

7.014 Problem Set 6 Solutions

Question 1

a) Define the following terms:

Dominant – In genetics, the ability of one allelic form of a gene to determine the phenotype of a heterozygous individual, in which the homologous chromosomes carries both it and a different (recessive) allele.

Recessive – In genetics, an allele that does not determine phenotype in the presence of a dominant allele.

Phenotype – The observable properties of an individual resulting from both genetic and environmental factors.

Genotype – An exact description of the genetic constitution of an individual, either with respect to a single trait or with respect to a larger set of traits.

Alleles – The alternate forms of a genetic character found at a given locus on a chromosome.

Homozygous – In a diploid organism, having identical alleles of a given gene on both homologous chromosomes. An individual may be a homozygote with respect to one gene and a heterozygote with respect to another.

Heterozygous – Of a diploid organism having different alleles of a given gene on the pair of homologues carrying that gene.

Mendel's First Law – Law of Segregation - In genetics, the separation of alleles, or of homologous chromosomes, from one another during meiosis so that each of the haploid daughter nuclei produced by meiosis contains one or the other member of the pair found in the diploid mother cell, but never both.

Mendel's Second Law – Law of Independent Assortment - During meiosis, the random separation of genes carried on nonhomologous chromosomes.

Sex-Linked – The pattern of inheritance characteristic of genes located on the sex chromosomes of organisms having a chromosomal mechanism for sex determination.

Haploid – Having a chromosome complement consisting of just one copy of each chromosome; designated $1n$ or n .

Diploid – Having a chromosome complement consisting of two copies (homologues) of each chromosome. Designated $2n$.

Question 1, continued

- b) One summer, you decide to become an UROP student for a professor who studies the genetics of corn, *Zea mays*. When you cross the following a true-breeding strain of corn with purpled, smooth kernels to a true-breeding strain of corn with yellow, wrinkled kernels you find that the F_1 progeny all have purple, smooth kernels. You then cross two F_1 progeny and see the following in the F_2 generation.

P₀ Phenotype

Purple, Smooth Kernels

Yellow, Wrinkled Kernels

P₀ Genotypes**

| |
|------|
| AABB |
|------|

| |
|------|
| aabb |
|------|

F₁ Phenotype

Purple, Smooth Kernels

F₂ Genotype**

| |
|------|
| AaBb |
|------|

F₂ Phenotypes*

Yellow, Wrinkled Kernels

Yellow, Smooth Kernels

Purple, Smooth Kernels

Purple, Wrinkled Kernels

of F₂ Progeny

56,251

167,879

508,746

170,435

F₂ Genotypes**

| |
|----------|
| aabb (1) |
|----------|

| |
|----------------|
| aaBb, aaBB (3) |
|----------------|

| |
|----------------------------|
| AaBb, AABB, AaBB, AABb (9) |
|----------------------------|

| |
|----------------|
| Aabb, AAbb (3) |
|----------------|

- i) Does the pattern above indicate a Mendelian method of inheritance for the traits of kernel color and kernel texture? Explain.

The pattern above corroborates with a Mendelian mode of inheritance: (1) The breeding of two true breeding strains results in a single F_1 progeny indicating that the purple kernel and the smooth kernel traits each correspond to dominant phenotypes, whereas the yellow kernel and the wrinkled kernel traits correspond to recessive traits. (2) In the F_2 generation, we find that the kernel color gene and the kernel texture gene are unlinked, or likely to be on different chromosomes. These F_2 progeny are the result of segregation of alleles and independent assortment during meiosis

- ii) Kernel color is encoded by gene *A*, where *A* is associated with the dominant phenotype and *a* is associated with the recessive phenotype. Similarly, kernel texture is encoded by gene *B*, where *B* is associated with the dominant phenotype and *b* is associated with the recessive phenotype. Indicate the genotype(s) for each of the corn plants in the above boxes under **Genotypes****.

Question 1, continued

c) You are unsure of the genotype of an individual F_2 corn plant (from Part b) that has purple, smooth kernels and would love to know its genotype.

i) What cross would allow you to determine the genotype of the purple, smooth corn plant?

