Meiosis and Fertilization – Understanding How Genes Are Inherited[1]

Almost all the cells in your body were produced by mitosis. The only exception is the **gametes** – sperm or eggs – which are produced by a different type of cell division called **meiosis**.

<u>Why your body can *not* use mitosis to make sperm or eggs</u>
During **fertilization** the sperm and egg unite to form a single cell called the **zygote** which contains the chromosomes from both the sperm and the egg. The zygote divides into two cells by mitosis, these cells each divide by mitosis, and mitosis is repeated many many times to produce the cells in a baby's body.

1. A typical human cell has 46 chromosomes (23 pairs of homologous chromosomes). Suppose that human sperm and eggs were produced by mitosis. How many chromosomes would each sperm or egg have? ____

- If a sperm of this type fertilized an egg of this type, and both the sperm and egg contributed all of their chromosomes to a zygote, how many chromosomes would the resulting zygote have? _____

- If the zygote had that number of chromosomes, how many chromosomes would be in each cell in the body of an embryo that developed from the zygote? _______

An embryo with that many chromosomes in each cell would be abnormal and would die before it could develop into a baby.

- To produce a normal zygote, how many chromosomes should each sperm and egg have? _____

To produce sperm and eggs with the needed number of chromosomes, <u>meiosis reduces the number of chromosomes by half</u>. For example, in humans each gamete produced by meiosis has only 23 chromosomes, including one chromosome from each pair of homologous chromosomes.

When an egg and sperm are united during fertilization, the egg and sperm each contribute 23 chromosomes to the resulting zygote. Thus, the zygote has 23 pairs of homologous chromosomes, one in each pair from the egg and one from the sperm. When the zygote undergoes mitosis to begin to form an embryo, each cell will have the normal number of 46 chromosomes.

Cells that have two copies of each chromosome (i.e. cells that have pairs of homologous chromosomes) are called **diploid** cells. Most of the cells in our bodies are diploid cells.

2. Cells that only have one copy of each chromosome are called **haploid** cells. Which types of cells in our bodies are haploid?

3. In this chart, write the correct number of chromosomes per human cell in each blank.

Mother's cells _____	Father's cells _____
Meiosis ↓	***Meiosis*** ↓
egg _____	sperm _____

Fertilization

zygote _____
Mitosis ↓
baby's cells _____

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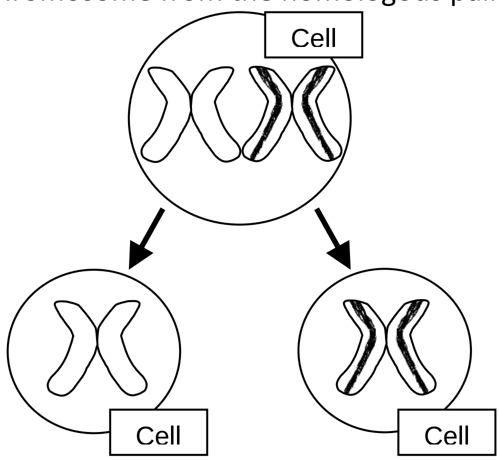
[1] by Drs. Ingrid Waldron, Jennifer Doherty, R. Scott Poethig, and Lori Spindler, Department of Biology, University of Pennsylvania, © 2014; Teachers are encouraged to copy this Student Handout for classroom use. A Word file for this Student Handout and Teacher Preparation Notes with background information and instructional suggestions are available at http://serendip.brynmawr.edu/sci_edu/waldron/#meiosis.

Meiosis – Cell Divisions to Produce Haploid Gametes

Before meiosis, the cell makes a copy of the DNA in each chromosome. Then, during meiosis there are two cell divisions, Meiosis I and Meiosis II. Meiosis reduces the chromosome number by half and produces four haploid daughter cells.

