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## Chapter 11

### Introduction to Genetics

### What You'll Learn

You will identify the basic concepts of genetics.

You will examine the process of meiosis.

### Section Objectives:

- Relate Mendel's two laws to the results he obtained in his experiments with garden peas.
- Predict the possible offspring of a genetic cross by using a Punnett square.

### Why Mendel Succeeded

- It was not until the mid-nineteenth century that Gregor Mendel, an Austrian monk, carried out important studies of **heredity**—the passing on of characteristics from parents to offspring.
- Characteristics that are inherited are called **traits**.

### Why Mendel Succeeded

- Mendel was the first person to succeed in predicting how traits are transferred from one generation to the next.
- A complete explanation requires the careful study of **genetics**—the branch of biology that studies heredity.

### Mendel chose his subject carefully

- Mendel chose to use the garden pea in his experiments for several reasons.



- Garden pea plants reproduce sexually, which means that they produce male and female sex cells, called **gametes**.

### Mendel chose his subject carefully

- The male gamete forms in the pollen grain, which is produced in the male reproductive organ.
- The female gamete forms in the female reproductive organ.
- In a process called **fertilization**, the male gamete unites with the female gamete.
- The resulting fertilized cell, called a **zygote** (ZI goht), then develops into a seed.

### Mendel chose his subject carefully

- The transfer of pollen grains from a male reproductive organ to a female reproductive organ in a plant is called **pollination**.

### Mendel chose his subject carefully



- When he wanted to breed, or cross, one plant with another, Mendel opened the petals of a flower and removed the male organs.

### Mendel chose his subject carefully

- He then dusted the female organ with pollen from the plant he wished to cross it with.



### Mendel chose his subject carefully

- This process is called cross-pollination.
- By using this technique, Mendel could be sure of the parents in his cross.

### Mendel was a careful researcher

- He studied only one trait at a time to control variables, and he analyzed his data mathematically.
- The tall pea plants he worked with were from populations of plants that had been tall for many generations and had always produced tall offspring.

### Mendel was a careful researcher

- Such plants are said to be true breeding for tallness.
- Likewise, the short plants he worked with were true breeding for shortness.

### Mendel's Monohybrid Crosses

- A **hybrid** is the offspring of parents that have different forms of a trait, such as tall and short height.
- Mendel's first experiments are called monohybrid crosses because *mono* means "one" and the two parent plants differed from each other by a single trait—height.

### The first generation

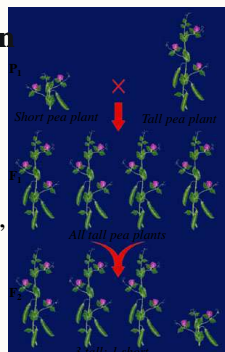
- Mendel selected a six-foot-tall pea plant that came from a population of pea plants, all of which were over six feet tall.
- He cross-pollinated this tall pea plant with pollen from a short pea plant.
- All of the offspring grew to be as tall as the taller parent.

### The second generation

- Mendel allowed the tall plants in this first generation to self-pollinate.
- After the seeds formed, he planted them and counted more than 1000 plants in this second generation.
- Three-fourths of the plants were as tall as the tall plants in the parent and first generations.

### The second generation















- One-fourth of the offspring were as short as the short plants in the parent generation.
- In the second generation, tall and short plants occurred in a ratio of about three tall plants to one short plant.



### The second generation

- The original parents, the true-breeding plants, are known as the P<sub>1</sub> generation.
- The offspring of the parent plants are known as the F<sub>1</sub> generation.
- When you cross two F<sub>1</sub> plants with each other, their offspring are the F<sub>2</sub> generation.

### The second generation

	Seed shape	Seed color	Flower color	Flower position	Pod color	Pod shape	Plant height
Dominant trait	 round	 yellow	 purple	 axial (side)	 green	 inflated	 tall
Recessive trait	 wrinkled	 green	 white	 terminal (top)	 yellow	 constricted	 short

### The second generation

- In every case, he found that one trait of a pair seemed to disappear in the F<sub>1</sub> generation, only to reappear unchanged in one-fourth of the F<sub>2</sub> plants.

### The rule of unit factors

- Mendel concluded that each organism has two factors that control each of its traits.
- We now know that these factors are genes and that they are located on chromosomes.
- Genes exist in alternative forms. We call these different gene forms **alleles**.

