

# CHAPTER 11 MENDELIAN PATTERNS OF INHERITANCE

## Chapter Outline

### 11.1 Gregor Mendel

- A. Gregor Mendel
  1. Mendel was an Austrian monk.
  2. Mendel formulated two fundamental laws of heredity in the early 1860s.
  3. He had previously studied science and mathematics at the University of Vienna.
  4. At time of his research, he was a substitute science teacher at a local technical high school.
- B. Blending Concept of Inheritance
  1. This theory stated that offspring would have traits intermediate between those of the parents.
  2. Red and white flowers produce pink flowers; any return to red or white offspring was considered instability in genetic material.
  3. Charles Darwin wanted to develop a theory of evolution based on hereditary principles; blending theory was of no help.
    - a. A blending theory did not account for variation and could not explain species diversity.
    - b. The particulate theory of inheritance proposed by Mendel can account for presence of differences among members of a population generation after generation.
    - c. Mendel's work was unrecognized until 1900; Darwin was never able to use it to support his theory of evolution.
- C. Mendel's Experimental Procedure
  1. Mendel had a mathematical background and did a statistical study.
  2. He prepared his experiments carefully and conducted preliminary studies.
    - a. He chose the garden pea, *Pisum sativum*, because peas were easy to cultivate, had a short generation time, and could be cross-pollinated by hand.
    - b. From many varieties, Mendel chose 22 true-breeding varieties for his experiments.
    - c. **True-breeding** varieties had all offspring like the parents and like each other.
    - d. Mendel studied simple traits (e.g., seed shape and color, flower color, etc.).
  3. Mendel traced inheritance of individual traits and kept careful records of numbers.
  4. He used his understanding of mathematical principles of probability to interpret results.

### 11.2 One-Trait Inheritance

- A. Cross-pollination Monohybrid Crosses
  1. Mendel confirmed that his tall plants always had tall offspring, etc. before crossing two different strains to produce a hybrid by conducting **reciprocal crosses**.
  2. A **hybrid** is the product of parent organisms that are true-breeding for different forms of one trait.
  3. A **monohybrid cross** is between two parent organisms true-breeding for two distinct forms of one trait.
  4. Mendel tracked each trait through two generations.
    - a. **P generation** is the parental generation in a breeding experiment.
    - b. **F<sub>1</sub> generation** is the first-generation offspring in a breeding experiment.
    - c. **F<sub>2</sub> generation** is the second-generation offspring in a breeding experiment.
    - d. He also performed reciprocal crosses of pollen on stigmas (e.g., tall-with-short and short-with-tall).
- B. Mendel's Results
  1. His results were contrary to those predicted by a blending theory of inheritance.
  2. He found that the F<sub>1</sub> plants resembled only one of the parents.
  3. Characteristic of other parent reappeared in about 1/4 of F<sub>2</sub> plants; 3/4 of offspring resembled the F<sub>1</sub> plants.
  4. Mendel saw that these 3:1 results were possible if:
    - a. F<sub>1</sub> hybrids contained two factors for each trait, one *dominant* and one *recessive*;
    - b. factors separated when gametes were formed; a gamete carried one copy of each factor;
    - c. and random fusion of all possible gametes occurred upon fertilization.

