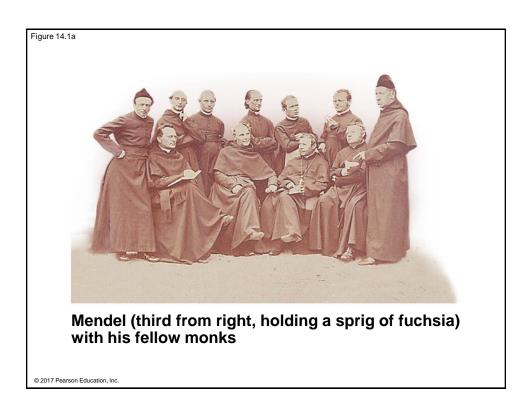


Chapter 14

Mendel and the Gene Idea

Lecture Presentations by Nicole Tunbridge and Kathleen Fitzpatrick



Concept 14.1: Mendel used the scientific approach to identify two laws of inheritance

 Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments

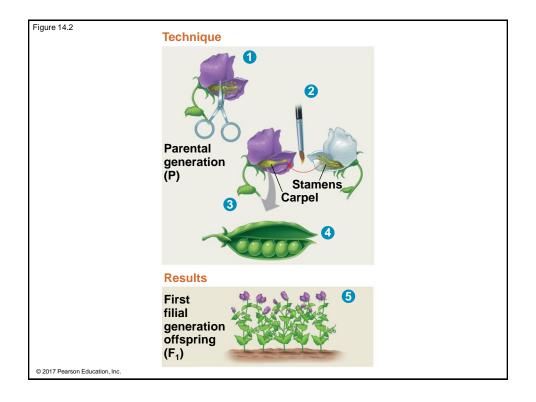
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Mendel's Experimental, Quantitative Approach

- Mendel's approach allowed him to deduce principles that had remained elusive to others
- A heritable feature that varies among individuals (such as flower color) is called a character
- Each variant for a character, such as purple or white color for flowers, is called a trait
- Peas were available to Mendel in many different varieties

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- Other advantages of using peas
 - Short generation time
 - Large numbers of offspring
 - Mating could be controlled; plants could be allowed to self-pollinate or could be cross-pollinated



- Mendel chose to track only those characters that occurred in two distinct alternative forms
- He also started with varieties that were truebreeding (plants that produce offspring of the same variety when they self-pollinate)

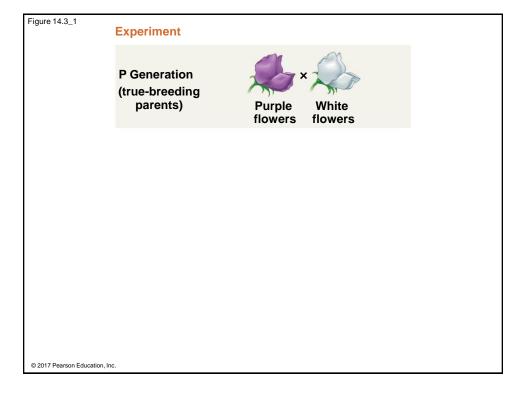
- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called hybridization
- The true-breeding parents are the P generation
- The hybrid offspring of the P generation are called the F₁ generation
- When F₁ individuals self-pollinate or cross-pollinate with other F₁ hybrids, the F₂ generation is produced

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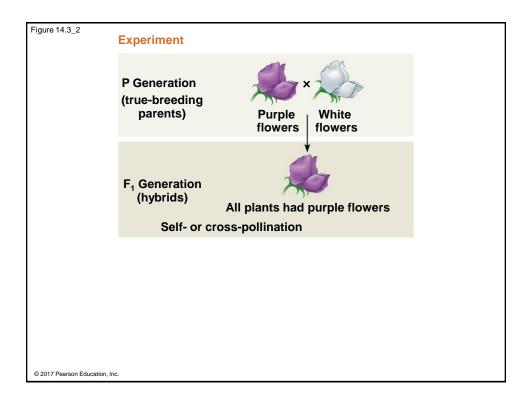
The Law of Segregation