### The rule of unit factors

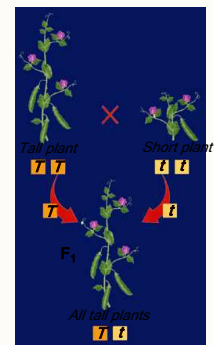
- An organism's two alleles are located on different copies of a chromosome—one inherited from the female parent and one from the male parent.

### The rule of dominance

- Mendel called the observed trait **dominant** and the trait that disappeared **recessive**.
- Mendel concluded that the allele for tall plants is dominant to the allele for short plants.

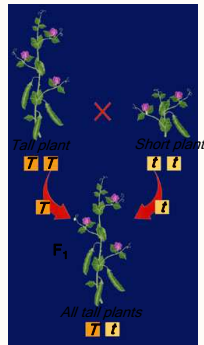
### The rule of dominance

- When recording the results of crosses, it is customary to use the same letter for different alleles of the same gene.



### The rule of dominance

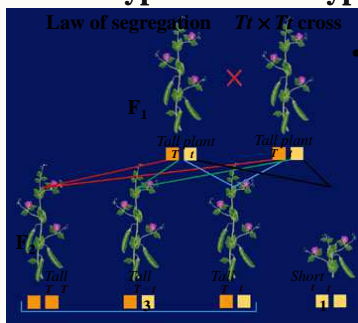
- An uppercase letter is used for the dominant allele and a lowercase letter for the recessive allele.
- The dominant allele is always written first.



### The law of segregation

- The law of segregation states that every individual has two alleles of each gene and when gametes are produced, each gamete receives one of these alleles.
- During fertilization, these gametes randomly pair to produce four combinations of alleles.

### Phenotypes and Genotypes



- Two organisms can look alike but have different underlying allele combinations.

### Phenotypes and Genotypes

- The way an organism looks and behaves is called its **phenotype**.
- The allele combination an organism contains is known as its **genotype**.
- An organism's genotype can't always be known by its phenotype.

### Phenotypes and Genotypes

- An organism is **homozygous** for a trait if its two alleles for the trait are the same.
- The true-breeding tall plant that had two alleles for tallness ( $TT$ ) would be homozygous for the trait of height.

### Phenotypes and Genotypes

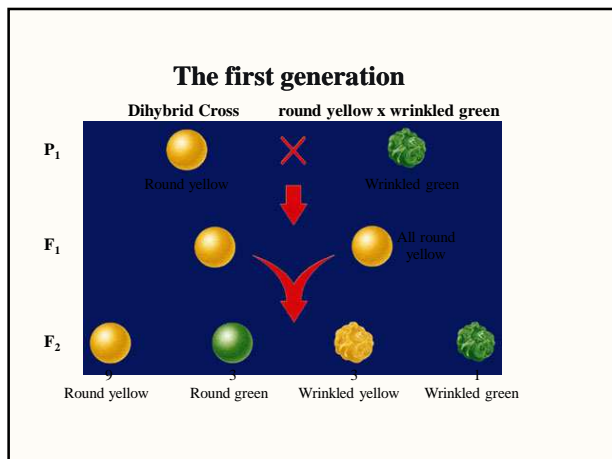
- An organism is **heterozygous** for a trait if its two alleles for the trait differ from each other.
- Therefore, the tall plant that had one allele for tallness and one allele for shortness ( $Tt$ ) is heterozygous for the trait of height.

## Mendel's Dihybrid Crosses

- Mendel performed another set of crosses in which he used peas that differed from each other in two traits rather than only one.
- Such a cross involving two different traits is called a dihybrid cross.

## The first generation

- Mendel took true-breeding pea plants that had round yellow seeds ( $RRYY$ ) and crossed them with true-breeding pea plants that had wrinkled green seeds ( $rryy$ ).
- He already knew the round-seeded trait was dominant to the wrinkled-seeded trait.
- He also knew that yellow was dominant to green.



## The second generation

- Mendel then let the F<sub>1</sub> plants pollinate themselves.
- He found some plants that produced round yellow seeds and others that produced wrinkled green seeds.
- He also found some plants with round green seeds and others with wrinkled yellow seeds.

## The second generation

- He found they appeared in a definite ratio of phenotypes—9 round yellow: 3 round green: 3 wrinkled yellow: 1 wrinkled green.

## The law of independent assortment

- Mendel's second law states that genes for different traits—for example, seed shape and seed color—are inherited independently of each other.
- This conclusion is known as the **law of independent assortment**.