5. Results of his experiments led Mendel to develop his first law of inheritance:
    - a. **Mendel's law of segregation:** Each organism contains two factors for each trait; factors segregate in formation of gametes; each gamete contains one factor for each trait; and fertilization gives each new individual two factors for each trait.
    - b. Mendel's law of segregation is consistent with a particulate theory of inheritance because many individual factors are passed on from generation to generation.
    - c. Reshuffling of factors explains variations and why offspring differ from their parents.
- C. As Viewed by Modern Genetics
1. Each trait in a pea plant is controlled by two **alleles**, alternate forms of a gene that occur at the same **gene locus** on homologous chromosomes.
    - a. A **dominant allele** masks or hides expression of a recessive allele; it is represented by an uppercase letter.
    - b. A **recessive allele** is an allele that exerts its effect only in the **homozygous** state; its expression is masked by a dominant allele; it is represented by a lowercase letter.
  2. **Gene locus** is specific location of a particular gene on homologous chromosomes.
  3. In Mendel's cross, the parents were true-breeding; each parent had two identical alleles for a trait—they were **homozygous**, indicating they possess two identical alleles for a trait.
    - a. **Homozygous dominant** genotypes possess two dominant alleles for a trait.
    - b. **Homozygous recessive** genotypes possess two recessive alleles for a trait.
  4. After cross-pollination, all individuals of the F<sub>1</sub> generation had one of each type of allele.
    - a. **Heterozygous** genotypes possess one of each allele for a particular trait.
    - b. The allele not expressed in a heterozygote is a recessive allele.
- D. Genotype Versus Phenotype
1. Two organisms with different allele combinations can have same outward appearance (e.g., *TT* and *Tt* pea plants are both tall; therefore, it is necessary to distinguish between alleles present and the appearance of organism).
  2. **Genotype** refers to the alleles an individual receives at fertilization.
  3. **Phenotype** refers to the physical appearance of the individual.
- E. One-trait Genetics Problems
1. First determine which characteristic is dominant; then code the alleles involved.
  2. Determine the genotype and gametes for both parents; an individual has two alleles for each trait; each gamete has only one allele for each trait.
  3. Each gamete has a 50% chance of receiving either allele.
- F. Laws of Probability
1. **Probability** is the likely outcome a given event will occur from random chance.
    - a. With each coin flip there is a 50% chance of heads and 50% chance of tails.
    - b. Chance of inheriting one of two alleles from a parent is also 50%.
  2. **Multiplicative law of probability** states that the chance of two or more independent events occurring together is the product of the probability of the events occurring separately.
    - a. Chance of inheriting a specific allele from one parent and a specific allele from another is  $\frac{1}{2} \times \frac{1}{2}$  or  $\frac{1}{4}$ .
    - b. Possible combinations for the alleles *Ee* of heterozygous parents are the following:  
 $EE = \frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$      $eE = \frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$      $Ee = \frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$      $ee = \frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$
  3. **Additive law of probability** calculates probability of an event that occurs in two or more independent ways; it is sum of individual probabilities of each way an event can occur; in the above example where unattached earlobes are dominant (*EE*, *Ee*, and *eE*), the chance for unattached earlobes is  $\frac{1}{4} + \frac{1}{4} + \frac{1}{4} = \frac{3}{4}$ .
- G. The Punnett Square
1. The Punnett square was introduced by R. C. Punnett and provides a simple method to calculate the probable results of a genetic cross.
  2. In a Punnett square, all possible types of sperm alleles are lined up vertically and all possible egg alleles are lined up horizontally; every possible combination is placed in squares.
  3. The larger the sample size examined, the more likely the outcome will reflect predicted ratios; a large number of offspring must be counted to observe the expected results; only in that way can all possible genetic types of sperm fertilize all possible types of eggs.
  4. We cannot testcross humans in order to count many offspring; therefore in humans, the phenotypic

- ratio is used to estimate the probability of any child having a particular characteristic.
5. Punnett square uses laws of probability; it does not dictate what the next child will inherit.
  6. "Chance has no memory": if two heterozygous parents have first child with attached earlobes (likely in 1/4th of children), a second child born still has 1/4 chance of having attached earlobes.

#### H. One-Trait Testcross

1. Mendel performed **test crosses** by crossing his F<sub>1</sub> plants with homozygous recessive plants.
2. Results indicated the recessive factor was present in the F<sub>1</sub> plants; they were heterozygous.
3. A monohybrid **testcross** is **used** between an individual with dominant phenotype and an individual with a recessive phenotype to see if the individual with dominant phenotype is homozygous or heterozygous.

### 11.3 Two-Trait Inheritance

#### A. Two-trait (Dihybrid) Crosses

1. This two-trait cross is between two parent organisms that are true-breeding for different forms of two traits; it produces offspring *heterozygous* for both traits.
2. Mendel observed that the F<sub>1</sub> individuals were dominant in both traits.

#### B. F<sub>1</sub> Plants Self-Pollinate

1. Mendel observed four phenotypes among F<sub>2</sub> offspring; he deduced second law of heredity.
2. **Mendel's law of independent assortment** states members of one pair of factors assort independently of members of another pair; all combinations of factors occur in gametes.

#### C. Two-trait Genetics Problems

1. Laws of probability indicate a 9:3:3:1 phenotypic ratio of F<sub>2</sub> offspring resulting in the following:
  - a. 9/16 of the offspring are dominant for both traits;
  - b. 3/16 of the offspring are dominant for one trait and recessive for the other trait;
  - c. 3/16 of the offspring are dominant and recessive opposite of the previous proportions; and
  - d. 1/16 of the offspring are recessive for both traits.
2. The Punnett Square for Two-trait Crosses
  - a. A larger Punnett square is used to calculate probable results of this cross.
  - b. A phenotypic ratio of 9:3:3:1 is expected when heterozygotes for two traits are crossed and simple dominance is present for both genes.
  - c. Independent assortment during meiosis explains these results.

