LECTURE PRESENTATIONS For CAMPBELL BIOLOGY, NINTH EDITION Jane B. Reece, Lisa A. Urry, Michael L. Cain, Steven A. Wasserman, Peter V. Minorsky, Robert B. Jackson

Chapter 14

Mendel and the Gene Idea

Lectures by Erin Barley Kathleen Fitzpatrick

Overview: Drawing from the Deck of Genes

- What genetic principles account for the passing of traits from parents to offspring?
- The "blending" hypothesis is the idea that genetic material from the two parents blends together (like blue and yellow paint blend to make green)

- The "particulate" hypothesis is the idea that parents pass on discrete heritable units (genes)
- This hypothesis can explain the reappearance of traits after several generations
- Mendel documented a particulate mechanism through his experiments with garden peas



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

Mendel's Experimental, Quantitative Approach

- Advantages of pea plants for genetic study
 - There are many varieties with distinct heritable features, or characters (such as flower color); character variants (such as purple or white flowers) are called traits
 - Mating can be controlled
 - Each flower has sperm-producing organs (stamens) and egg-producing organ (carpel)
 - Cross-pollination (fertilization between different plants) involves dusting one plant with pollen from another

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- Mendel chose to track only those characters that occurred in two distinct alternative forms
- He also used varieties that were true-breeding (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 crosspollinate with other F₁ hybrids, the F₂ generation is produced

The Law of Segregation

- When Mendel crossed contrasting, truebreeding 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 to one, purple to white flowers, in the F₂ generation

EXPERIMENT

Figure 14.3-3



- 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

Table 14.1

| Character | rs in Pea Pla | nts | | crosses for 3 | even |
|--------------------|-------------------|-----|--------------------|--|--------|
| Character | Dominant Trait | × | Recessive Trait | F ₂ Generation Dominant: Recessive | Ratio |
| Flower color | Purple | × | White | 705:224 | 3.15:1 |
| Flower position | Axial | × | Terminal | 651:207 | 3.14:1 |
| Seed color | Yellow | × | Green | 6,022:2,001 | 3.01:1 |
| Seed shape | Round | × | Wrinkled | 5,474:1,850 | 2.96:1 |
| Pod shape | Inflated | × | Constricted | 882:299 | 2.95:1 |
| Pod color | Green | × | Yellow | 428:152 | 2.82:1 |
| Stem length | Tall | × | Dwarf | 787:277 | 2.84:1 |

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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

- 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 now 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 the role of chromosomes
- The two alleles at a particular locus may be identical, as in the true-breeding plants of Mendel's P generation
- Alternatively, 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

- Fourth: (now known as 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

- Mendel's segregation model accounts for the 3:1 ratio he observed in the F₂ generation of his numerous crosses
- The possible combinations of sperm and egg can be shown using a **Punnett square**, a diagram for predicting the results of a genetic cross between individuals of known genetic makeup
- A capital letter represents a dominant allele, and a lowercase letter represents a recessive allele



Useful Genetic Vocabulary

- An organism with two identical alleles for a character is said to be homozygous for the gene controlling that character
- An organism that has two different alleles for a gene is said to be heterozygous for the gene controlling that character
- Unlike homozygotes, heterozygotes are not true-breeding

- Because of the different effects of dominant and recessive alleles, 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



Ratio 1:2:1

The Testcross

- How can we tell the genotype of an individual with the dominant phenotype?
- Such an individual could be either homozygous dominant or heterozygous
- The answer is to 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



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, individuals that are heterozygous for one character
- A cross between such heterozygotes is called a monohybrid cross

- 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

Figure 14.8

EXPERIMENT



RESULTS

- Using a dihybrid cross, Mendel developed the law of independent assortment
- The law of independent assortment states that each pair of alleles segregates independently of each other pair of alleles during gamete formation
- Strictly speaking, 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

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

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

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

Figure 14.10-3



The Relation Between Dominance and Phenotype

- A dominant allele does not subdue a recessive allele; alleles don't interact that way
- Alleles are simply variations in a gene's nucleotide sequence
- For any character, dominance/recessiveness relationships of alleles depend on the level at which we examine the phenotype

- 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

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 (I) 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

| (a) The three alleles for the ABO blood groups and their carbohydrates | | | | | | | |
|--|-----|------------|------|--|--|--|--|
| Allele / ^A | | J B | i | | | | |
| Carbohydrate | Α 🛆 | B ဝ | none | | | | |

| (b) Blood group genotypes and phenotypes | | | | | | | | |
|--|---|---|--------------|----|--|--|--|--|
| Genotype | I ^A I ^A or I ^A i | l ^B l ^B or l ^B i | I ^I₿ | ii | | | | |
| Red blood cell appearance | | | | | | | | |
| Phenotype (blood group) | Α | В | AB | Ο | | | | |