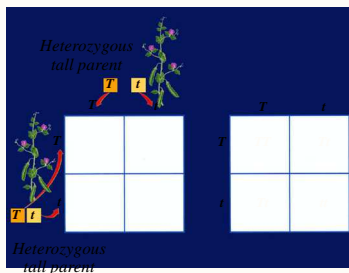
## Punnett Squares

- In 1905, Reginald Punnett, an English biologist, devised a shorthand way of finding the expected proportions of possible genotypes in the offspring of a cross.
- This method is called a Punnett square.

## Punnett Squares

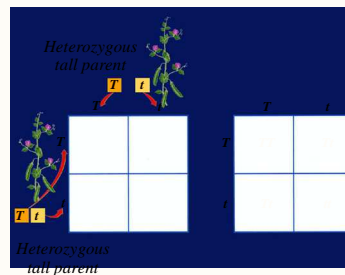
- If you know the genotypes of the parents, you can use a Punnett square to predict the possible genotypes of their offspring.

## Monohybrid crosses



- A Punnett square for this cross is two boxes tall and two boxes wide because each parent can produce two kinds of gametes for this trait.

## Monohybrid crosses

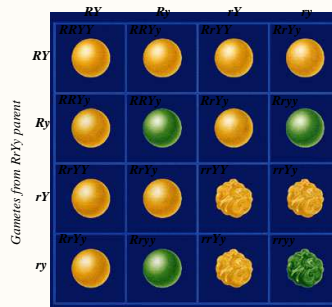


- The two kinds of gametes from one parent are listed on top of the square, and the two kinds of gametes from the other parent are listed on the left side.

## Monohybrid crosses

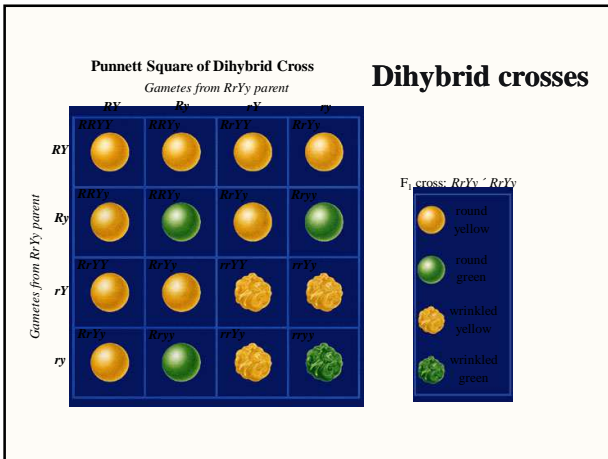
- It doesn't matter which set of gametes is on top and which is on the side.
- Each box is filled in with the gametes above and to the left side of that box. You can see that each box then contains two alleles—one possible genotype.
- After the genotypes have been determined, you can determine the phenotypes.

## Punnett Square of Dihybrid Cross



## Dihybrid crosses

- A Punnett square for a dihybrid cross will need to be four boxes on each side for a total of 16 boxes.

