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Chapter 2: Mendelian Inheritance

Key Terms

Allele
Binomial expansion equation
Characters
Crossed
Dihybrid cross
Dominant
Empirical approach
    Cross-fertilization
Forked-line method
Gene
Generations
    F_2 generation
Genotype
Heterozygous
Hybridization
Hypothesis testing
Law of Segregation
Monohybrid cross
Multinomial expansion equation
Nonparentals
Ovaries
P values
Particulate theory of inheritance
Phenotype
Probability
Punnett square
Recessive

Anthers
Blending inheritance
Chi square test
Degrees of freedom
Dihybrid testcross
Eggs
Fertilization
    Self-fertilization
Gamete
Genetic recombination
    F_1 generation
    Parental (P) generation
Goodness of fit
Homozygous
Hybrids
Law of Independent Assortment
Loss-of-function alleles
Monohybrids
Multiplication method
Null hypothesis
Ovules
Pangenesis
Pedigree analysis
Pollen grains
Product rule
Random sampling error
Segregate

Single-factor cross
Stigma
Sum rule
True-breeding line
Variant
Sperm
Strain
Trait
Two-factor cross

Chapter Outline

Introduction

1. The concept of heredity predates the time of Mendel. The ancient Greek philosopher Hippocrates (around 400 B.C.E) proposed the concept of pangenesis where "seeds" are produced by all parts of the body and are transmitted to the offspring.
2. Greek theories persisted in some form for over 2,000 years.
   a. homunculus – a tiny, fully formed human that lived within the sperm cells
   b. ovists – egg was responsible for human characteristics of the offspring
   c. spermists – sperm was responsible for the human characteristics of the offspring
3. Work of Joseph Kölreuter (18^{th} century) supported the blending theory of inheritance.
   a. The idea that the factors that dictate heredity are blended together from one generation to the next.
   b. By the mid-19^{th} century it was believed that these blended traits could change over generations.

2.1 Mendel's Study of Pea Plants

Learning Outcomes:

1. Explain the characteristics of pea plants that make them a suitable organism to study genetically.
2. Outline the steps that Mendel followed to make crosses between different strains of pea plants.
3. List the seven characteristics of pea plants that Mendel chose to study.

1. Facts regarding Mendel's (1822-1884) life (Figure 2.1):
   a. ordained Augustinian minister (1847) from the monastery of St. Thomas
   b. interested in teaching, but failed license exams because he was weak in physics and mathematics.
   c. enrolled at the University of Vienna to study physics and mathematics
   d. learned that natural laws can be stated as simple mathematical relationships
2. Mendel's experimental approach to heredity began in 1856 and lasted only 8 years.
   a. worked in a small (115x23 foot) plot
   b. kept detailed records that included quantitative data
   c. 1866 paper, "Experiments on Plant Hybrids," was not well recognized by the scientific community
3. Mendel's work was independently rediscovered in 1900 by Hugh de Vries (Holland), Carl Correns (Germany), and Erich von Tschermak (Austria).

Mendel Chose Pea Plants as His Experimental Organism

1. Prior to Mendel, plant breeders had crossed (mated) distinct individuals to produce

hybrids in a process called hybridization

2. Mendel believed that the patterns of traits in these hybridization experiments were rooted in physical laws that could be explained by mathematical principles.

3. Mendel used the garden pea (Pisum sativum) as his experimental system.
   a. existed in several varieties with easily recognizable characteristics
   b. pea plants can be easily mated (see #4)
4. Plant reproduction occurs by pollination (Figure 2.2).
   a. Male gametes (sperm) are produced within pollen grains, which are formed within the anthers of the plant.
   b. Female gametes are produced within the ovules, which are formed within the ovaries of the plant.
   c. Pollen grains first land on the stigma. This is followed by the formation of a pollen tube, which delivers the sperm to the egg cell.
5. With plants, self-fertilization (sperm and egg from the same individual) is possible. The structure of a pea plant favors self-fertilization since the stamens are covered by a protective petal (the keel).
6. Cross-fertilization involves the use of two parents. The large flowers of the pea plant make it possible to remove stamens from the flower, preventing self-fertilization.
   a. allowed selective breeding of pea plants to produce desired hybrids (Figure 2.3)

Mendel Studied Seven Characteristics That Bred True

1. Mendel chose varieties of pea plants that had distinct morphological differences in their traits (characters).
2. These lines were true-breeding, or did not show any variation in the trait over time.
3. Mendel identified seven traits that existed in two variants (Figure 2.4)
   a. flower color, flower position, seed color, seed shape, pod shape, pod color, height
4. Mendel conducted crosses between variants of a single trait.
   a. called a monohybrid or single-factor cross

2.2 Mendel's Study of Pea Plants

<u>Learning Outcomes:</u>

1. Analyze Mendel's experiments involving single-factor crosses.
2. Define the law of segregation and explain how it is related to gamete formation and fertilization.
3. Predict the outcome of single-factor crosses using a Punnett square.

