and Unlinked genes: During prophase of Meiosis I, genes at different loci on the same chromosome can be separated from one another and recombined by crossing over. When crossing over occurs between two linked genes, not all the progeny of a cross have the parental phenotypes. Instead the recombinant offspring appear as well, in proportions called recombination frequency, which is calculated by dividing the number of recombinants by the total number of progeny. Recombination

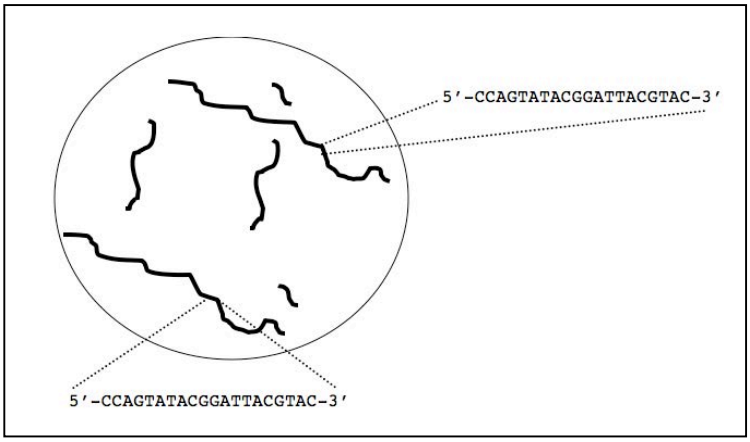
frequency is greater for loci that are far apart on the chromosome than for the loci that are close to each other.

For example during crossing over/ homologous recombination, two homologs (e.g. the chromosome #7 inherited from dad and the chromosome #7 inherited from mom) break at the same place and switch ends with each other. Homologous recombination results in a change in which alleles are on the same chromosomes together (e.g. a cell with "AB" on the maternal homolog and "ab" on the paternal homolog can give rise to gametes containing "Ab" or "aB" chromosomes). The two recombinants ("Ab" and "aB") will always be created in equal frequencies because one of each kind is created by a single recombination event. Again you can identify the offspring with recombinant phenotypes by doing a test cross (crossing with a homozygous recessive parent)!

Two genes can either be unlinked, completely linked or somewhat linked. Genes that are unlinked are usually located on different chromosomes from each other or are located far apart (50cM or 50 map units) from each other on the same chromosome. Genes that are completely linked are very close to each other on the same chromosome (map distance = 0cM). Genes that are somewhat linked are located on the same chromosome with a map distance of <50cM. Unlinked genes assort independently from each other; this means that an AaBb parent would create their four possible gametes (AB, ab, Ab, and aB) each 25% of the time. Genes that are completely linked always segregate with each other. This means that an AaBb parent would create only two (parental types and no recombinants) of the four possible gametes. Genes that are somewhat linked to each other show a pattern that is in between what is seen for unlinked genes and completely linked genes.

Questions:

1. Below is pictured the nucleus from a skin cell taken from a mouse (named Mouse #1), with the chromosomes shown in bold.



a) Is the cell haploid or diploid? _____

b) Let's say we blew up a region of the longest chromosome to look at its sequence at one small gene.

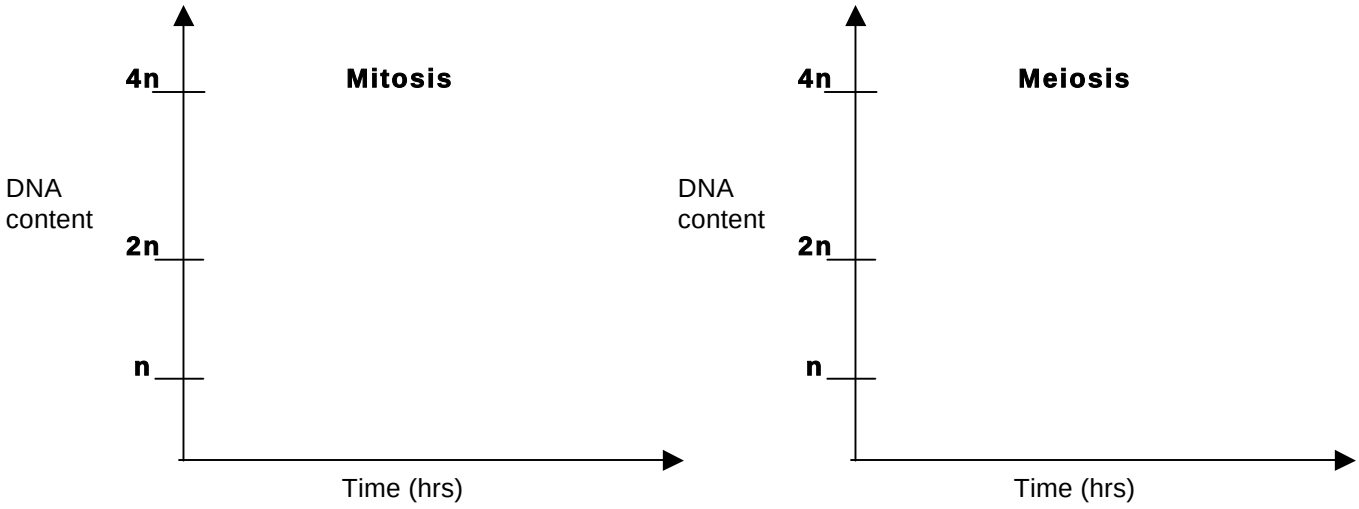
i. Is Mouse #1 homozygous or heterozygous for this gene?

