Massachusetts Institute of Technology HHMI
StarGenetics

Name_____

Meiosis and Mendel's Law of Segregation

Introduction

In this worksheet, we are going to demonstrate how chromosomes and alleles segregate during meiosis.

Meiosis

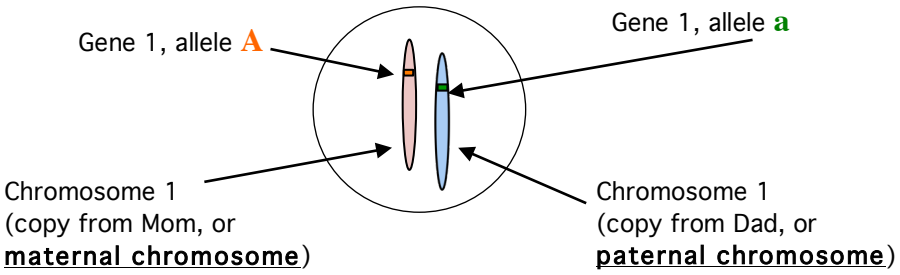
Gametes (sperm and eggs) are produced from germ cells (the progenitors of sperm and eggs) through the process of **meiosis**. Meiosis is the process in which a diploid germ cell, **diploid** meaning that the cell has two sets of chromosomes – one from each parent, first replicates its DNA and then undergoes two rounds of division to produce four haploid gametes. The resulting products of meiosis, or gametes, are **haploid** because each has only one set of chromosomes, and as a result, half the number of chromosomes as the diploid parent germ cell.

In humans, **diploid** cells in the body have 46 chromosomes or 23 pairs (2 copies for chromosomes 1-22 and 1 pair of sex chromosomes). One set of 23 chromosomes is inherited from the mother and the other set is inherited from the father. After meiosis, each gamete (eggs or sperm) has 23 chromosomes or a single set and is therefore **haploid**. When two gametes come together in the process of **fertilization**, the resulting cell has 46 chromosomes or 23 chromosome pairs and is diploid.

For simplicity, we are going to diagram only one chromosome, chromosome 1, and one hypothetical gene found on Chromosome 1, *Gene 1*, within a simplified version of a cell (only the cell membrane is shown). *Gene 1* has two different variations, or alleles: **A** and **a**.

Figure 1 illustrates what chromosome 1 looks like in a normal **diploid cell**:

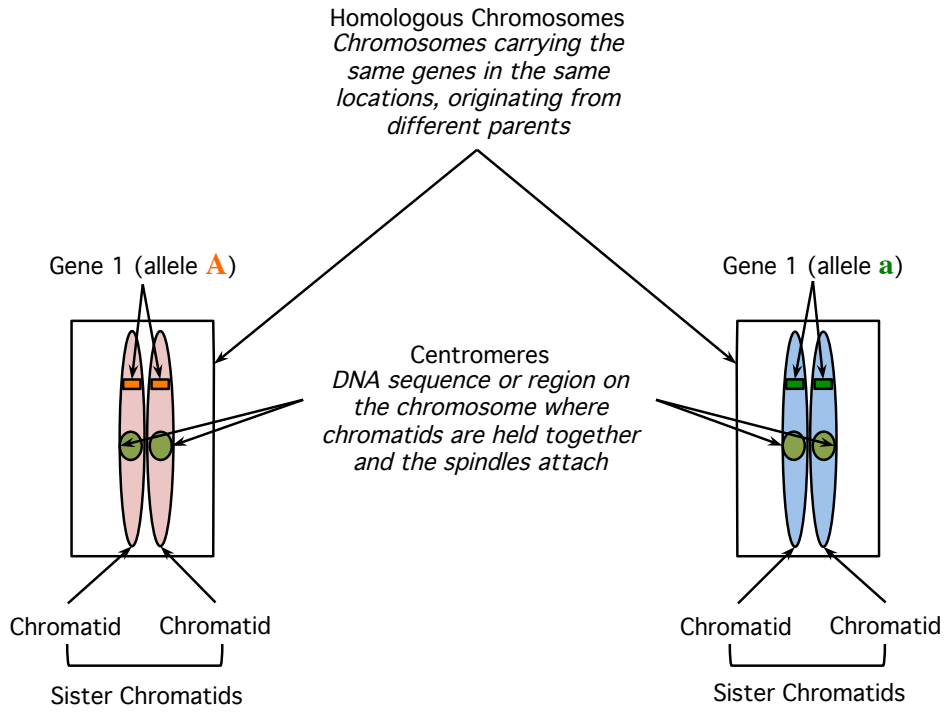
Figure 1



Following DNA replication, which occurs before the chromosomes segregate, the chromosomes look like this:

Name_____

Figure 2



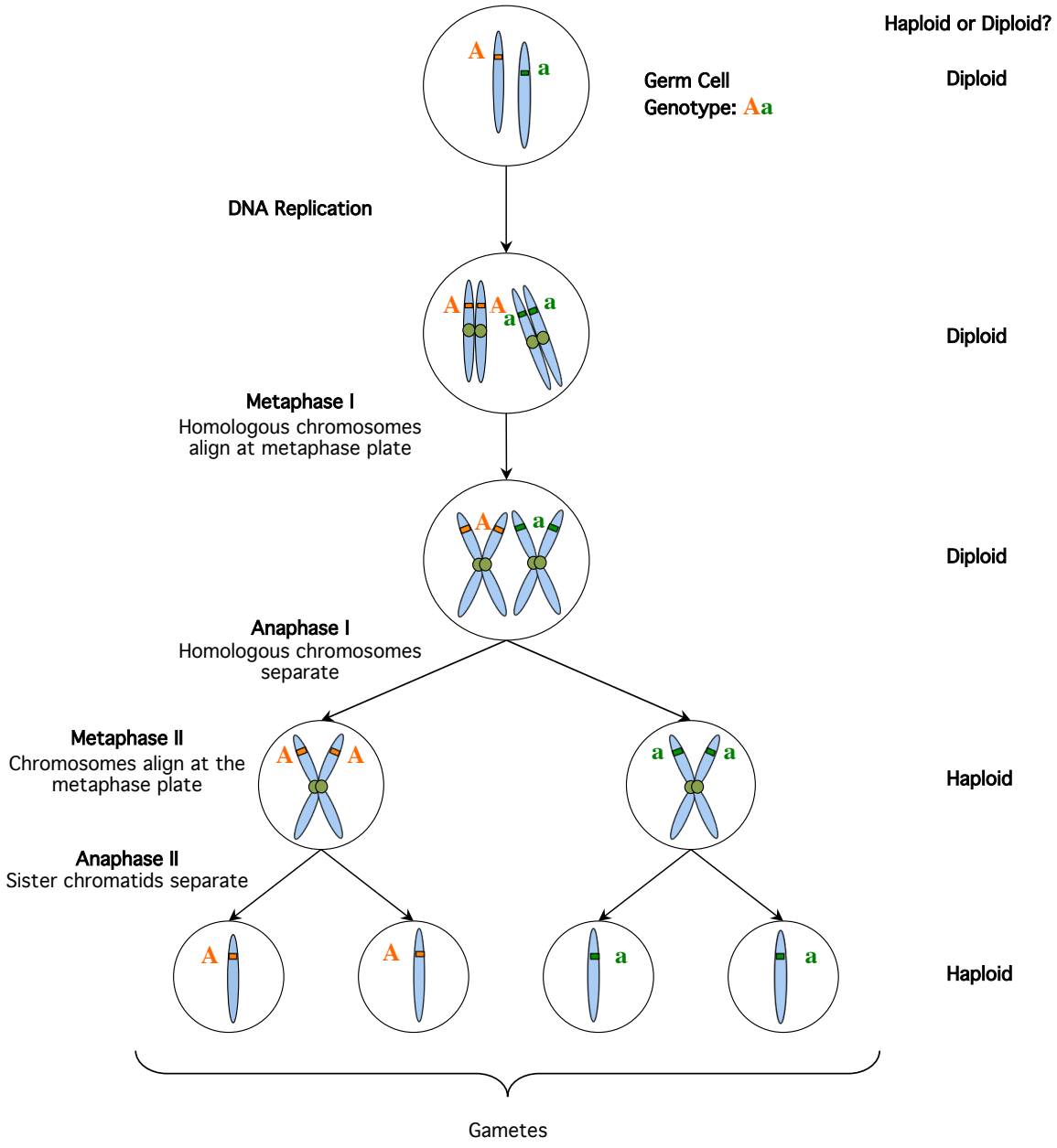
After DNA replication, we begin to call each duplicated chromosome a "chromatid." Each duplicated chromosome contains two chromatids, or "sister chromatids", which are identical in DNA sequence until recombination occurs later in meiosis. The word chromatid is only used to refer to each copy of the duplicated chromosome before sister chromatids segregate in Anaphase of Meiosis II. After this, each chromatid is again referred to as a chromosome.

