Chapter 14

Mendel and the Gene Idea

PowerPoint Lectures for
Biology, Seventh Edition
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Questions of heredity

- Babylonians & Ancient Egyptians ~ 6000 ya: agriculture, pedigrees, cross-pollination
- Pythagoras c. 500 BC: male parent dominant
- Empedocles c. 453 BC: blending
- Aristotle: semen was purified blood (this theory lasted 2000 years!
- Harvey & Leeuwenhoek: discovered eggs & fertilization: 17th & 18th centuries

Genetic inheritance

- What genetic principles account for the transmission of traits from parents to offspring?
  - Blending of traits (Darwin) – contradicted Darwin's own theory (eg bottle of ink)
  - "Particulate" hypothesis of inheritance: the gene idea
    - Parents pass on discrete heritable units, genes

- A Moravian monk who discovered the principals of particulate inheritance through carefully conducted plant breeding experiments

Gregor Mendel
(1822- 1884)

Mendel's Experimental, Quantitative Approach

- Mendel chose to work with peas
  - Because they are available in many varieties
  - Because he could strictly control which plants mated with which

How to cross pea plants

1. Remove stamens
2. Transfer pollen
3. Let carpal mature into pod
4. Plant seeds from pod
5. Examine offspring: all purple

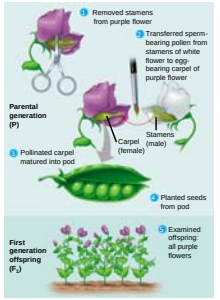
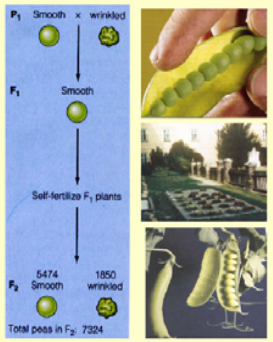


Figure 14.2

Mendel's breeding experiments

- Mendel created "pure breeding" strains for various traits.
- He noted that in 1st generation (F1) cross of these strains, certain (recessive) traits disappeared.
- However, in the F2 crosses they reappeared.
- This was strong evidence against blending inheritance



Vocabulary

- Character: a heritable feature, such as flower color
- Trait: a variant of a character, such as purple or white flowers
- Homozygous: an organism has identical alleles for a trait
- Heterozygous: an organism has different alleles for a trait
- Phenotype: physical traits
- Genotype: contributing alleles to traits

- Mendel discovered a ratio of about 3:1 purple to white flowers, in the F_2 generation

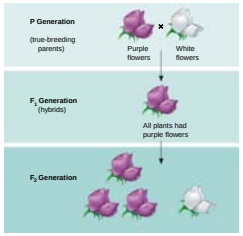


Figure 14.3

- Mendel reasoned that
  - In the F_1 plants, only the purple flower factor was affecting flower color in these hybrids
  - Purple flower color was dominant, and white flower color was recessive

Mendel's Model to explain 3:1 inheritance

1. There are alternative versions of genes: alleles
2. For each character an organism inherits two alleles, one from each parent
3. If the two alleles at a locus differ
   - Then one, the dominant allele, determines the organism's appearance, the other has no noticeable effect
4. The law of segregation
   - The two alleles separate (segregate) during gamete formation and end up in different gametes

Alternative alleles

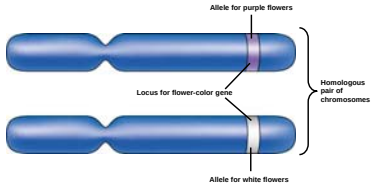


Figure 14.4

- Does Mendel's segregation model account for the 3:1 ratio he observed in the F_2 generation of his numerous crosses?
  - We can answer this question using a Punnett square