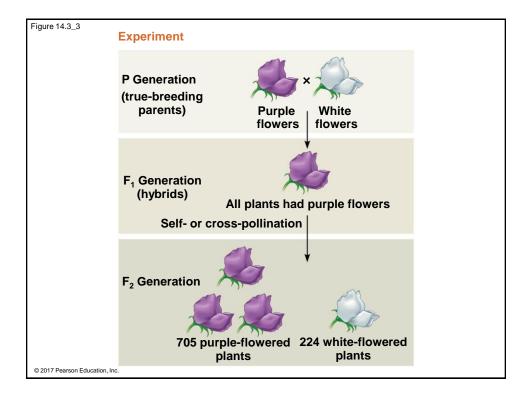
- When Mendel crossed contrasting, true-breeding white- and purple-flowered pea plants, all of the F₁ hybrids were purple
- When Mendel crossed the F₁ hybrids, many of the F₂ plants had purple flowers, but some had white
- Mendel discovered a ratio of about three purple flowers to one white flower in the F₂ generation

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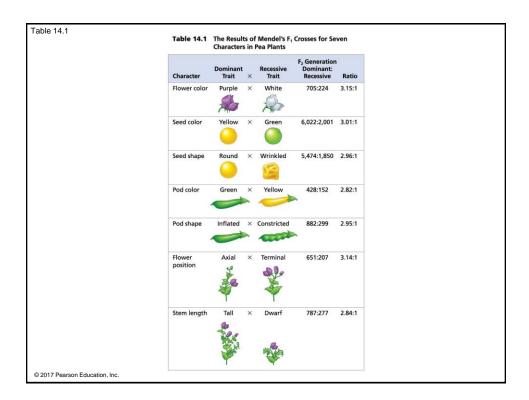




- Mendel reasoned that only the purple flower factor was affecting flower color in the F₁ hybrids
- Mendel called the purple flower color a dominant trait and the white flower color a recessive trait
- The factor for white flowers was not diluted or destroyed because it reappeared in the F₂ generation

- Mendel observed the same pattern of inheritance in six other pea plant characters, each represented by two traits
- What Mendel called a "heritable factor" is what we now call a gene

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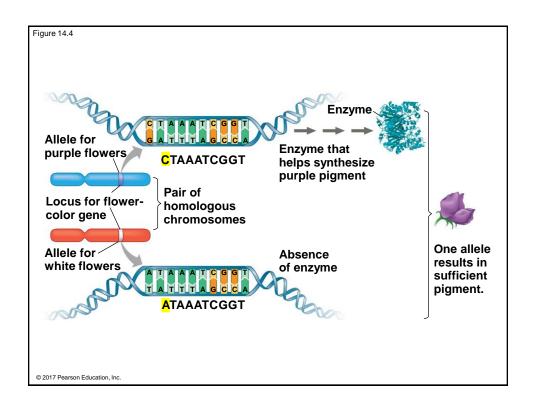


Mendel's Model

- Mendel developed a hypothesis to explain the 3:1 inheritance pattern he observed in F₂ offspring
- Four related concepts make up this model
- These concepts can be related to what we now know about genes and chromosomes

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- First: alternative versions of genes account for variations in inherited characters
- For example, the gene for flower color in pea plants exists in two versions, one for purple flowers and the other for white flowers
- These alternative versions of a gene are called alleles
- Each gene resides at a specific locus on a specific chromosome



- Second: for each character, an organism inherits two alleles, one from each parent
- Mendel made this deduction without knowing about chromosomes
- The two alleles at a particular locus may be identical, as in the true-breeding plants of Mendel's P generation
- Or the two alleles at a locus may differ, as in the F₁ hybrids

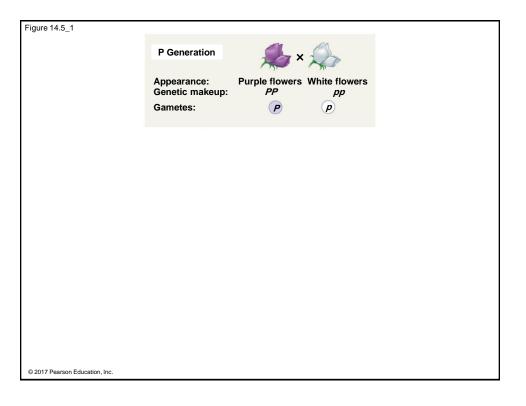
- Third: if the two alleles at a locus differ, then one (the dominant allele) determines the organism's appearance, and the other (the recessive allele) has no noticeable effect on appearance
- In the flower-color example, the F₁ plants had purple flowers because the allele for that trait is dominant