Chromosome & Allele Segregation in Meiosis

Now we are going to diagram the process of chromosome segregation during meiosis. **Figure 3** illustrates the segregation of *Gene 1* in a male germ cell that is undergoing meiosis in the gonad, which is the gamete-producing organ (ex: testes in humans). This diagram only illustrates Chromosome 1 and *Gene 1*, which is located on Chromosome 1.

Name_____

Figure 3



Recombination during meiosis:

Recombination is the exchange of DNA sequences between two chromatids at a given loci, which may result in single chromatids that contain both maternal and paternal DNA sequences. Recombination occurs in Prophase of Meiosis I (when the homologous chromosomes pair up), which takes place after DNA replication and before Metaphase I. In **Figure 3**, recombination is not shown for simplicity.

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Use **Figure 3** to answer questions a-g below:

a) What is the **genotype** of the germ cell? _____

b) How many different **alleles** does the germ cell have? _____

c) Is the genotype of the germ cell **homozygous** or **heterozygous**? _____

d) Write the **genotypes** of the gametes in the spaces provided in the figure above.

e) How many **copies** of Chromosome 1 does each gamete have? ____

f) How many **alleles** of *Gene 1* does each gamete have? ____

g) How many gametes have the genotype **A**: ___/4= ___ %

a: ___/4 = ___%

h) Now imagine that there is a female germ cell which also has the **Aa** genotype with respect to *Gene 1*. How many <u>different</u> gamete genotypes will result from meiosis in the **Aa** female germ cell? _____

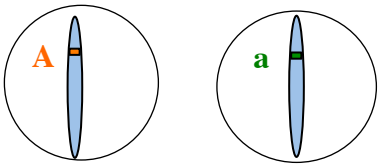
SUMMARY

Each **DIPLOID** germ cell has **2 COPIES** of a gene. Through meiosis, germ cells produce gametes, which contain only a single copy of the gene. In other words, alleles **segregate** through the process of generating reproductive cells, or meiosis. THIS DESCRIBES MENDEL'S LAW OF SEGREGATION

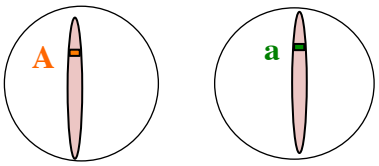
Fertilization

Now, let's think about what will happen if a sperm from the **Aa** male parent fertilizes an egg from the **Aa** female parent.

In the male parent, (Parent 1, **blue** chromosomes), meiosis will generate two types of gametes with either the genotype **A** or **a** with respect to *Gene 1*, as illustrated above. Therefore, the resulting offspring will inherit either allele, **A** or **a**, of Gene 1 based on random chance:

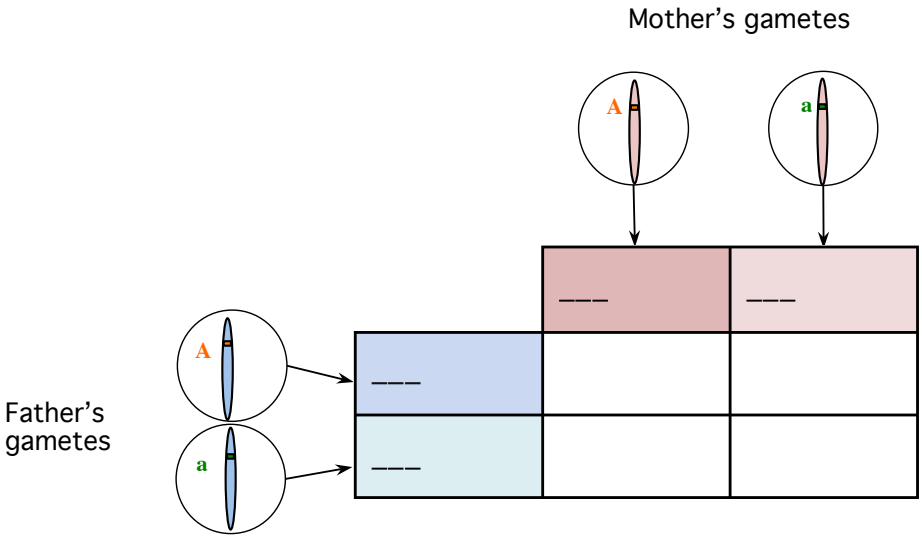


If the genotype of the female parent, (Parent 2, **pink** chromosomes) with respect to *Gene1* is also **Aa**, then the offspring will also inherit either allele, **A** or **a**, of *Gene 1* based on random chance:



Name_____

i) What are the possible genotypes of the offspring of a cross between an **Aa** male parent x **Aa** female parent? Fill in Punnett Square below with the possible genotypes of the gametes and offspring.