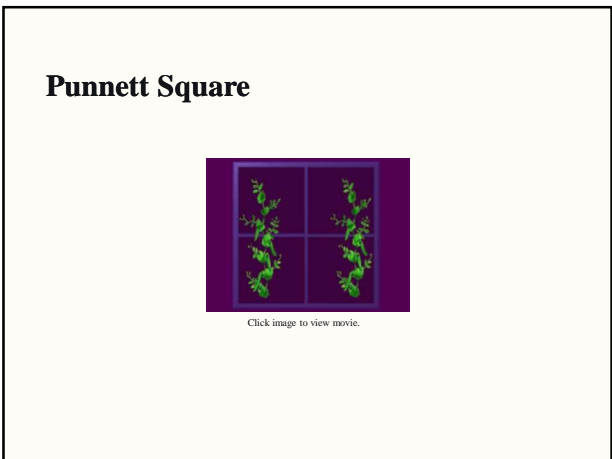
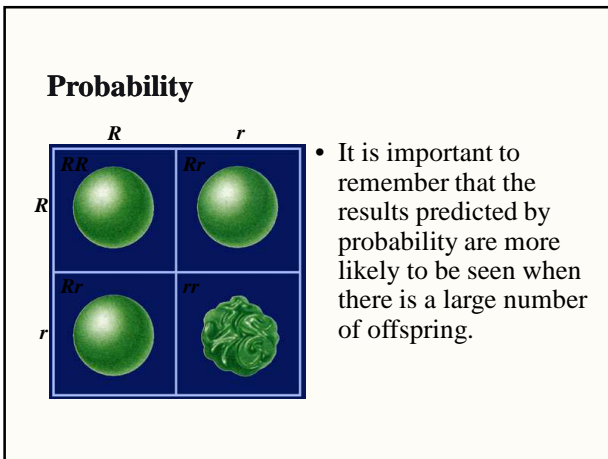
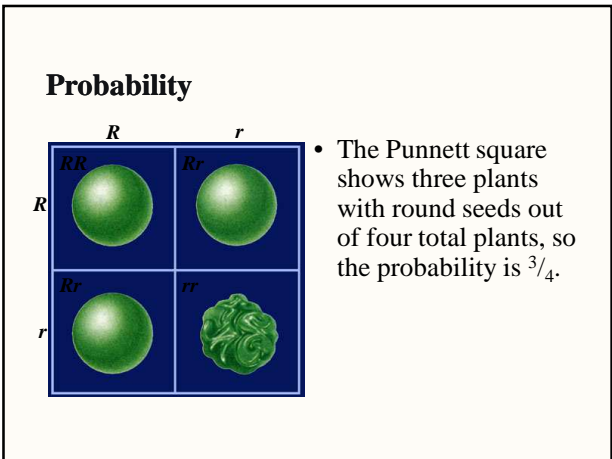


**Probability**

- In reality you don't get the exact ratio of results shown in the square.
- That's because, in some ways, genetics is like flipping a coin—it follows the rules of chance.
- The probability or chance that an event will occur can be determined by dividing the number of desired outcomes by the total number of possible outcomes.

**Probability**

- A Punnett square can be used to determine the probability of getting a pea plant that produces round seeds when two plants that are heterozygous (*Rr*) are crossed.





## Section Objectives

- Analyze how meiosis maintains a constant number of chromosomes within a species.
- Infer how meiosis leads to variation in a species.
- Relate Mendel's laws of heredity to the events of meiosis.

## Genes, Chromosomes, and Numbers

- Genes do not exist free in the nucleus of a cell; they are lined up on chromosomes.
- Typically, a chromosome can contain a thousand or more genes along its length.

## Diploid and haploid cells

- In the body cells of animals and most plants, chromosomes occur in pairs.
- A cell with two of each kind of chromosome is called a **diploid** cell and is said to contain a diploid, or  $2n$ , number of chromosomes.

## Diploid and haploid cells

- This pairing supports Mendel's conclusion that organisms have two factors—alleles—for each trait.
- Organisms produce gametes that contain one of each kind of chromosome.
- A cell containing one of each kind of chromosome is called a **haploid** cell and is said to contain a haploid, or  $n$ , number of chromosomes.

## Diploid and haploid cells

Organism	Body Cell ( $2n$ )	Gamete ( $n$ )
Garden pea	14	7
Tomato	24	12
Apple	34	17
Chimpanzee	48	24
Alder's tongue fern	1260	630

- This fact supports Mendel's conclusion that parent organisms give one allele for each trait to each of their offspring.

## Diploid and haploid cells

Organism	Body Cell ( $2n$ )	Gamete ( $n$ )
Garden pea	14	7
Tomato	24	12
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Alder's tongue fern	1260	630

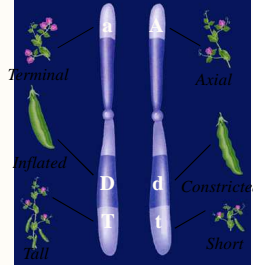
- This table shows the diploid and haploid number of chromosomes of some species.

### Homologous chromosomes

- The two chromosomes of each pair in a diploid cell are called **homologous chromosomes**.
- Each pair of homologous chromosomes has genes for the same traits.

### Homologous chromosomes

Homologous Chromosome 4



- On homologous chromosomes, these genes are arranged in the same order, but because there are different possible alleles for the same gene, the two chromosomes in a homologous pair are not always identical to each other.

### Why meiosis?

- When cells divide by mitosis, the new cells have exactly the same number and kind of chromosomes as the original cells.
- Imagine if mitosis were the only means of cell division.
- Each pea plant parent, which has 14 chromosomes, would produce gametes that contained a complete set of 14 chromosomes.