- Mendel's law of segregation, probability and the Punnett square

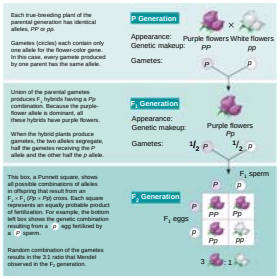


Figure 14.5

- Phenotype versus genotype

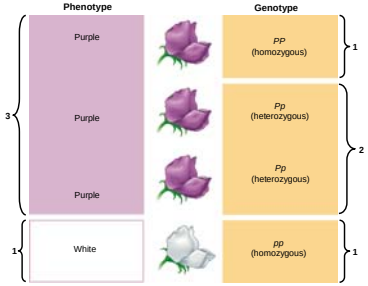


Figure 14.6

The Testcross

- In pea plants with purple flowers
  - The genotype is not immediately obvious
  - A testcross:
    - Allows us to determine the genotype of an organism with the dominant phenotype, but unknown genotype
    - P _ x pp

- The testcross

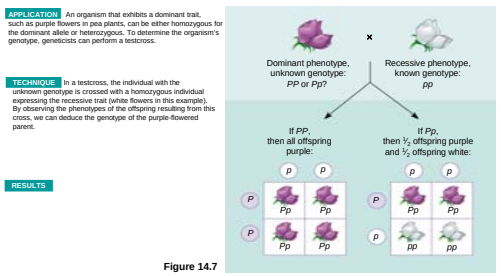


Figure 14.7

Law of independent assortment

- Are alleles segregated independently of each other?
- Crossing two, true-breeding parents differing in two characters
  - Produces dihybrids in the F_1 generation, heterozygous for both characters

- A dihybrid cross
  - Illustrates the inheritance of two characters
- Produces four phenotypes in the F_2 generation

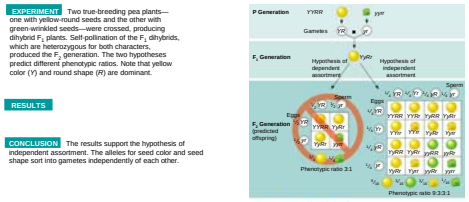
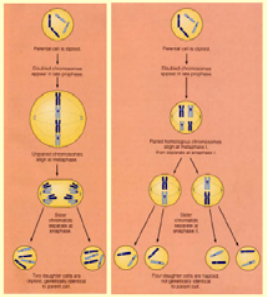


Figure 14.8 315 108 101 32 Phenotypic ratio approximately 9:3:3:1

Mitosis vs. Meiosis: the duplication of chromosomes during cell division explains the pattern of inheritance Mendel observed

- **Mitosis**
  - Cell division during normal growth and development.
  - Normally results in a diploid number of chromosomes (46 in humans).
  - Mutations are not heritable
- **Meiosis**
  - Cell division during the formation of sex cells (eggs and sperm).
  - Normally results in a haploid numbers of chromosomes (23 in humans).
  - Mutations are heritable



Probability

- Laws of segregation and independent assortment
  - Reflect the rules of probability
- Multiplication rule
  - the probability that two or more independent events will occur together is the product of their individual probabilities
- Example: toss one nickel twice, probability of heads once: .5 (what is probability of heads twice in a row – heads AND heads)
- .5 x .5 = .25

- Multiplication rule applies to probability in a monohybrid cross

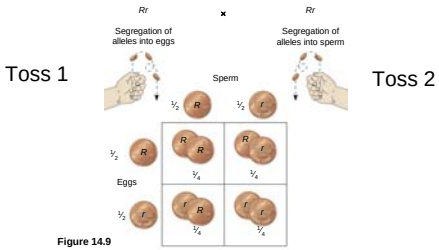


Figure 14.9

- Addition rule
  - The probability that any one of two or more mutually exclusive events will occur is calculated by adding together their individual probabilities
  - Toss a nickel, heads = .5, tails = .5
  - Probability of heads OR tails?
  - .5 plus .5 = 1.0

Solving Complex Genetics Problems with the Rules of Probability

- We can apply the rules of probability
  - To predict the outcome of crosses involving multiple characters