Meiosis I is different from mitosis because homologous chromosomes line up next to each other and then the two homologous chromosomes separate (see figure). Thus, Meiosis I produces daughter cells with half as many chromosomes as the parent cell, so these daughter cells are haploid. This diagram shows Meiosis I for a cell with a single pair of homologous chromosomes. Notice that each daughter cell has a different chromosome from the homologous pair of chromosomes.

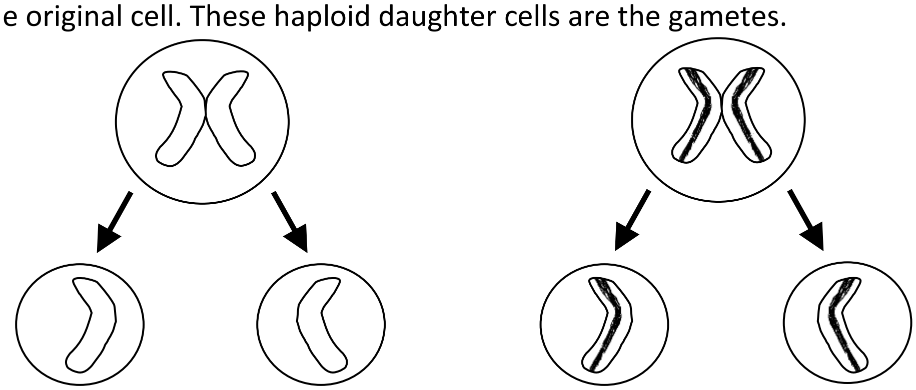


4. In this figure, label the diploid cell,

- the pair of homologous chromosomes in this diploid cell, and
- the two sister chromatids in one of these chromosomes.

5. Do the chromosomes in the two daughter cells produced by Meiosis I have the same alleles for each gene? Explain your reasoning.

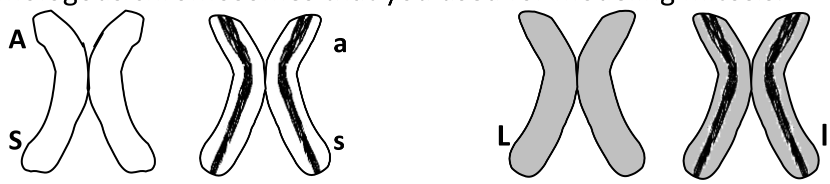
Meiosis II is like mitosis, since the sister chromatids of each chromosome are separated. As a result, each daughter cell gets one copy of one chromosome from the pair of homologous chromosomes that was in the original cell. These haploid daughter cells are the gametes.



6. Use asterisks to indicate the cells in this figure that represent the sperm produced by meiosis.

Modeling Meiosis

To model meiosis, you will be using the same model chromosomes for the same two pairs of homologous chromosomes that you used for modeling mitosis.



7. For each of the above chromosomes, write in the appropriate allele(s) on the unlabeled chromatid.

8. The genotype for a person with these chromosomes would be **AaSsLl**. What phenotypic characteristics would a person with this genotype have? Circle the appropriate phenotypic characteristics in this table.

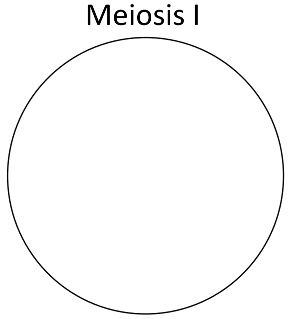
Genotype	→	**Protein**	→	**Phenotype** (characteristics)
AA or **Aa**	→	Enough normal enzyme to make melanin in skin and hair	→	Normal skin and hair color
aa	→	Defective enzyme for melanin production	→	Albino (very pale skin and hair color)
SS or **Ss**	→	Enough normal hemoglobin to prevent red blood cells from becoming sickle shaped	→	Normal blood (no sickle cell anemia)
ss	→	Sickle cell hemoglobin	→	Sickle cell anemia
LL or **Ll**	→	Defective enzyme (defective protein inactivates any normal enzyme)	→	Alcohol sensitive (skin flush and discomfort after drinking alcohol)
ll	→	Normal enzyme	→	Not alcohol sensitive

➢ Find two model chromosomes that have the two different alleles for the gene for sensitivity to alcohol (**L** and **l**). Use these model chromosomes to <u>model each step of meiosis</u> (see page 2). Repeat this until you are confident that you understand meiosis.