Mendel Followed the Outcome of a Single Character for Two Generations

1. Mendel conducted experiments to determine the mathematical relationship between hereditary traits.
   a. this process is called the empirical approach
2. In a single-factor cross the parental generation (P generation) is a true-breeding line for the variant of the trait being studied (purple flower color, tall height, etc.)
   a. The offspring of the parental generation are called the first filial (F_1) generation.
   b. The offspring of the F_1 generation are called the second filial (F_2) generation.
3. Mendel's single-factor cross followed the following steps (Figure 2.5):

a. Two true-breeding lines were crosses that differed only for one trait.
b. The F_1 generation are allowed to self-fertilize, producing an F_2 generation.

4. The data (pg. 22) from these experiments yielded the following information regarding inheritance:
   a. The F_1 generation did not exhibit blending. Rather, it showed that one of the traits was dominant over the other (recessive) trait.
   b. The dominant trait was always displayed in the F_1 generation. In the F_2 generation the dominant trait was present in the majority (75%) of the plants, while the recessive trait was present in the minority (25%) of the plants.
   c. The genetic information is passed on from one generation to the next as "unit factors," which are now called genes. This supported the particulate theory of inheritance which suggests that the units governing traits remain unchanged (unblended) from generation to generation.
   d. The 3:1 ratio of dominant to recessive offspring in the F_2 generation suggested that each parent possesses two traits, which segregate during the formation of gametes.
   e. Mendel was the first to apply quantitative analyses to the study of inheritance.

Mendel's 3:1 Phenotypic Ratio Is Consistent with the Law of Segregation

1. Mendel was unaware of the concept of DNA or genes.
   a. the term gene was first introduced by Wilhelm Johannsen
   b. genes reside on chromosomes
   c. the variants in the traits are due to versions of the gene called an allele
2. Mendel's law of segregation: *The two copies of a gene segregate from each other during transmission from parent to offspring*.
3. Alleles for a gene are typically represented using uppercase (for the dominant trait) and lowercase (for the recessive trait) letters (Figure 2.6).
4. The genotype is the genetic combination of an individual.
   a. homozygous indicates individuals with two identical alleles
   b. heterozygous indicates individuals with two different alleles
5. The observable characteristics of an organism are called the phenotype.

A Punnett Square Can Be Used to Predict the Outcome of Crosses

1. Allows you to predict the types of offspring the parents will produce and the proportion of the trait in the offspring.
2. Steps for preparing a Punnett Square
   a. write down the genotypes of both parents
   b. write down the possible gametes that each parent can make
   c. create an empty Punnett square in which the number of columns equals the number of male gametes and the number of rows equals the number of female gametes
   d. fill in the possible genotypes of the offspring by combining the alleles of the gametes in the empty boxes
   e. determine the relative proportions of genotypes and phenotypes of the offspring

2.3 Law of Independent Assortment

Learning Outcomes:

1. Analyze Mendel's experiments involving two-factor crosses.
2. Define the law of independent assortment.
3. Predict the outcome of two-factor crosses using a Punnett square.
4. Define a loss-of-function allele and explain why such alleles are useful to study.

Mendel Also Analyzed Crosses Involving Two Different Characters

1. Mendel conducted crosses using two-factors to see if additional information regarding patterns of inheritance could be determined. These are now known as dihybrid crosses.
2. In a two-factor cross there are two possibilities of how the traits can be inherited (Figure 2.7)
   a. They may be linked to one another and inherited as a single unit.
   b. They may be unlinked and assort themselves independently during inheritance.
3. Mendel's experimental system followed the same pattern as the single-factor cross (Figure 2.8).
   a. Two true-breeding lines were selected that were different with regards to two different traits (seed shape, seed color).
   b. The F_1 plants were allowed to self-fertilize.
   c. The phenotypic ratio of the F_2 generation was determined.
4. Mendel's experimental data (page 26) indicated the following:
   a. The F_2 generation of seeds possessed a 9:3:3:1 phenotypic ratio, not the 1:2:1 ratio expected by a linked model.
   b. Some seeds of the F_2 generation were nonparentals, thus further disproving that the traits were linked.
5. Mendel's law of independent assortment states that *two different genes will randomly assort their alleles during the formation of haploid reproductive cells*.
6. Independent assortment means that a single individual can produce a vast array of genetically different gametes (Figure 2.9).
7. An offspring receiving a different combination of alleles than are seen in the parental generation is known as genetic recombination.

A Punnett Square Can Also Be Used to Solve Independent Assortment Problems

1. For a two-factor cross, each parent can produce four types of gametes. Thus the Punnett square would have 16 cells (4 rows x 4 columns) (Figure 2.10).
2. Process is the same as the single-factor Punnett square.
3. Punnett squares are not practical for more than two traits. The forked-line method or multiplication method are more useful for larger crosses.
4. The dihybrid test cross involves using an individual who is homozygous recessive for both traits in the cross.