A test cross - A cross of a dominant-phenotype individual (which may be either heterozygous or homozygous) with a homozygous-recessive individual. That is, A-B- (purple, smooth) × aabb (yellow, wrinkled)

ii) Describe the four different possible scenarios from your cross above. Indicate phenotypes and genotypes.

(1) $AaBb \times aabb \rightarrow$

- _ $AaBb$ (purple, smooth)
- _ $Aabb$ (purple, wrinkled)
- _ $aaBb$ (yellow, smooth)
- _ $aabb$ (yellow, wrinkled)

(2) $AABb \times aabb \rightarrow$

- _ $AaBb$ (purple, smooth)
- _ $Aabb$ (purple, wrinkled)

(3) $AaBB \times aabb \rightarrow$

- _ $AaBb$ (purple, smooth)
- _ $aaBb$ (yellow, smooth)

(4) $AABb \times aabb \rightarrow$

- _ $AaBb$ (purple, smooth)
- _ $Aabb$ (yellow, wrinkled)

Question 2

The fruitfly *Drosophila melanogaster* has been a focus of genetic studies since the early twentieth century with the pioneering experiments of Thomas Hunt Morgan. It is a good system for genetic studies because of its small size, ease of cultivation, and short generation time.

As a junior Drosophologist, you discover a mutation that results in flies with curly wings. You gather 30 curly-winged flies and perform the following P_0 and F_1 crosses and receive the results below:

P_0 30 curly-winged flies × 30 normal-winged flies

F_1 603 curly-winged flies 197 normal-winged flies

You then perform several self-crosses with the two kinds of F_1 flies

| | Self-Cross of curly-winged flies | Self-Cross of normal-winged flies |
|-------|--|-----------------------------------|
| F_2 | 598 curly-winged flies & 205 normal-winged flies | All normal-winged flies |

a) Is the curly-winged phenotype dominant or recessive?

The curly-winged phenotype is dominant.

Question 2, continued

- b) The curly phenotype follows a Mendelian mode of inheritance, so why don't the numbers of offspring follow Mendelian ratios?

The Curly-winged flies in the parental generation are not true-breeding, i.e., there are some heterozygotes in the mix.

- c) Enamored by the robustness of *Drosophila* genetics, by the cuteness of curly-winged flies, and by the prospects of an entrepreneurial venture selling flies to frog and lizard pet owners, you decide to breed flies with altered wings. (FYI: Curly- and short-winged flies do not fly as well as normal-winged flies so frogs and lizards can feed easily.) You create two different pure-breeding strains of short winged flies and call them *sht1* and *sht2*. In both strains, the short-winged phenotype is recessive.

- i) What experiment would allow you to determine whether these two strains carry mutations in the same gene or in two different genes? What results would you expect if the two strains have mutations in different genes?

You would perform a **complementation test**, whereby you cross the short-winged *sht1* strain (*sht1/sht1*) with the short-winged *sht2* strain (*sht2/sht2*). If they were in the same gene, the progeny from this cross would have short wings (*sht1/sht2*). However, if the mutations represent two different genes, you would only get normal-winged flies (if they are on the same chromosome: *sht1 +/+ sht2*. Or, if they are on a different chromosome *sht1/+; sht2/+*).

- d) You identify mutations in two genes that result in the following recessive phenotypes: (1) flies homozygous for *dbl1* have double-wings and (2) flies homozygous for *prpl1* have purple eyes rather than red eyes. You then perform the following cross:

| | | |
|--|---|--|
| Normal-winged, Red-eyed flies <i>dbl1⁺ dbl1⁻ prpl1⁺ prpl1⁻</i> | × | Curly-winged, Purple-eyed flies <i>dbl1⁻ dbl1⁻ prpl1⁻ prpl1⁻</i> |
|--|---|--|

- i) What would the phenotypic ratios of the progeny be if *dbl1* and *prpl1* were unlinked genes?