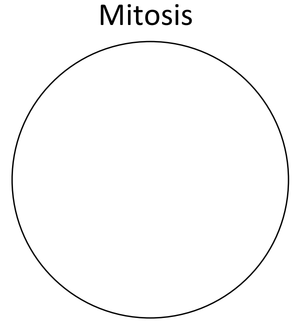
9. You have been modeling meiosis beginning with a diploid cell that has the genetic makeup **Ll.** The genetic makeup of the haploid gametes produced by meiosis is: __ **L** or **l** __ **LL** or **ll** __ **Ll**

10. Draw diagrams to show the difference in the way that a pair of homologous chromosomes is lined up in a cell at the beginning of Meiosis I vs. the beginning of Mitosis.

Meiosis I



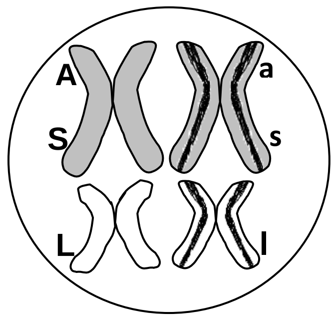
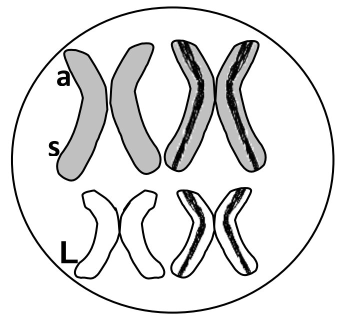
Mitosis



➢ Model mitosis and then meiosis. Repeat this comparison until you are confident that you understand the differences between mitosis and meiosis.

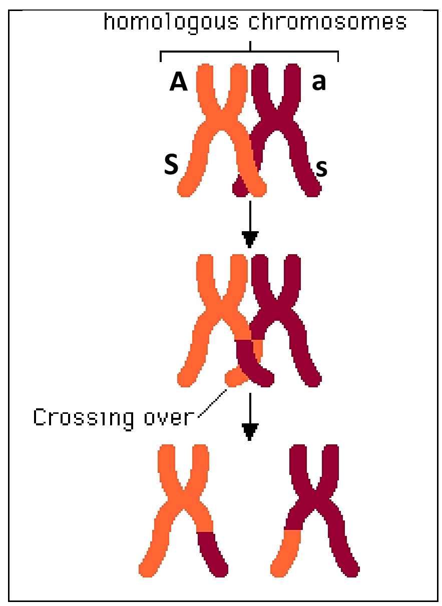
- Next, find two model chromosomes that have the two different alleles for the genes for albinism and sickle cell anemia. Use all four model chromosomes to model meiosis in a cell which has two pairs of homologous chromosomes and the genetic makeup **AaSsLl**. Begin by lining up the model chromosomes as shown in the diagram on the left in the chart. Model what would happen during meiosis, and write the genetic makeup of the resulting gametes in the chart.
- In the diagram on the right, label the alleles for the chromosomes on the right to show the other possible way of lining up the model chromosomes at the beginning of Meiosis I. Model meiosis for this way of lining up the model chromosomes. Show the genetic makeup of the resulting gametes.

11. Complete this chart to summarize the results of your modeling.

Chromosomes at the beginning of Meiosis I		
Genetic makeup of gametes	________ or ________	________ or ________

During Meiosis I each pair of homologous chromosomes lines up independently of how the other pairs of homologous chromosomes have lined up. This is called **independent assortment**. Because of independent assortment, different gametes have different combinations of the alleles on different chromosomes.

