Chapter 10

OBSERVING PATTERNS IN INHERITED TRAITS
Mendel, Pea Plants, and Inheritance Patterns

By experimenting with pea plants, **Mendel** was the first to gather evidence of patterns by which parents transmit genes to offspring.
a Garden pea flower, cut in half. Sperm form in pollen grains, which originate in male floral parts (stamens). Eggs develop, fertilization takes place, and seeds mature in female floral parts (carpels).

b Pollen from a plant that breeds true for purple flowers is brushed onto a floral bud of a plant that breeds true for white flowers. The white flower had its stamens snipped off. This is one way to assure cross-fertilization of plants.

c Later, seeds develop inside pods of the cross-fertilized plant. An embryo within each seed develops into a mature pea plant.

d Each new plant’s flower color is indirect but observable evidence that hereditary material has been transmitted from the parent plants.
Heritable Units of Information

• **Genes**
  – Heritable units of information about traits
  – Each has its own **locus** on the **chromosome**

• **Alleles**
  – Different molecular forms of the same gene

• **Mutation**
  – Permanent change in a gene’s information
a A **pair of homologous chromosomes**, both unduplicated. In most species, one is inherited from a female parent and its partner from a male parent.

b A **gene locus** (plural, loci), the location for a specific gene on a chromosome. ** Alleles ** are at corresponding loci on a pair of homologous chromosomes.

c A pair of alleles may be identical or not. Alleles are represented in the text by letters such as $D$ or $d$.

d Three **pairs of genes** (at three loci on this pair of homologous chromosomes); same thing as three pairs of alleles.
Modern Genetic Terms

Dominant allele may mask effect of recessive allele on the homologous chromosome

Genotype

• It is the genetic make up or constitution of an individual with reference to the traits

Phenotype

– An individual’s observable traits
– Visible expression of particular character
Modern Genetic Terms

• **Homozygous dominant**
  – Has two dominant alleles for a trait \((AA)\)

• **Homozygous recessive**
  – Has two recessive alleles \((aa)\)

• **Heterozygote**
  – Has two nonidentical alleles \((Aa)\)
Producing Hybrids

• **Hybrids**
  – Offspring of a cross between two individuals that breed true for different forms of a trait

• Each inherits nonidentical alleles for a trait being studied
homozygous dominant parent

chromosomes duplicated before meiosis

meiosis I

meiosis II

(gametes)

(fertilization produces heterozygous offspring)

gametes

homozygous recessive parent
Key Concepts:

MODERN GENETICS

• Gregor Mendel gathered the first indirect, experimental evidence of the genetic basis of inheritance

• His meticulous work tracking traits in many generations of pea plants gave him clues that heritable traits are specified in units

• The units, distributed into gametes in predictable patterns, were later identified as genes
Mendel’s Theory of Segregation

Mendel’s Theory of Segregation:

– *Diploid organisms have pairs of genes, on pairs of homologous chromosomes*

– Based on monohybrid experiments

• During meiosis

  – Genes of each pair separate
  – Each gamete gets one or the other gene

**Law of Segregation** – The separation of alleles into separate gametes
Producing Hybrid Offspring

• Crossing two true-breeding parents of different genotypes yields **hybrid** offspring

• All F₁ offspring are **heterozygous** for a gene, and can be used in monohybrid experiments
  – All F₁ offspring of parental cross **AA x aa** are **Aa**
Cross between two plants that breed true for different forms of a trait.
A Monohybrid Cross

- Crosses between F₁ monohybrids resulted in these allelic combinations among F₂ offspring
  - Phenotype ratio 3:1
  - Evidence of dominant and recessive traits

\[
\begin{array}{c|c|c}
 & A & a \\
\hline
A & AA & Aa \\
\hline
a & Aa & aa \\
\end{array}
\]

\{ AA \text{ (dominant)} \  
\text{Aa (dominant)} \  
\text{Aa (dominant)} \  
\text{aa (recessive)} \}

the expected phenotypic ratio of 3:1
From left to right, step-by-step construction of a Punnett square. Circles signify gametes. $A$ stands for a dominant allele and $a$ for a recessive allele at the same gene locus. Offspring genotypes are indicated inside the squares.
Predicting $F_1$ Offspring

True-breeding homozygous recessive parent plant

True-breeding homozygous dominant parent plant

Cross between two plants that breed true for different forms of a trait.
Predicting $F_2$ Offspring

Heterozygous $F_1$ offspring

Cross between heterozygous $F_1$ offspring.