The outside of the Punnett Square is reserved for the alleles that make up the genotype of the parental gametes. The inside of the Punnett Square illustrates the four possible outcomes of a fertilization event involving the two specific parental gametes. All of the combinations are equally likely, meaning that there is a **1/4 chance of each outcome occurring** due to **Mendel's Law of Segregation**.

If the mother contributes the **A** allele, the resulting offspring's genotypes are those in the dark pink column. If the mother contributes the **a** allele, the resulting offspring's genotypes are those found in the light pink column.

If the father contributes the **A** allele, the genotype of the resulting offspring are in the dark blue row, and if he contributes the **a** allele, the genotype of the resulting offspring are those in the light blue row.

j) Answer questions i-vi below based on your Punnett Square in **i**). How many gametes have:

i) A homozygous genotype? _____

ii) A heterozygous genotype? _____

iii) The genotype **aa**? _____

iv) The mother's genotype? _____

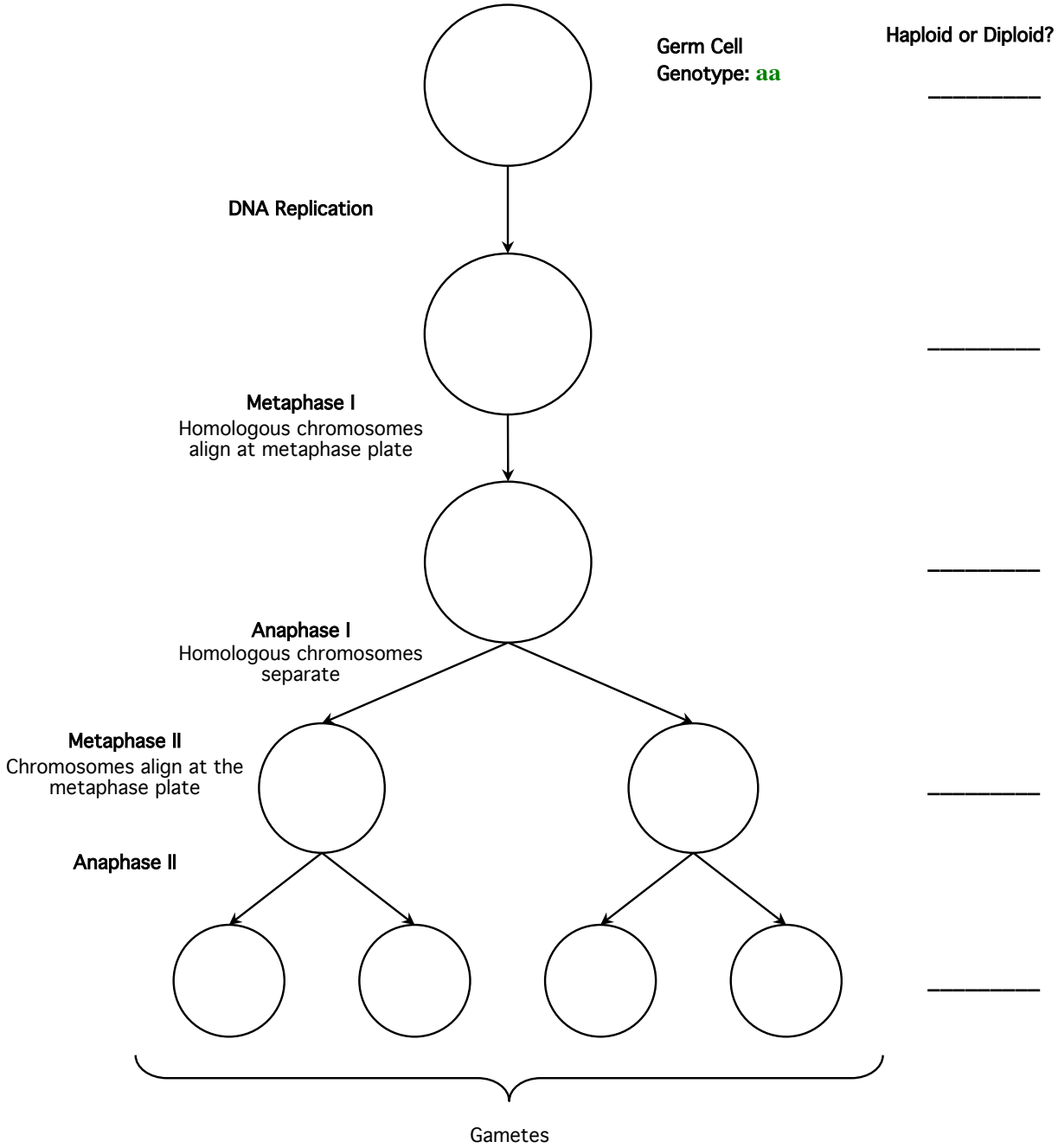
v) A genotype different from the father? _____