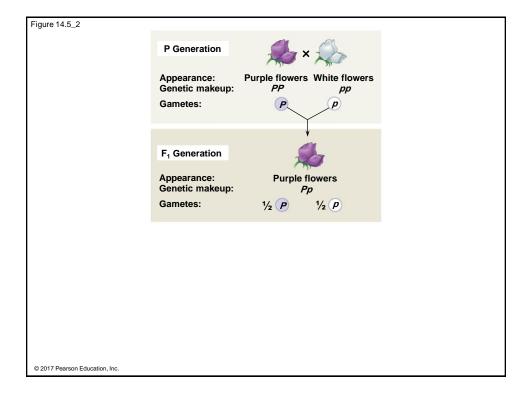
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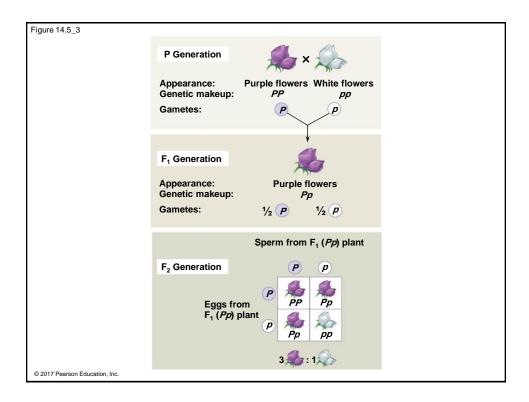
- Fourth (the law of segregation): the two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes
- Thus, an egg or a sperm gets only one of the two alleles that are present in the organism
- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis

- The model accounts for the 3:1 ratio observed in the F₂ generation of Mendel's crosses
- Possible combinations of sperm and egg can be shown using a **Punnett square**
- A capital letter represents a dominant allele, and a lowercase letter represents a recessive allele

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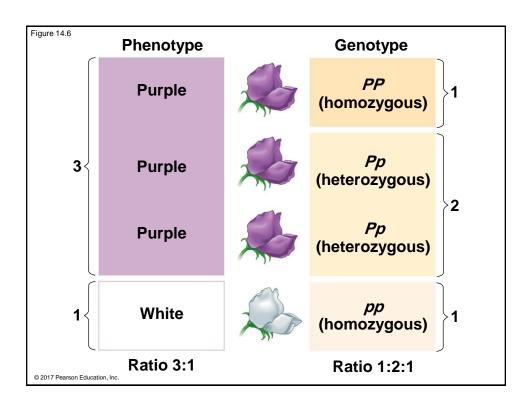


Useful Genetic Vocabulary

- An organism with two identical alleles for a character is called a homozygote
- It is said to be homozygous for the gene controlling that character
- An organism with two different alleles for a gene is a heterozygote and is said to be heterozygous for the gene controlling that character
- Unlike homozygotes, heterozygotes are not truebreeding

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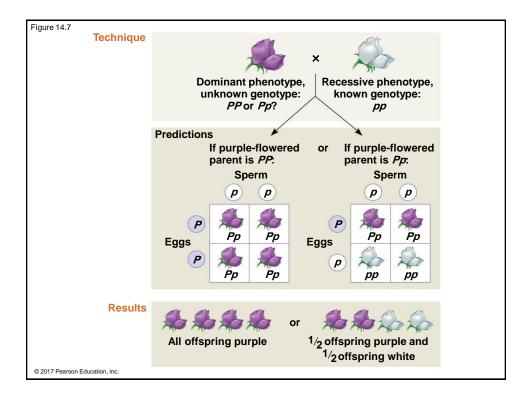
- An organism's traits do not always reveal its genetic composition
- Therefore, we distinguish between an organism's phenotype, or physical appearance, and its genotype, or genetic makeup
- In the example of flower color in pea plants, PP and Pp plants have the same phenotype (purple) but different genotypes



The Testcross

- An individual with the dominant phenotype could be either homozygous dominant or heterozygous
- To determine the genotype we can carry out a testcross: breeding the mystery individual with a homozygous recessive individual
- If any offspring display the recessive phenotype, the mystery parent must be heterozygous

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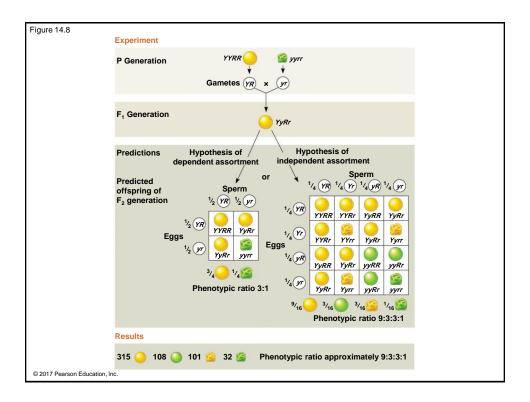
The Law of Independent Assortment