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Key Concepts:

**MONOHYBRID EXPERIMENTS**

- Some experiments yielded evidence of gene segregation

- When one chromosome separates from its homologous partner during meiosis, the pairs of alleles on those chromosomes also separate and end up in different gametes
Mendel’s Theory of Independent Assortment

Mendel’s Theory of Independent Assortment:
The independent segregation of each pair of alleles during gamete formation

- Based on dihybrid experiments

- Pairs of homologous chromosomes align randomly at metaphase I
Independent Assortment in Meiosis I

One of two possible alignments

- Chromosome alignments at metaphase I:
  - a: $Aa$ and $Bb$
  - b: $Aa$ and $Bb$

The only other possible alignment

- The resulting alignments at metaphase II:
  - a: $Aa$ and $Bb$
  - b: $Aa$ and $Bb$

- Possible combinations of alleles in gametes:
  - c: $AB$, $ab$, $Ab$, $aB$
Meiosis, gamete formation in true-breeding parent plants

Parent homozygous dominant for purple flowers, tall stems

Parent homozygous recessive for white flowers, short stems

Gametes at fertilization

Possible genotypes resulting from a cross between two F₁ plants:

All F₁ plants are AaBb heterozygotes with purple flowers and tall stems.
Dihybrid Experiments

• Start with a cross between true-breeding heterozygous parents that differ for alleles of two genes (AABB x aabb)

• All F₁ offspring are heterozygous for both genes (AaBb)
Mendel’s Dihybrid Experiments

• $AaBb \times AaBb$

• Phenotypes of the $F_2$ offspring of $F_1$ hybrids were close to a 9:3:3:1 ratio
  – 9 dominant for both traits
  – 3 dominant for $A$, recessive for $b$
  – 3 dominant for $B$, recessive for $a$
  – 1 recessive for both traits
Key Concepts:

DIHYBRID EXPERIMENTS

• Some experiments yielded evidence of independent assortment

• During meiosis, the members of a pair of homologous chromosomes are distributed into gametes independently of all other pairs
Beyond Simple Dominance

- Other types of gene expression
  - Codominant alleles
  - Incomplete dominance
  - Epistasis
  - Pleiotropy
Codominant Alleles

- Both expressed at the same time in heterozygotes
- Nonidentical alleles for a gene are fully expressed
  - *Example*: Multiple alleles in ABO blood typing
Incomplete Dominance

• An allele is not fully dominant over its partner on a homologous chromosome
  – Both are expressed
  – Produces a phenotype between the two homozygous conditions
Cross two of the F1 plants, and the F2 offspring will show three phenotypes in a 1:2:1 ratio:

- **Homozygous parent (RR)**
- **Homozygous parent (rr)**
- **Heterozygous F1 offspring (Rr)**
Epistasis

- The masking of the phenotypic effect of alleles at one gene by alleles of another gene.
- A gene is said to be epistatic when its presence suppresses the effect of a gene at another locus.
Pleiotropy

• A single gene may affects two or more traits
  – Example: Marfan syndrome
  – Mutation in fibrillin gene
Linkage Groups

• All genes on the same chromosome are part of one linkage group

• Crossing over between homologous chromosomes disrupts gene linkages
Linkage Groups and Meiosis

• During meiosis, genes relatively close together on a chromosome tend to stay together
  – Few crossover events occur between them

• Genes that are relatively far apart tend to assort independently into gametes
  – Greater frequency of crossing over between them
Linkage and Crossing Over

Parental generation

F₁ offspring

Gametes

Most gametes have parental genotypes
A smaller number have recombinant genotypes

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Genes and Environment

- Environmental factors may affect gene expression in individuals
  - *Example*: Temperature and fur color
Complex Variations in Traits

- **Polygenic Inheritance**
  - When products of many genes influence a trait, individuals of a population show a range of continuous variation for the trait.
Variations in Gene Expression

• Gene interactions and environmental factors affect most phenotypes
  – Gene products control metabolic pathways
  – Mutations may alter or block pathways
VARIATIONS ON MENDEL’S THEME (cont.)

• Two or more gene pairs often influence the same trait, and some single genes influence many traits

• The environment also influences variation in gene expression