### Why meiosis?

- The  $F_1$  pea plants would have cell nuclei with 28 chromosomes, and the  $F_2$  plants would have cell nuclei with 56 chromosomes.

### Why meiosis?

- There must be another form of cell division that allows offspring to have the same number of chromosomes as their parents.
- This kind of cell division, which produces gametes containing half the number of chromosomes as a parent's body cell, is called **meiosis**.

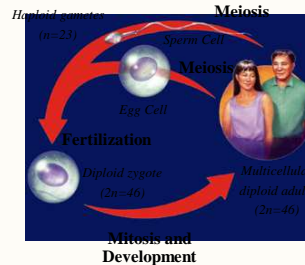
### Why meiosis?

- Meiosis consists of two separate divisions, known as meiosis I and meiosis II.
- Meiosis I begins with one diploid ( $2n$ ) cell.
- By the end of meiosis II, there are four haploid ( $n$ ) cells.

### Why meiosis?

- These haploid cells are called sex cells—gametes.
- Male gametes are called **sperm**.
- Female gametes are called **eggs**.
- When a sperm fertilizes an egg, the resulting zygote once again has the diploid number of chromosomes.

### Why meiosis?

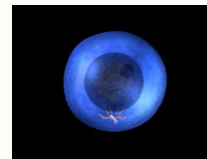


- This pattern of reproduction, involving the production and subsequent fusion of haploid sex cells, is called **sexual reproduction**.

### The Phases of Meiosis

- During meiosis, a spindle forms and the cytoplasm divides in the same ways they do during mitosis.
- However, what happens to the chromosomes in meiosis is very different.

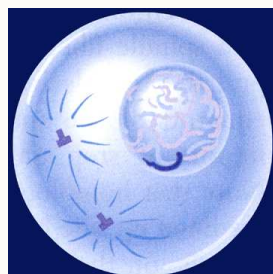
### The Phases of Meiosis



Click image to view movie.

### Interphase

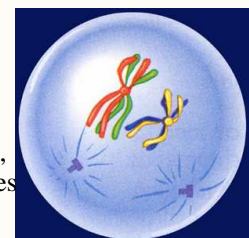
- During interphase, the cell replicates its chromosomes.
- After replication, each chromosome consists of two identical sister chromatids, held together by a centromere.



Interphase

### Prophase I

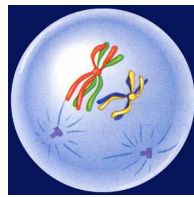
- The chromosomes coil up and a spindle forms.
- As the chromosomes coil, homologous chromosomes line up with each other gene by gene along their length, to form a four-part structure called a tetrad.



Prophase I

### Prophase I

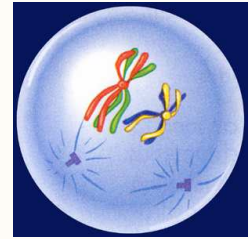
- The chromatids in a tetrad pair tightly.
- In fact, they pair so tightly that non-sister chromatids from homologous chromosomes can actually break and exchange genetic material in a process known as **crossing over**.



*Prophase I*

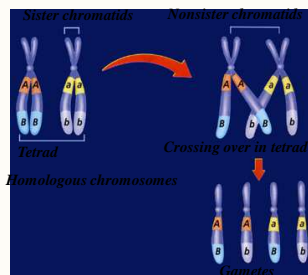
### Prophase I

- Crossing over can occur at any location on a chromosome, and it can occur at several locations at the same time.



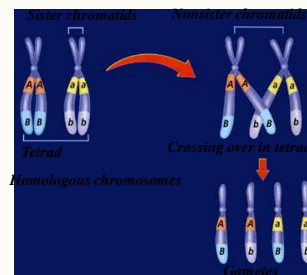
*Prophase I*

### Prophase I



- It is estimated that during prophase I of meiosis in humans, there is an average of two to three crossovers for each pair of homologous chromosomes.

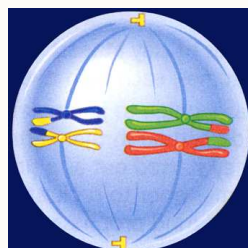
### Prophase I



- Crossing over results in new combinations of alleles on a chromosome.

### Metaphase I

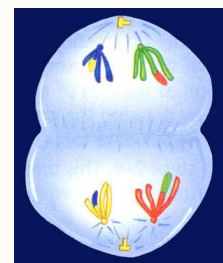
- During metaphase I, the centromere of each chromosome becomes attached to a spindle fiber.
- The spindle fibers pull the tetrads into the middle, or equator, of the spindle.