ii. When you zoom in on the same locus on the long chromosome in a cell from Mouse #2 you find that its two homologous chromosomes have the sequences. How might we write the genotypes of Mouse #1 and Mouse #2 if Sequence #1 encodes a functional protein but Sequence #2 encodes a protein that no longer works? *(**Note:** Use the upper case or lowercase A while writing the genotypes).*

5'-CCAGTATACGGA**G**TACGTAC-3'
Sequence 1

5'-CCAGTATACGGATTACGTAC-3'
Sequence 2

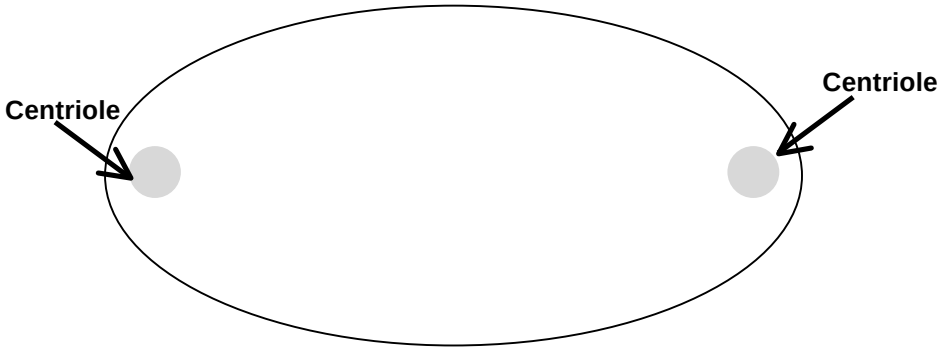
2. In a diploid cell (2n), that is undergoing cell division, draw a graph showing the variation in the DNA content (n = haploid) against time for the mitosis and meiosis.



3. You are following the segregation of three genes; A, B and D in Heidi's cells (**genotype: $AAX^{BD}X^{bd}$**). ***Note:*** *Gene A is located on the autosome whereas Genes B and D are on the X chromosome.*

a) Give the possible genotype(s) of a cell from **Heidi's dad** for A, B and D genes. ***Note:*** *Assume that Heidi's dad is homozygous for Gene A.*

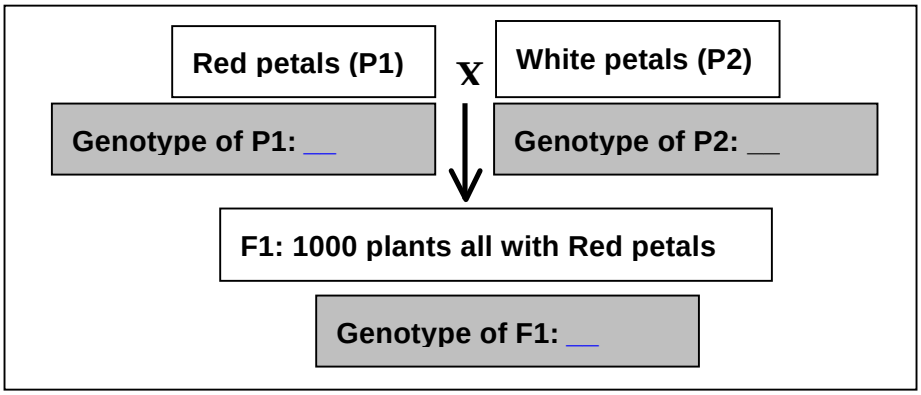
b) Assume that a **somatic cell** (shown below) from Heidi undergoes cell division. Draw the arrangement of A, B and D genes at the metaphase plate of this dividing cell. ***Note:*** *The centrioles at the two ends are drawn for you.*



c) Assume that the **somatic cell** drawn above undergoes **nondisjunction** of **one** X chromosome carrying B and D genes. All other chromosomes separate normally. Give the genotypes of **all** possible daughter cells in terms of A, B and D genes.

d) Heidi has a daughter whose genotype is **$AAX^{BD}X^{bd}X^{bd}$**. Could the presence of the extra X chromosome be explained by nondisjunction in part (c)? Why or why not?

4. You want to study petal color (red or white) in a variety of plants. Gene A that is located on an autosome, regulates this trait. You perform the following **monohybrid cross**.



a) In the shaded boxes to the left, give the **genotype(s)** of P1, P2 and F1 plants. ***Note:*** Represent the allele of Gene A associated with the dominant phenotype as ***"A"*** *and recessive phenotype as* ***"a"***

b) You mate two **F1 plants** to obtain 1000 plants in F2 generation.

i Fill in the Punnett square to the right for this cross.

Gametes-→ ↓		

ii List the **phenotypes and corresponding ratio** of the F2 plants for petal color.