1 (or $\frac{1}{16}$) normal-winged, red-eyed flies (*dbl1⁺/dbl1⁻ ; prpl1⁺/prpl1⁻*)
1 (or $\frac{1}{16}$) normal-winged, purple-eyed flies (*dbl1⁺/dbl1⁻ ; prpl1⁻/prpl1⁻*)
1 (or $\frac{1}{16}$) double-winged, red-eyed flies (*dbl1⁻/dbl1⁻ ; prpl1⁺/prpl1⁻*)
1 (or $\frac{1}{16}$) double-winged, purple-eyed flies (*dbl1⁻/dbl1⁻ ; prpl1⁻/prpl1⁻*)

- ii) Instead of finding that the genes *dbl1* and *prpl1* are unlinked, you find that *dbl1* and *prpl1* are linked. When you perform the above cross, you find the following progeny:

68 normal-winged, red-eyed flies
73 double-winged, purple-eyed flies
6 normal-winged, purple-eyed flies
9 double-winged, red-eyed flies

Calculate the recombination frequency between the *dbl1* and *prpl1* genes

Recombination frequency = (total # recombinants)/(total # progeny) = (6+9)/(68+73+6+9) = 15/156 = 0.096

Question 3

There are four blood types in human: A, B, AB, and O. This is a single gene trait with three alleles. The A and B blood types (associated with alleles I^A and I^B , respectively) are both dominant to the O blood type (associated with i allele). The A and B blood types are co-dominant to each other. You are a genetic counselor and physician at the Massachusetts General Hospital. The combinations for genotypes and phenotypes are listed below:

| Genotype | Phenotype |
|-----------------------|------------------|
| $I^A I^A$ and $I^A i$ | A |
| $I^B I^B$ and $I^B i$ | B |
| $I^A I^B$ | AB |
| ii | O |

a) One of your female patients makes an inquiry to see if her friend was actually the father of her child. The mother is blood type B and the child is blood type O.

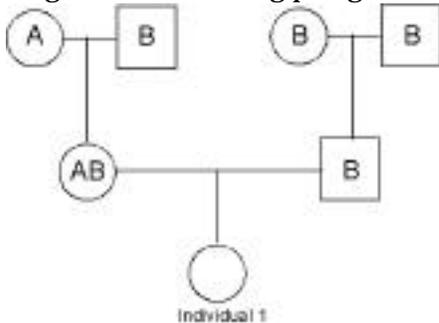
- i) Unfortunately, the possible father was killed while performing dangerous genetic experiments with corn and fruitflies. Your patient believes that this man had blood type A. Would this information exclude this man as the father of the child? Explain.

No, the mother (blood type B) may have a genotype of $I^B i$ and the accused father (blood type A) may have a genotype of $I^A i$. Therefore, they may each pass their i allele to their child who is O.

- ii) Luckily, the possible father's parents are still alive and they submit blood samples to be tested: his father and mother are both blood type AB. You also find records that the possible father was actually blood type A. Is the accused father the actual father of the child? Explain.

The accused father cannot be the actual father. Since his parents are both blood type AB, they are both genotypically $I^A I^B$. Thus, since the father is blood type A, his parents would pass only the I^A allele to their son. Therefore, the accused father is $I^A I^A$ and would not be able to pass an i allele to the fatherless child.

b) You are given the following pedigree for family blood types:



- i) If Individual 1 is blood type B, what is the probability that she will have the genotype $I^B i$?
If both grandparents are BB then the Dad is BB and the probability is 0. If either or both grandparent is BO and the dad is BO , then the probability is $1/4$.

- ii) **The parents of Individual 1 now have a newborn child that is blood type A.** What is the probability that Individual 1 would have the genotype $I^A I^B$?

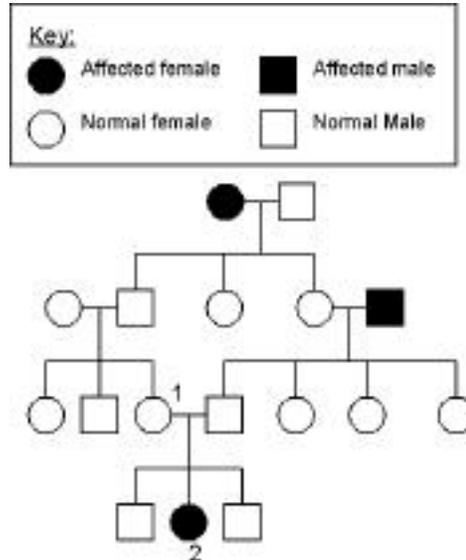
Probability = (50% probability of getting I^A allele from AB mother) \times (50% probability of getting I^B allele from B ($I^B i$) father)