*Metaphase I*

### Anaphase I

- Anaphase I begins as homologous chromosomes, each with its two chromatids, separate and move to opposite ends of the cell.
- This critical step ensures that each new cell will receive only one chromosome from each homologous pair.



*Anaphase I*

### Telophase I

- Events occur in the reverse order from the events of prophase I.
- The spindle is broken down, the chromosomes uncoil, and the cytoplasm divides to yield two new cells.



*Telophase I*

### Telophase I

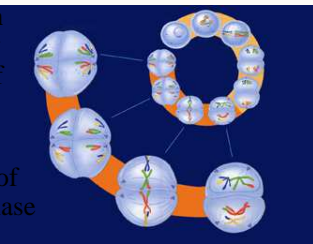
- Each cell has half the genetic information of the original cell because it has only one chromosome from each homologous pair.



*Telophase I*

### The phases of meiosis II

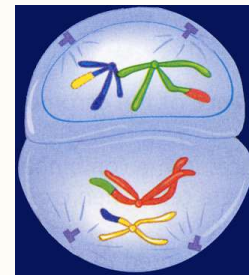
- The second division in meiosis is simply a mitotic division of the products of meiosis I.
- Meiosis II consists of prophase II, metaphase II, anaphase II, and telophase II.



*Meiosis II*

### The phases of meiosis II

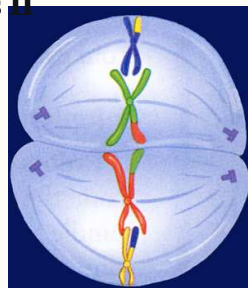
- During prophase II, a spindle forms in each of the two new cells and the spindle fibers attach to the chromosomes.



*Prophase II*

### The phases of meiosis II

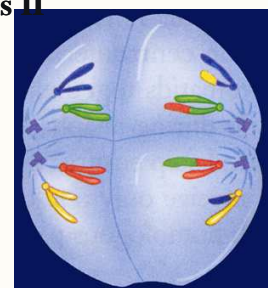
- The chromosomes, still made up of sister chromatids, are pulled to the center of the cell and line up randomly at the equator during metaphase II.



*Metaphase II*

### The phases of meiosis II

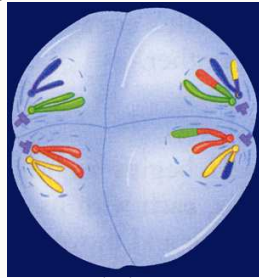
- Anaphase II begins as the centromere of each chromosome splits, allowing the sister chromatids to separate and move to opposite poles.



*Anaphase II*

### The phases of meiosis II

- Finally nuclei, reform, the spindles break down, and the cytoplasm divides during telophase II.



### The phases of meiosis II

- At the end of meiosis II, four haploid cells have been formed from one diploid cell.
- These haploid cells will become gametes, transmitting the genes they contain to offspring.

### Meiosis Provides for Genetic Variation

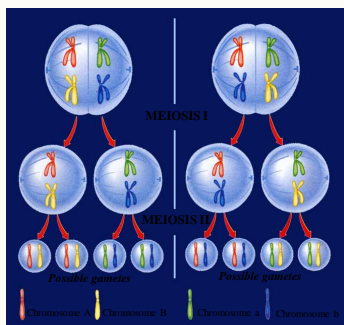
- Cells that are formed by mitosis are identical to each other and to the parent cell.
- Crossing over during meiosis, however, provides a way to rearrange allele combinations.
- Thus, variability is increased.

### Genetic recombination

- Reassortment of chromosomes and the genetic information they carry, either by crossing over or by independent segregation of homologous chromosomes, is called **genetic recombination**.

### Genetic recombination

- It is a major source of variation among organisms.



### Meiosis explains Mendel's results

- The segregation of chromosomes in anaphase I of meiosis explains Mendel's observation that each parent gives one allele for each trait at random to each offspring, regardless of whether the allele is expressed.

### **Meiosis explains Mendel's results**

- The segregation of chromosomes at random during anaphase I also explains how factors, or genes, for different traits are inherited independently of each other.

### **Nondisjunction**

- The failure of homologous chromosomes to separate properly during meiosis is called **nondisjunction**.

### **Nondisjunction**

- Recall that during meiosis I, one chromosome from each homologous pair moves to each pole of the cell.
- In nondisjunction, both chromosomes of a homologous pair move to the same pole of the cell.