vi) The genotype **Aa**? **Hint**: Does the order in which you write the alleles of a genotype change the genotype? _____

k) Using **Figure 3** as an example, complete the diagram below to demonstrate meiosis in a parent with an **aa** genotype for *Gene 1*:

i. Draw Chromosome 1 in the cells below to demonstrate how this representative chromosome segregates during the different stages of meiosis. Show the location of *Gene 1* on the chromosome with a line, label the alleles (**A** or **a**) appropriately in **ALL STAGES**

ii. List the ploidy and the genotypes of the gametes at each stage provided below.



l) Based on your figure above, how many different genotypes do you observe in the resulting gametes?

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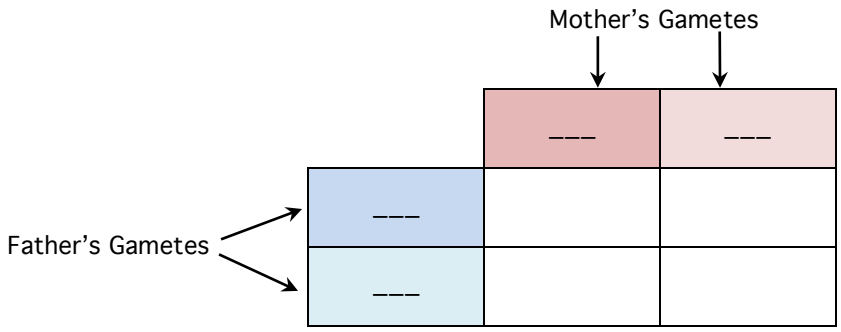
m) If an **aa** male parent mates with an **Aa** female, what are the possible genotypes of the resulting offspring? Fill in the Punnett Square below with the possible outcomes.



SUMMARY

Each offspring gets a total of **2 COPIES** of each gene through the process of **FERTILIZATION.**

One copy (one allele, ex: A or a) is inherited from the mother and one copy (one allele, ex: A or a) is inherited from the father.

These two alleles may be the same or different, depending on the genotype of the parents and random chance of two particular gametes

If the alleles (ex: A or a) for a single gene are **THE SAME** (ex: AA or aa), this individual is **HOMOZYGOUS** for this gene.

If the alleles (ex: A or a) for a single gene are **DIFFERENT** (ex: Aa), the individual is **HETEROZYGOUS** for this gene.

Applying Your Knowledge: Meiotic Errors and Non-disjunction

An error can occur in meiosis when chromosomes or chromatids do not separate properly in meiosis. This is called **non-disjunction.** There are two ways in which non-disjunction can occur: 1) both homologous chromosomes migrate together to one pole instead of separating to opposite poles in Anaphase I or 2) sister chromatids fail to separate properly and both sister chromatids move together to one pole instead of to opposite poles in Anaphase II.

n) What are the **genotypes** of the gametes in the **Aa** male we first examined at the beginning of this worksheet, if a non-disjunction event occurred in Anaphase II and the sister chromatids failed to separate into two daughter cells? Fill in the table below with the genotypes of each of the 4 gametes.

Note: Assume the non-disjunction event occurs when the precursor cell that gives rise to gametes 1 and 2 divides improperly.

Name_____

	Gamete 1	Gamete 2	Gamete 3	Gamete 4
Normal Meiosis				
Non-Disjunction in Meiosis II				

o) When a gamete with an abnormal number of chromosomes due to a non-disjunction event is fertilized, the resulting zygote will be **aneuploid** (having an irregular chromosome number). Two specific types of aneuploidy are **trisomy**, where each cell in the offspring has three copies of a single chromosome instead of two, and **monosomy**, where the offspring has only one copy of a single chromosome instead of two.

The cross of an **aa** male and **Aa** female produced an offspring with the genotype **AAa**. Therefore, the resulting offspring has 3 copies of gene 1 (trisomy for Chromosome 1). At what stage during meiosis did the non-disjunction event occur and in which parent? Circle all of the answers that apply.

a) Paternal non-disjunction in Meiosis I
b) Maternal non-disjunction in Meiosis I
c) Paternal non-disjunction in Meiosis II
d) Maternal non-disjunction in Meiosis II