When a pair of homologous chromosomes is lined up next to each other during Meiosis I, the two homologous chromosomes can exchange parts of a chromatid. This is called **crossing over**.



12. Label the alleles for the genes for albinism and sickle cell anemia on each chromatid of the chromosomes in the bottom row of this diagram to show where these alleles are located after crossing over.

13. List the four different combinations of alleles for the genes for albinism and sickle cell anemia that would be observed in the haploid gametes produced by this type of meiosis with crossing over.

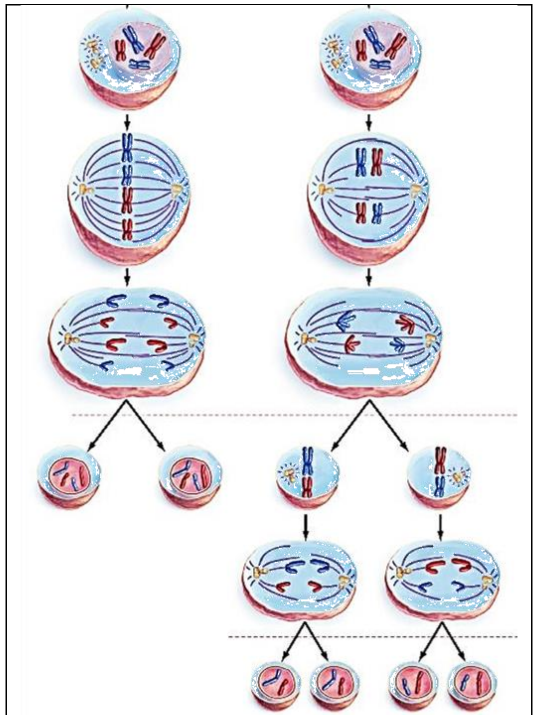
Notice that, because of crossing over, different gametes can have different combinations of alleles for two genes that are located far apart on the same chromosome.

14. Explain why different sperm or eggs produced by the same person have different genetic makeup (i.e. contain different alleles). Give three different reasons for the genetic diversity of different gametes produced by the same person.

15. In this figure, label the column that shows meiosis and the column that shows mitosis.

16. What are some similarities between cell division by mitosis and cell division by meiosis?

17. Describe the differences between mitosis and meiosis.



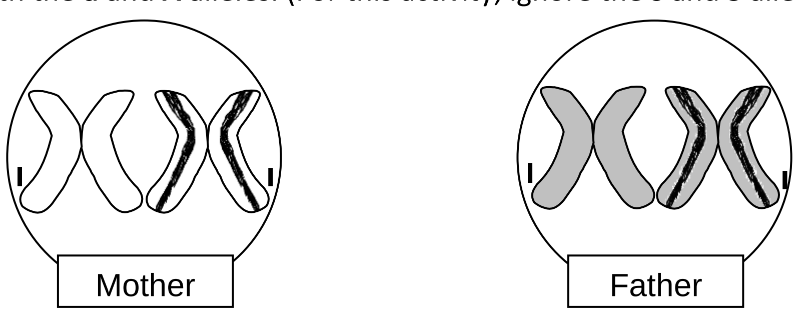
These diagrams show mitosis and meiosis after DNA has been replicated and wound tightly into sister chromatids. The dotted lines represent cytokinesis.

Analyzing Meiosis and Fertilization to Understand Inheritance

In this section you will investigate how events during meiosis and fertilization determine the genetic makeup of the zygote, which in turn determines the genetic makeup of the baby that develops from the zygote.

You already know that sisters or brothers can have different characteristics, even though they have the same parents. One major reason for these different characteristics is that the processes of meiosis and fertilization result in a different combination of alleles in each child.