Question 3, continued

c) For the following pedigrees determine:

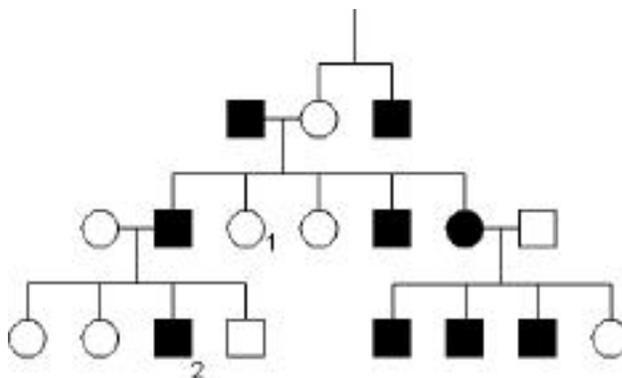
- The most likely mode of inheritance (e.g., dominant, recessive, autosomal, X-linked, new mutation, etc.) of the genetic disease
- The probable genotype of the numbered individual(s). Indicate any ambiguities. Be sure to define your genotype symbols clearly.

Disease A Pedigree



- Mode of inheritance: *Autosomal Recessive (Trait often skips generations. Two unaffected parents may have affected offspring. Consanguinity present in the family. Members of both sexes have equal probability of inheriting the disease allele.)*
- Genotypes: (1) Aa (2) aa

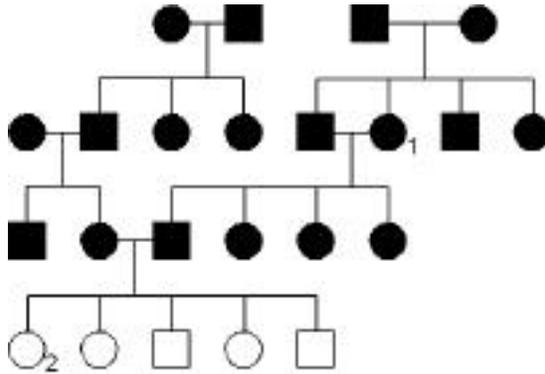
Disease B Pedigree



- Mode of inheritance: *X-linked recessive (More males than females express the trait. Affected females transmit disease allele to all of her sons.)*
- Genotypes: (1) $X^A X^a$ (2) $X^a Y$

Question 3, continued

Disease C Pedigree



- i) Mode of inheritance: (Hint: Think complementation) *The disease is caused by two different genes that are both autosomal recessive.*
- ii) Genotypes: (1) *AAbb or aaBB* (2) *AaBb*.

Question 4

You are now studying the process of mitosis and meiosis in the simple organism, *Notuu briitus*, and have prepared microscope slides of fixed (preserved) and treated these cells with DAPI, a chemical that will mark chromosomes/DNA of a cell under fluorescent microscopy. *N. briiitus* only have two chromosomes. You also have the ability identify the location of two **unlinked** genes A and B (A is dominant, a is recessive; B is dominant b is recessive); this facilitates your ability to genotype the cells. You have forgotten to label your slides while preparing them and attempt to sort the stage of the cell cycle that each may contain: G₁, S, G₂, Mitosis (Prophase, Metaphase, Anaphase, Telophase), Meiosis (Prophase I, Metaphase I, Anaphase I, Telophase I, Prophase II, Metaphase II, Anaphase II, Telophase II), or cytokinesis.

Answer the following questions to determine the characteristics of the slide that you are viewing under the microscope.

- a) Slide 1: You look in the microscope and find a single cell with decondensed chromosomes. The genotype of the cell is AAaaBBbb.
 - i) Is the cell haploid, diploid, or tetraploid? *Tetraploid*.
 - ii) What phase of the cell cycle does this belong to? *Interphase G₂ following S phase of the cell cycle*.
- b) In the space below, draw what you would see in the scope. Indicate where the alleles—A, a, B, and b—are located on the chromosomes.

(See attached figures.)