Phenotype(s) and corresponding ratio: _____

iii List the **genotypes and corresponding ratio** of the F2 plants for petal color.

Genotype(s) and corresponding ratio: _____

c) Using a Punnett square, design a mating experiment that can help you test whether a plant having red petals is homozygous or heterozygous for the alleles of Gene A.

Gametes-→ ↓		

i. If the test plant was **homozygous** for the alleles of Gene A…

- **Phenotype(s) and ratio** of the resulting plants: _____
- **Genotype(s) and ratio** of resulting plants: _____

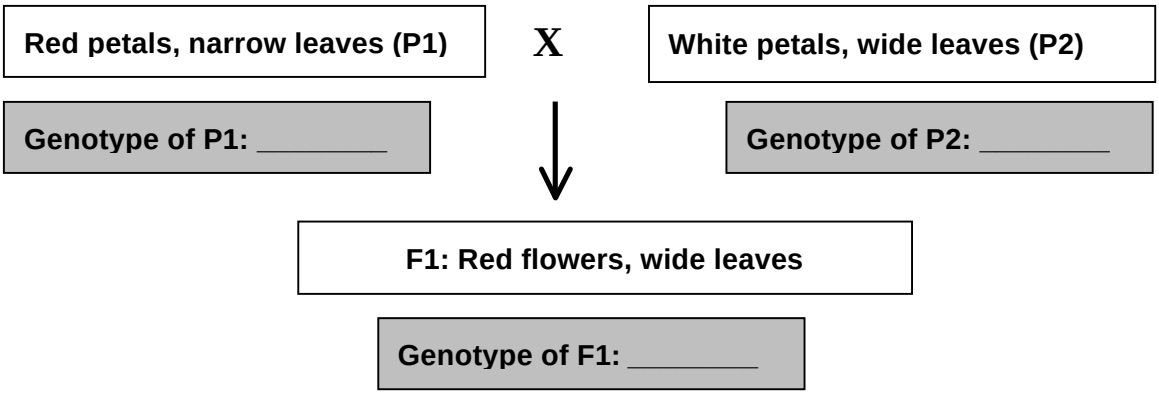
Gametes-→ ↓		

ii. If the test plant was **heterozygous** for the alleles of Gene A…

- **Phenotype(s) ratio** of the resulting plants: _____
- **Genotype(s) ratio** of resulting plants: _____

d) You also want to study leaf shape (wide or narrow) that is regulated by Gene B in the same variety of plant. **Note:** Genes A and B *are located on two different autosomes. Gene B can exist as* ***"allele B"*** *(associated with the dominant phenotype) or* ***"allele b"*** *(associated with the recessive phenotype).*

You perform a dihybrid cross between P1 and P2 plants both of which are true-breeding for the above traits.



In the shaded boxes above, give the **genotype(s)** of P1, P2 and F1 plants by using the specified notation for the alleles of Gene A (petal color) and Gene B (leaf shape)

e) You mate an **F1 plant with another plant that has white petals and narrow leaves.** You obtain 1600 plants in the F2 generation.

i. Give the likely **genotype** of the plant that has white petals and narrow leaves: _____

ii. If Gene A and Gene B **assort independently,** how many F2 plants will have white petals and narrow leaves? _____

f) If you **mate two F1 plants** to get 1600 plants in the F2 generation, how many of these will have white petals and narrow leaves? _____

5. Tomato plants can be **tall** or **short** and have **notched** or **smooth** leaves. You cross a tall, smooth leafed plant with a short, notched leafed plant. All of the progeny are **tall**, and **notched** leafed.

a) Which traits are dominant and which are recessive? _____

b) What are the genotypes of the two **true-breeding** parents? *Use the letters H or h to represent the alleles of the height gene and the letters S or s to represent the alleles of the leaf gene. In each case, use the uppercase letter for the allele associated with the dominant phenotype and the lower case letter for the allele associated with the recessive phenotype.*

Tall & smooth: _____ **Short and notched;** _____

c) Two tall, notched F1 plants were crossed (self cross) to get 1600 F2 plants

i. What ratio of phenotype do you expect in the F2 generation?

ii. If you get 400 plants in F2, how many of these will be homozygous recessive for both traits?

d) You cross an F1 Plant with a double homozygous recessive plant (genotype: hhbb) and get 1600 plants in F2 generation. Give the four classes of the F2 plants that you will see. Give the genotype, phenotype and the number of each of F2 classes generated.

6. In fruit flies, the “B/b” gene and the “G/g” gene are linked. In one specific fly whose genotype is GgBb, “B” is linked to “g” on the maternal chromosome #2, and “b” is linked to “G” on the paternal chromosome #2.

a) List all parental type (non-recombinant) gametes by genotype that could be produced by this GgBb fly.

b) List all recombinant gametes by genotype that could be produced by this GgBb fly.

c) Say that 40% of all gametes produced by the GgBb fly are “Bg.” What percentage of all gametes produced by this fly are recombinants?

d) As the recombination frequency between two genes on the same chromosome rises, does the physical distance between those two genes lower **or** rise?

7.013 Introductory Biology

Spring 2018