Modern Geneticists Are Often Interested in the Relationship Between the Molecular Expression of Genes and the Outcome of Traits

1. Genes encode proteins that perform the majority of cellular functions. Proteins influence an individual's expressed traits.
2. The study of loss-of-function alleles can assist geneticists in understanding the

relationship between a gene and a phenotype.

a. The white flower color in Mendel's pea plants is an example of a loss-of-function allele (unknown to Mendel).

2.4 Studying Inheritance Patterns in Humans

Learning Outcomes:

1. Describe the features of a pedigree.
2. Analyze a pedigree to determine if a trait or disease is dominant or recessive.

Pedigree Analysis Can Be Used to Follow the Mendelian Inheritance of Traits in Humans

1. Mendel's approach works when large numbers of offspring can be produced and matings can be controlled. This is unethical in humans and impractical for many organisms.
2. A pedigree (family tree) is used to examine inheritance patterns in humans (Figure 2.11).
   a. often used to study human disease
3. Example is the study of cystic fibrosis (CF) in humans
   a. gene for CF encodes a protein called CFTR (cystic fibrosis transmembrane conductance regulator
   b. altered CFTR effects the ionic balance across the membrane
   c. the mutated CFTR associated with CF is a recessive trait

2.5 Probability and Statistics

Learning Outcomes:

1. Define probability.
2. Predict the outcome of crosses using the product rule and binomial expansion equation.
3. Evaluate the validity of a hypothesis using a chi square test.

1. Mendel's work demonstrated that laws of inheritance can be used to predict the outcomes of genetic crosses.
2. Probability calculations are used to predict the outcomes of genetic crosses.

Probability Is the Likelihood That an Event Will Occur

1. Probability is the chance that a future event will occur

$$\text{Probability} = \frac{\text{number of times an event occurs}}{\text{total number of events}}$$

2. The accuracy of a probability calculation is determined by sample size. The larger the sample size, the more accurately the calculation reflects the larger population.
3. The deviation between the observed outcome and the expected outcome is called the random sampling error.

The Sum Rule Can Be Used to Predict the Occurrence of Mutually Exclusive Events

1. A probability calculation involving the sum rule states that *the probability that one of two or more mutually exclusive independent events will occur is equal to the sum of the individual probabilities of the events*. For example: the probability a baby will be

a boy OR a girl.

2. Based on an “or” event.
   a. What is the probability that event A or B will occur?
      $P = p(A) + p(B)$

The Product Rule Can Be Used to Predict the Probability of Independent Events

1. A probability calculation involving the product rule states that *the probability that two or more independent events will occur is equal to the product of their individual probabilities*. For example: A pea plant will be tall AND purple-flowered.
2. Based on an “and” event.
3. What is the probability that event A and B will occur?
   $P = p(A) \times p(B)$
4. The product rule can replace the use of the Punnett square for crosses involving two or more genes.

The Binomial Expansion Equation Can Be Used to Predict the Probability of an Unordered Combination of Events

1. Binomial expansion is used to give the probability that a certain proportion of offspring will be produced, regardless of order.
2. Equation (pg. 33)
   $$P = \frac{n!}{x!(n-x!)} p^{x} q^{n-x}$$
   P = probability that an unordered number of events will occur
   n = total number of events
   x = the total number of events in one category
   p = individual probability of x
   q = individual probability of the other category
   ! = factorial
3. The multinomial expansion equation is needed to solve unordered genetic problems that involve three or more phenotypic categories.

The Chi-Square Test Can Be Used to Test the Validity of a Genetic Hypothesis

1. Involved in hypothesis testing, such as determining if the data from a given genetic cross is consistent with a certain pattern of inheritance.
   a. tests the goodness of fit between the observed data and that predicted by the hypothesis
   b. This is sometimes called a null hypothesis because it assumes there is no real difference between the observed and expected values.
   c. does not prove that a hypothesis is correct or incorrect
2. Formula (pg. 34)
   $$X^2 = \sum \frac{(O - E)}{E}$$
   O = observed data in each category
   E = expected data in each category
   ∑ = sum of each category
3. Steps of a Chi square test.

   a. Propose a hypothesis that allows us to calculate the expected values based on Mendel's laws.
   b. Calculate the expected values for each phenotype.
   c. Apply the Chi square formula, using the data for the observed and expected values as calculated in step 2.
   d. Interpret the results using a Chi square table.
4. P values (Table 2.1) allow us to determine the likelihood that the variation indicated by the Chi square calculation is due to random chance alone.
   a. Hypotheses are usually rejected if the chi-square value results in a P value less than 0.05
5. Degrees of freedom (df) is the measure of the number of categories in the experiment that are independent of one another.
   a. represented as n-1, where n equals the number of categories