Question 4, continued

c) Slide 2: You look in the microscope and find **all chromosomes** are aligned at the center of a single cell. The genotype of the cell is AAaaBBbb.

i) Is the cell haploid, diploid, or tetraploid? *Tetraploid.*

ii) What phase of the cell cycle does this belong to? *Metaphase of Mitosis*

iii) If the cell completed its current path along the cell cycle what would be the genotypes of the resulting cells.

Two mitotic products: 2 AaBb

iv) In the space below, draw what you would see in the scope **in Slide 2**. Indicate where the alleles—A, a, B, and b—are located on the chromosomes.

(See attached figures.)

d) Slide 3: On this slide, you find two cells where sister chromatids are paired but are beginning to be pulled to one side of the chromosome by the spindle located on either side of the cell poles. The genotype of one cell is AA bb . The other cell is aa BB .

i) Are the cells haploid, diploid, or tetraploid? *Diploid.*

ii) What phase of the cell cycle do these cells belong to? *Anaphase II of Meiosis II.*

iii) If the cell completed its current path along the cell cycle what would be the genotypes of the resulting cells.

Four meiotic products: 2 Ab cells & 2 aB cells

iv) In the space below, draw what you would see in the scope. Indicate where the alleles—A, a, B, and b—are located on the chromosomes.

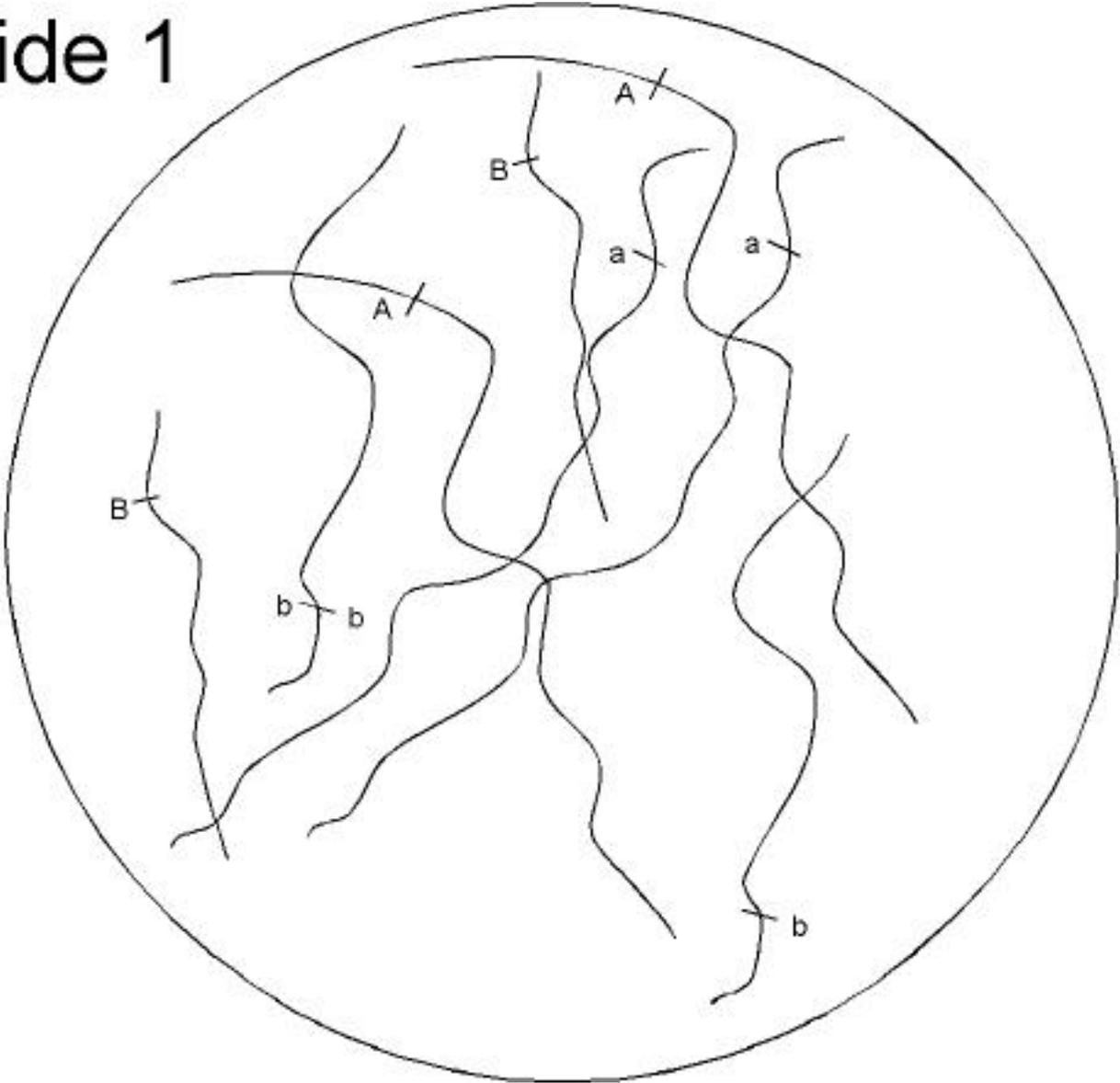
(See attached figures.)