### **Nondisjunction**

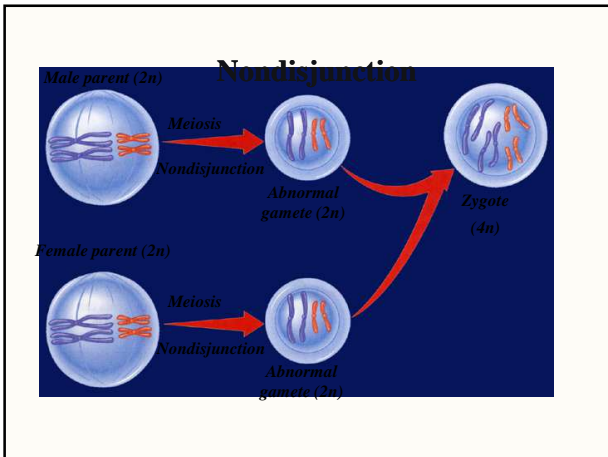
- The effects of nondisjunction are often seen after gametes fuse.
- When a gamete with an extra chromosome is fertilized by a normal gamete, the zygote will have an extra chromosome.
- This condition is called trisomy.

### **Nondisjunction**

- Although organisms with extra chromosomes often survive, organisms lacking one or more chromosomes usually do not.
- When a gamete with a missing chromosome fuses with a normal gamete during fertilization, the resulting zygote lacks a chromosome.
- This condition is called monosomy.

### **Nondisjunction**

- An example of monosomy that is not lethal is Turner syndrome, in which human females have only a single X chromosome instead of two.



### Nondisjunction

- When a gamete with an extra set of chromosomes is fertilized by a normal haploid gamete, the offspring has three sets of chromosomes and is triploid.
- The fusion of two gametes, each with an extra set of chromosomes, produces offspring with four sets of chromosomes—a tetraploid.

### Chromosome Mapping

- Crossing over produces new allele combinations. Geneticists use the frequency of crossing over to map the relative positions of genes on a chromosome.

The diagram shows four genes labeled A, B, C, and D on a chromosome. Recombination frequencies are indicated by double-headed arrows: A-B is 50%, A-D is 10%, B-C is 5%, and C-D is 35%.

### Chromosome Mapping

- Genes that are farther apart on a chromosome are more likely to have crossing over occur between them than are genes that are closer together.

The diagram shows four genes labeled A, B, C, and D on a chromosome. Recombination frequencies are indicated by double-headed arrows: A-B is 50%, A-D is 10%, B-C is 5%, and C-D is 35%.

### Chromosome Mapping

- Suppose there are four genes—A, B, C, and D—on a chromosome.

The diagram shows a horizontal line representing a chromosome with four genes labeled A, B, C, and D. The distances between the genes are indicated by double-headed arrows: A-B is 10, D-C is 35, and C-B is 5. A long arrow below the line indicates the total length of the chromosome is 50.

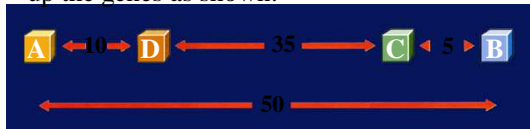
### Chromosome Mapping

- Geneticists determine that the frequencies of recombination among them are as follows: between A and B—50%; between A and D—10%; between B and C—5%; between C and D—35%.
- The recombination frequencies can be converted to map units: A-B = 50; A-D = 10; B-C = 5; C-D = 35.



### Chromosome Mapping

- These map units are not actual distances on the chromosome, but they give relative distances between genes. Geneticists line up the genes as shown.



### Chromosome Mapping

- The genes can be arranged in the sequence that reflects the recombination data.
- This sequence is a chromosome map.



### Polyploidy

- Organisms with more than the usual number of chromosome sets are called polyploids.
- Polyploidy is rare in animals and almost always causes death of the zygote.

### Polyploidy



- However, polyploidy frequently occurs in plants.
- Many polyploid plants are of great commercial value.

### Gene Linkage and Maps

- If genes are close together on the same chromosome, they usually are inherited together.
- These genes are said to be linked.

### Gene Linkage and Maps

- Linked genes may become separated on different homologous chromosomes as a result of crossing over.
- When crossing over produces new gene combinations, geneticists can use the frequencies of these new gene combinations to make a chromosome map showing the relative locations of the genes.