#### D. Two-Trait Testcross

1. A **two-trait testcross** tests if individuals showing two dominant characteristics are homozygous for both or for one trait only, or is heterozygous for both.
2. If an organism heterozygous for two traits is crossed with another recessive for both traits, the expected phenotypic ratio is 1:1:1:1.
3. In dihybrid genetics problems, the individual has four alleles, two for each trait.

### 11.4 Human Genetic Disorders

#### A. Patterns of Inheritance

1. Genetic disorders are medical conditions caused by alleles inherited from parents.
2. An autosome is any chromosome other than a sex (X or Y) chromosome.
3. In a pedigree chart, males are designated by squares, females by circles; shaded circles and squares are affected individuals; line between square and circle represents a union; vertical line leads to offspring.
4. A **carrier** is a heterozygous individual who has no apparent abnormality but can pass on an allele for a recessively inherited genetic disorder.
5. Autosomal dominant and autosomal recessive alleles have different patterns of inheritance.
  - a. Characteristics of autosomal dominant disorders
    - 1) Affected children usually have an affected parent.
    - 2) Heterozygotes are affected. Two affected parents can produce unaffected child; two unaffected parents will not have affected children.
  - b. Characteristics of autosomal recessive disorders
    - 1) Most affected children have normal parents since heterozygotes have a normal phenotype.

- 2) Two affected parents always produce an affected child.
- 3) Close relatives who reproduce together are more likely to have affected children.

## B. Autosomal Recessive Disorders

### 1. Tay-Sachs Disease

- a. Usually occurs among Jewish people in the U.S. of central and eastern European descent.
- b. Symptoms are not initially apparent; infant's development begins to slow between four to eight months, neurological and psychomotor difficulties become apparent, child gradually becomes blind and helpless, develops seizures, eventually becomes paralyzed and dies by age of three or four.
- c. This results from lack of enzyme hexosaminidase A (Hex A) and the subsequent storage of its substrate, glycosphingolipid, in lysosomes.
- d. Primary sites of storage are cells of the brain; accounts for progressive deterioration.
- e. There is no treatment or cure.
- f. Prenatal diagnosis is possible by amniocentesis or chorionic villi sampling.

### 2. Cystic Fibrosis

- a. This is most common lethal genetic disease in Caucasians in U.S.
- b. About 1 in 20 Caucasians is a carrier, and about 1 in 3,000 newborns has this disorder.
- c. It increases the production of a viscous form of mucus in the lungs and pancreatic ducts.
  - 1) The resultant accumulation of mucus in the respiratory tract interferes with gas exchange.
  - 2) Digestive enzymes must be mixed with food to supplant the pancreatic juices.
- d. New treatments have raised the average life expectancy to up to 35 years.
- e. Chloride ions ( $\text{Cl}^-$ ) fail to pass plasma membrane proteins.
- f. Since water normally follows  $\text{Cl}^-$ , lack of water in the lungs causes thick mucus.
- g. The cause is a gene on chromosome 7; attempt to insert gene into nasal epithelium has had little success.
- h. Genetic testing for adult carriers and fetuses is possible.

### 3. Phenylketonuria (PKU)

- a. PKU occurs once in every 5,000 births; it is the most common inherited disease of nervous system.
- b. It is caused by a lack of an enzyme needed to metabolize amino acid phenylalanine; this results in accumulation of the amino acid in nerve cells of the brain and impairs nervous system development.
- c. PKU is caused by a gene on chromosome 12.
- d. Now newborns are routinely tested in hospital for high levels of phenylalanine in the blood.
- e. If an infant has PKU, the child is placed on diet low in phenylalanine until the brain is fully developed near age seven

## B. Autosomal Dominant Disorders

### 1. Neurofibromatosis

- a. This is an autosomal dominant disorder that affects one in 3,500 newborns and is distributed equally around the world.
- b. Affected individuals have tan skin spots at birth, which develop into benign tumors.
- c. Neurofibromas are lumps under the skin comprised of fibrous coverings of nerves.
- d. In most cases, symptoms are mild and patients live a normal life; sometimes symptoms are severe:
  - 1) skeletal deformities, including a large head;
  - 2) eye and ear tumors that can lead to blindness and hearing loss; and
  - 3) learning disabilities and hyperactivity.
  - 4) Such variation is called **variable expressivity**.
- e. The gene that codes for neurofibromatosis was discovered in 1990 to be on chromosome 17.
  - 1) The gene controls production of neurofibromin protein that normally blocks growth signals for cell division.
  - 2) Many types of mutations result in this effect.
  - 3) Some mutations are caused by a gene that moves from another location in the genome.

### 2. Huntington Disease

- a. This leads to progressive degeneration of brain cells, which in turn causes severe muscle spasm, personality disorders, and death in 10–15 years after onset.