Question 4, continued

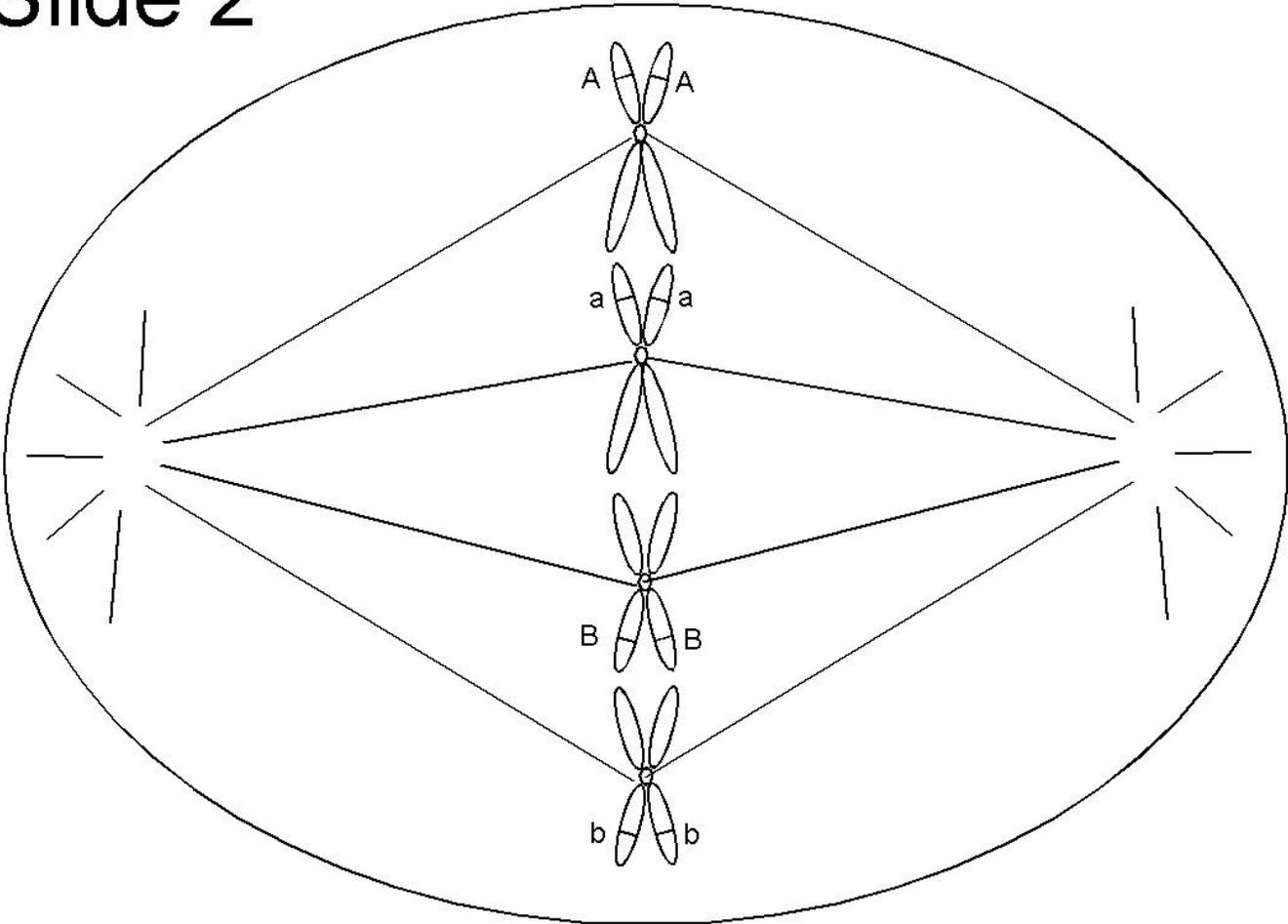
- e) Slide 4: In the following slide, you see four cells that have just completed cytokinesis. The genotypes of the cells are as follows: AB, aaB, Ab, b.
- i) What is wrong with these cells? *These should be the products of meiosis. However, non-disjunction at one of the steps in meiosis resulted in a failure of a sister chromatid to separate during meiosis II.*
 - ii) What phase of the cell cycle could this defect have occurred (Interphase, Mitosis, Meiosis I, Meiosis II)? *Meiosis II.*
 - iii) In the space below, draw what you would see in the scope. Indicate where the alleles—A, a, B, and b—are located on the chromosomes.