To begin to understand this genetic variability, you will model meiosis and fertilization for a very simplified case where there is only one pair of homologous chromosomes per cell. In both the mother and the father, the two homologous chromosomes will have different alleles for a gene. Your group may have four model chromosomes with the **l** and **L** alleles (see figure) or you may have four model chromosomes with the **a** and **A** alleles. (For this activity, ignore the **s** and **S** alleles.)



Modeling Meiosis and Fertilization

- One of you should be the father and demonstrate how <u>meiosis</u> produces different types of sperm, and your partner should be the mother and demonstrate how meiosis produces different types of eggs. Write the genetic makeup of each type of egg and sperm in the white boxes in the chart.
- Use one of the sperm to <u>fertilize</u> one of the eggs to produce a zygote. The resulting zygote will have a pair of homologous chromosomes including one chromosome from the egg and one from the sperm. Write the genetic makeup of this zygote in the appropriate shaded box in the chart.
- Repeat this step to <u>produce as many different types of zygotes</u> as you can by pairing each type of sperm with each type of egg. Write the genetic makeup of each type of zygote in the appropriate box in the shaded area in the chart.

18. Record the results of your modeling in the chart. (If you have the model chromosomes with the genes for both albinism and sickle cell anemia, <u>*record only*</u> the results for the alleles for the albinism gene.)

Allele in each type of egg produced by meiosis in the mother

	____	____
____	______	______
____	______	______

Allele in each type of sperm produced by meiosis in the father

Alleles in the zygotes produced by fertilization

19. Each person began as a zygote. Explain why each person has the same genetic makeup as the zygote he or she developed from. (Hint: Review the information on page 1.)

20. In the above chart, write in the phenotypic characteristic for the mother, the father, and the person that will develop from each zygote. (If you used the model chromosomes with the **L** and **l** alleles, indicate whether or not each of these individuals is sensitive to alcohol. If you used the model chromosomes with the **A** and **a** alleles, indicate whether they have albinism.)
- Circle the zygotes that will develop into a person with the same phenotypic characteristic as the parents. Use an * to mark the zygote that will develop into a person who will have a different phenotypic characteristic that neither parent has.

21. Explain why many children have the same phenotypic characteristics as their parents.

- Explain how a child can have a different phenotypic characteristic that neither parent has.

Next, we will consider the genetic variability and phenotypic variability of offspring produced by the same parents when we consider three different genes, each with two alleles.

22. As you saw on page 4, **AaSsLl** parents can produce multiple different types of gametes with different combinations of the alleles for the albinism gene, sickle cell gene, and alcohol sensitivity gene. Consult question 11 on page 4 and list here the genetic makeup of the four different types of gametes that each parent can produce as a result of independent assortment with no crossing over.

As a result of crossing over, **AaSsLl** parents can produce four additional types of gametes:

AsL, **Asl**, **aSL** and **aSl.**

Obviously, fertilization of the eight different types of eggs by the eight different types of sperm can result in offspring with many different genotypes. In the next question, you will describe the outcomes for fertilization of a few of the possible types of eggs by one of the possible types of sperm.

23. Complete the following chart to describe the genotype and phenotype of some of the possible outcomes of fertilization between the different types of possible eggs and sperm.

Alleles in egg	Alleles in sperm	Genotype of zygote	Phenotype of person who will develop from this zygote (See the table in question 8 on page 3.)
ASL	**asl**		
ASl	**asl**		
aSl	**asl**		

This illustrates how meiosis and fertilization can produce zygotes with multiple different combinations of alleles and multiple different phenotypes, even when we consider only three genes with two alleles each. The actual amount of <u>genetic diversity</u> possible in the children produced by one couple is much greater, since each person has thousands of genes on 23 pairs of homologous chromosomes. As a result of independent assortment of the 23 pairs of homologous chromosomes, more than 8 million different combinations of chromosomes could be found in the different eggs or sperm produced by one person. If each different type of egg from one mother could be fertilized by each different type of sperm from one father, they could produce zygotes with approximately 70 trillion different combinations of chromosomes! Crossing over results in an even greater amount of genetic diversity. You can see why no two people are genetically alike, except for identical twins who both develop from the same zygote.