- b. Most appear normal until they are of middle age and already have had children who might carry the gene; occasionally, first signs of the disease are seen in teenagers or even younger.
- c. The gene for Huntington disease is located on chromosome 4.
- e. This gene contains many repeats of a base triplet that codes for glutamine in the huntingtin protein; normal persons have 10–15 glutamines; affected persons have 36 or more.
- f. A huntingtin protein with over 36 glutamines changes shape and forms large clumps inside neurons; it also attracts other proteins to clump with it .

## 11.5 Beyond Mendelian Genetics

### A. Incomplete Dominance

- 1. **Incomplete dominance:** offspring show traits intermediate between two parental phenotypes.
  - a. True-breeding red and white-flowered four-o'clocks produce pink-flowered offspring.
  - b. Incomplete dominance has a biochemical basis; the level of gene-directed protein production may be between that of the two homozygotes.
  - c. One allele of a heterozygous pair only partially dominates expression of its partner.
  - d. This does not support a blending theory; parental phenotypes reappear in F<sub>2</sub> generation.

### B. Human Examples of Incomplete Dominance

- 1. Curly versus Straight Hair
  - a. A curly-haired Caucasian and a straight-haired Caucasian will have wavy-haired offspring.
  - b. Two wavy-haired parents will produce a 1:2:1 ratio of curly-wavy-straight hair children.
- 2. **Sickle-cell disease** is a blood disorder controlled by incompletely dominant alleles.
  - a. Codominance occurs when alleles are equally expressed in a heterozygote.
  - b.  $Hb^A Hb^A$  individuals are normal;  $Hb^S Hb^S$  have sickle-cell disease;  $Hb^A Hb^S$  have sickle-cell trait.
  - c. With sickle-cell disease, red blood cells are irregular in shape (sickle-shaped) rather than biconcave, due to abnormal hemoglobin that the cells contain.
  - d. Due to irregular shape, sickle-shaped red blood cells clog vessels and break down; results in poor circulation, anemia, low resistance to infection, hemorrhaging, damage to organs, jaundice, and pain of abdomen and joints; when a gene affects many traits, this is called **pleiotropy**.
  - e. Persons heterozygous for sickle-cell ( $Hb^A Hb^S$ ) usually lack sickle-cell symptoms unless deprived of water or oxygen.
  - f. In malaria regions of Africa, infants heterozygous ( $Hb^A Hb^S$ ) for sickle-cell allele have better chance of surviving; malaria parasite dies as potassium leaks from sickled cells.
  - g. Possible cures focus on bone marrow transplants and drugs that turn on genes for fetal hemoglobin in adults.

### C. Multiple Allelic Traits

- 1. This occurs when a gene has many allelic forms or alternative expressions.
- 2. ABO Blood Types
  - a. The ABO system of human blood types is a multiple allele system.
  - b. Two dominant alleles ( $I^A$  and  $I^B$ ) code for presence of A and B glycoproteins on red blood cells.
  - c. This also includes a recessive allele ( $i^O$ ) coding for no A or B glycoproteins on red blood cells.
  - d. As a result, there are four possible phenotypes (blood types): A, B, AB, and O
  - e. This is a case of **codominance**, where both alleles are fully expressed.
- 3. The Rh factor is inherited independently from the ABO system; the Rh<sup>+</sup> allele is dominant.

### D. Polygenic Inheritance

- 1. Polygenic inheritance occurs when one trait is governed by two or more sets of alleles.
- 2. Dominant alleles have a quantitative effect on the phenotype: each adds to the effect.
- 3. The more genes involved, the more continuous is the variation in phenotypes, resulting in a bell-shaped curve.
- 4. Crosses of white and dark-red wheat seeds produce seeds with seven degrees of intermediate colors due to genes at three separate loci.
- 5. Human Examples of Polygenic Inheritance
  - a. A hybrid cross for skin color provides a range of intermediates.
  - b. Parents with intermediate skin color can produce children with the full range of skin colors.
  - c. Albinism, where one gene interferes with the expression of others, is an example of **epistasis**.

### E. Polygenic Disorders

- 1. This includes cleft lip, clubfoot, congenital dislocations of the hip, hypertension, diabetes, schizophrenia, allergies and cancers.

2. Behavioral traits including suicide, phobias, alcoholism, and homosexuality may be associated with particular genes but are not likely completely predetermined.
3. Environment and the Phenotype
  - a. In water buttercups, the aquatic environment dramatically influences the structure of the plant.
  - b. Temperature triggers primrose to develop white flowers when grown above 32°C and red flowers when grown at 24°C.
  - c. The coats of Siamese cats and Himalayan rabbits have darker tipped ears, nose, paws, etc. due to the enzyme encoded by an allele is only active at low temperatures at the extremities.