(See attached figures.)

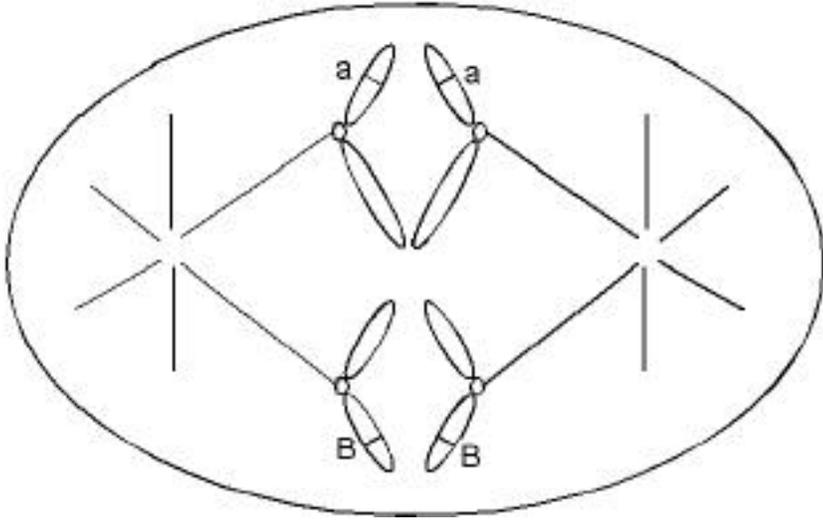
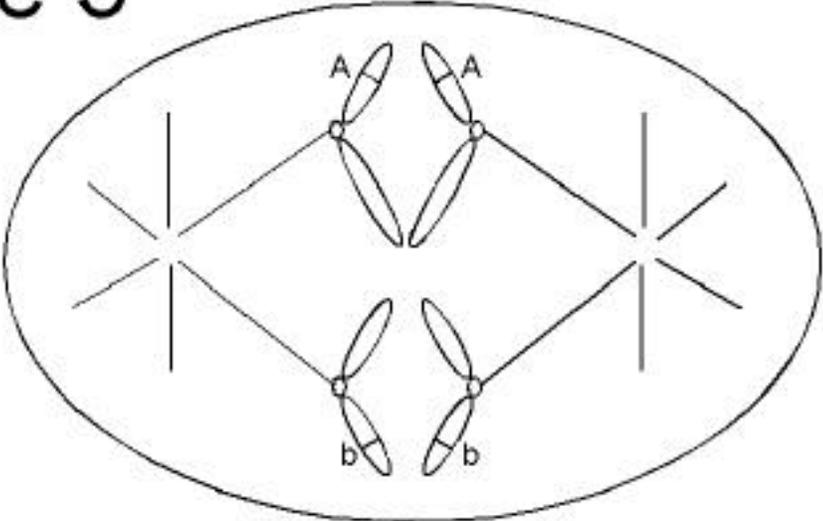
Slide 1



Slide 2



Slide 3



Slide 4