24. Sally and Harry fall in love. They introduce Sally's identical twin, Emily, to Harry's identical twin, Ken. Soon there is a double wedding where Sally marries Harry and Emily marries Ken. Both Sally and Emily get pregnant. They wonder "Will their babies look exactly alike?" Answer their question, and explain your reasoning.

A Mistake in Meiosis Can Cause Down Syndrome

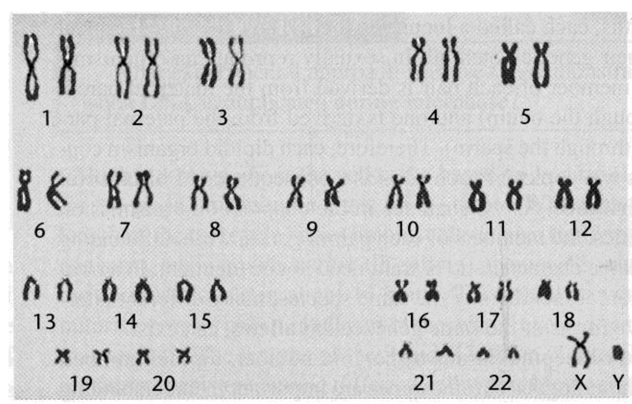
You have seen that normal meiosis and fertilization can produce a lot of genetic variability in the different children produced by the same parents. Additional genetic variability can result from mistakes in DNA replication (which can cause mutations) or mistakes in meiosis. For example, when meiosis does not happen perfectly, the chromosomes are not divided equally between the daughter cells produced by meiosis, so an egg or a sperm may receive two copies of the same chromosome.

25. If a human egg receives two copies of a chromosome, and this egg is fertilized by a normal sperm, how many copies of this chromosome would there be in the resulting zygote? ____
- How many copies of this chromosome would there be in each cell in the resulting embryo? ____

When a cell has three copies of a chromosome, the extra copies of the genes on this chromosome result in abnormal cell function and abnormal embryonic development. To understand how an extra copy of one chromosome could result in abnormalities, remember that each chromosome has genes with the instructions to make specific types of proteins, so the extra chromosome could result in too many copies of these specific proteins. Think about what would happen if you added too much milk to a box of macaroni and cheese. The mac and cheese would have too much liquid and be runny instead of creamy. Cells are much more complicated than mac and cheese, and a cell cannot function properly when there are too many copies of some types of proteins due to an extra copy of one of the chromosomes.

When the cells in an embryo do not function properly, the embryo develops abnormalities. For example, some babies are born with an extra copy of chromosome 21 in each cell. This results in **Down syndrome** with multiple abnormalities, including mental retardation, a broad flat face, a big tongue, short height, and often heart defects.

This figure shows a karyotype from a normal boy. A **karyotype** is a photograph of a magnified view of the chromosomes from a human cell, with pairs of homologous chromosomes arranged next to each other and numbered. In the karyotype, each chromosome has double copies of its DNA, contained in a pair of sister chromatids linked at a centromere.



26. Label the sister chromatids in chromosome 3 in the karyotype.
- Draw in an extra chromosome 21 to show the karyotype of a boy with Down syndrome.

27. In most cases, an embryo which has an extra chromosome in each cell develops such severe abnormalities that the embryo dies, resulting in a miscarriage. For example, an extra copy of any of the chromosomes in the top row of the karyotype results in such severe abnormalities that the embryo always dies. In contrast, an extra copy of chromosome 21 results in less severe abnormalities so the embryo can often survive to be born as a baby with Down syndrome. What do you think is the reason why a third copy of chromosome 1, 2, 3, 4 or 5 results in more severe abnormalities than a third copy of chromosome 21?