- Mendel derived the law of segregation by following a single character
- The F₁ offspring produced in this cross were monohybrids, heterozygous for one character
- A cross between such heterozygotes is called a monohybrid cross

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- Mendel identified his second law of inheritance by following two characters at the same time
- Crossing two true-breeding parents differing in two characters produces dihybrids in the F₁ generation, heterozygous for both characters
- A dihybrid cross, a cross between F₁ dihybrids, can determine whether two characters are transmitted to offspring as a package or independently

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- Using a dihybrid cross, Mendel developed the law of independent assortment
- It states that each pair of alleles segregates independently of any other pair of alleles during gamete formation
- This law applies only to genes on different, nonhomologous chromosomes or those far apart on the same chromosome
- Genes located near each other on the same chromosome tend to be inherited together

Concept 14.3: Inheritance patterns are often more complex than predicted by simple Mendelian genetics

- The relationship between genotype and phenotype is rarely as simple as in the pea plant characters
 Mendel studied
- Many heritable characters are not determined by only one gene with two alleles
- However, the basic principles of segregation and independent assortment apply even to more complex patterns of inheritance

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Extending Mendelian Genetics for a Single Gene

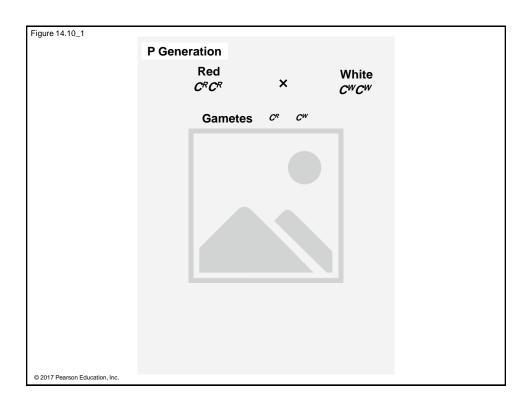
- Inheritance of characters by a single gene may deviate from simple Mendelian patterns in the following situations:
 - When alleles are not completely dominant or recessive
 - When a gene has more than two alleles
 - When a gene produces multiple phenotypes

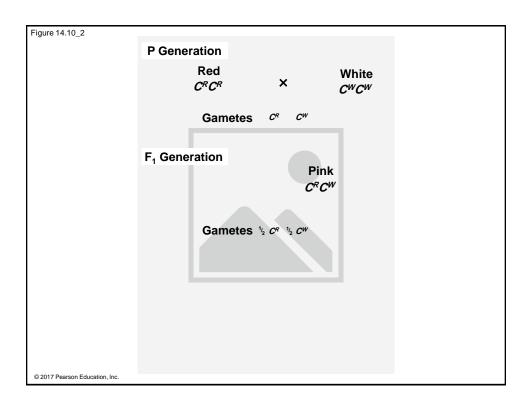
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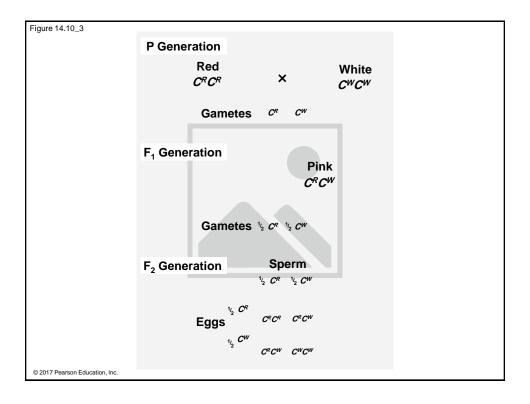
Degrees of Dominance

- Complete dominance occurs when phenotypes of the heterozygote and dominant homozygote are identical
- In incomplete dominance, the phenotype of F₁
 hybrids is somewhere between the phenotypes of
 the two parental varieties
- In codominance, two dominant alleles affect the phenotype in separate, distinguishable ways

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The Relationship Between Dominance and Phenotype

- In the case of pea shape, the dominant allele codes for an enzyme that converts an unbranched form of starch in the seed to a branched form
- The recessive allele codes for a defective form of the enzyme, which leads to an accumulation of unbranched starch
- This causes water to enter the seed, which then wrinkles as it dries

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- Tay-Sachs disease is fatal; a dysfunctional enzyme causes an accumulation of lipids in the brain
 - At the organismal level, the allele is recessive
 - At the biochemical level, the phenotype (i.e., the enzyme activity level) is incompletely dominant
 - At the molecular level, the alleles are codominant

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Frequency of Dominant Alleles

- Dominant alleles are not necessarily more common in populations than recessive alleles
- One baby out of 400 in the United States is born with extra fingers or toes
- This condition, polydactyly, is caused by a dominant allele, found much less frequently in the population than the recessive allele

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Multiple Alleles

- Most genes exist in populations in more than two allelic forms
- For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme that attaches A or B carbohydrates to red blood cells: I^A, I^B, and i
- The enzyme encoded by the I^A allele adds the A carbohydrate, whereas the enzyme encoded by the I^B allele adds the B carbohydrate; the enzyme encoded by the I allele adds neither

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(a) The three al carbohydra		ABO blood g	roups and	u tileli
Allele	I ^A	I ^B	I ^B	
Carbohydrate	Α	В		none
(b) Blood grou	p genotypes	and phenoty	pes	
Genotype	<i>I^AI^A</i> or <i>I^Ai</i>	<i>I^BI^B</i> or <i>I^Bi</i>	I ^A I ^B	ii
Red blood cell with surface carbohydrates				
Phenotype (blood group)	Α	В	АВ	0

Pleiotropy

- Most genes have multiple phenotypic effects, a property called **pleiotropy**
- For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as cystic fibrosis and sickle-cell disease

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Extending Mendelian Genetics for Two or More Genes

- Some traits may be determined by two or more genes
- In epistasis, one gene affects the phenotype of another due to interaction of their gene products
- In polygenic inheritance, multiple genes independently affect a single trait

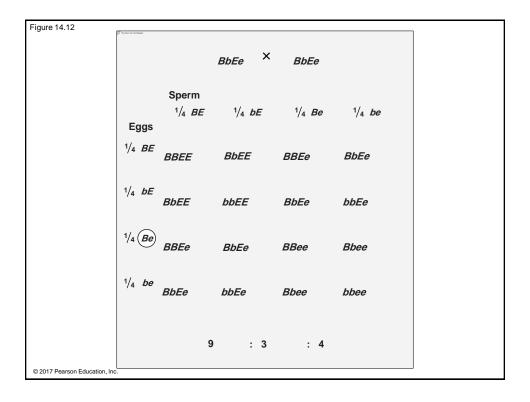
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Epistasis

- In epistasis, expression of a gene at one locus alters the phenotypic expression of a gene at a second locus
- For example, in Labrador retrievers and many other mammals, coat color depends on two genes
- One gene determines the pigment color (with alleles B for black and b for brown)
- The other gene (with alleles E for color and e for no color) determines whether the pigment will be deposited in the hair

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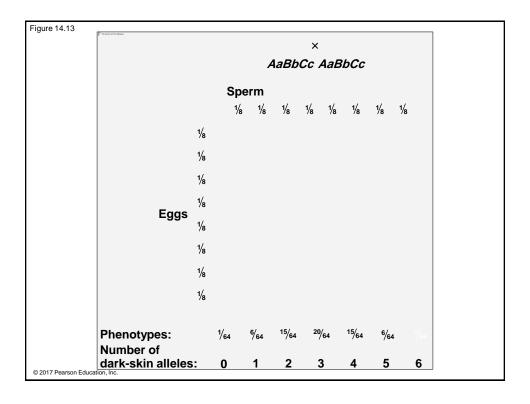
- If heterozygous black labs (genotype BbEe) are mated, we might expect the dihybrid F₂ ratio of 9:3:3:1
- However, a Punnett square shows that the phenotypic ratio will be 9 black to 3 chocolate to 4 yellow labs
- Epistatic interactions produce a variety of ratios, all of which are modified versions of 9:3:3:1



Polygenic Inheritance

- Quantitative characters are those that vary in the population along a continuum
- Quantitative variation usually indicates polygenic inheritance, an additive effect of two or more genes on a single phenotype
- Height is a good example of polygenic inheritance:
 Over 180 genes affect height
- Skin color in humans is also controlled by many separately inherited genes

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A Mendelian View of Heredity and Variation

- An organism's phenotype includes its physical appearance, internal anatomy, physiology, and behavior
- An organism's phenotype reflects its overall genotype and unique environmental history

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