Predicting the outcome of multicharacter crosses

- A dihybrid or other multicharacter cross
  - Is equivalent to two or more independent monohybrid crosses occurring simultaneously
- In calculating the chances for various genotypes from such crosses
  - Each character first is considered separately and then the individual probabilities are multiplied together

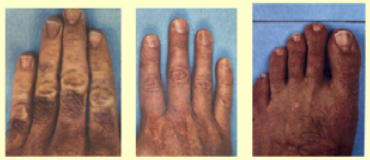
Non mendelian inheritance patterns

- Inheritance patterns are often more complex than predicted by simple Mendelian genetics

Single gene:

- Complete dominance
- Co-dominance
- Incomplete dominance

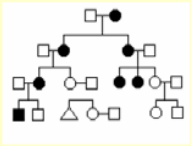
Some traits with dominant inheritance



Mid-digital hair Bent Little finger Sort Big Toe

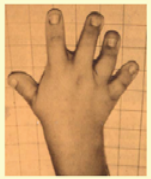
White Forelock

- A trait with Dominant inheritance
- Sometimes associated with defective hearing and other problems
- Notice that dominant traits such as this don't skip generations

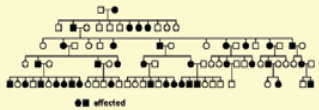


Brachydactyly

- Refers to shortening of the fingers and toes of the hands and feet.
- There are many different forms of this condition.
- Dominant inheritance



Dominant Inheritance of Pedigree of brachydactyly



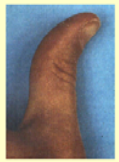
Achondroplastic Dwarfism

- An autosomal dominant trait
- If two achondroplastic people marry, about 2/3 of their children dwarf and 1/3 of normal height.
- There is also a higher than normal frequency of spontaneous abortions and stillbirths
- Homozygosity for the allele is probably incompatible with life

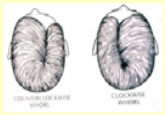
Porphyria

- **Dominant inheritance**
- A hemoglobin abnormality that afflicted King George III
- Part of the blood pigment hemoglobin, called a porphyrin ring, is not broken down and metabolized by cells.
- Porphyrin builds up and attacks the nervous system, causing mental problems, blood red urine, and other symptoms.


Some traits with recessive inheritance



Hitchhiker's thumb

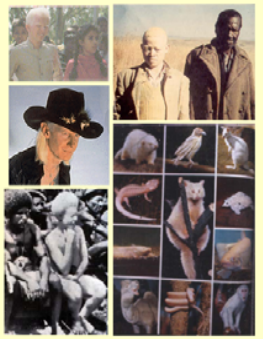


Counter clockwise whorl

Albinism

- The most common form is an **autosomal recessive** disorder
- Afflicted individuals lack the enzyme (tyrosinase) necessary for producing the pigment melanin.
- characterized by absence of pigment in hair, skin, and eyes
- Eye problems and sun sensitivity are common features.
- The "albino locus" can be filled with several different alleles.

They all affect the production of melanin.



Recessive inheritance of albinism

- Notice generations are sometimes skipped in the expression of traits with recessive inheritance
- If two people with albinism have children, what will the phenotype of their children be?

	A	a
A	AA	Aa
a	Aa	aa

Incomplete Dominance

For some traits the heterozygote has a phenotype intermediate between those of the homozygous parents

Parental Generation

F1

F2

	A	a
A	AA	Aa
a	Aa	aa

- Dominant & recessive alleles don't interact, rather they produce different proteins
- Dominant alleles
  - Are not necessarily more common in populations than recessive alleles
- Most genes exist in populations in more than two allelic forms…

Multiple alleles

- The ABO blood group in humans
  - Is determined by multiple alleles

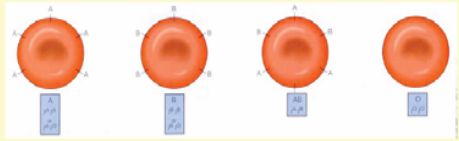
Table 14.2 Determination of ABO Blood Group by Multiple Alleles

Genotype	Phenotype (Blood Group)	Red Blood Cells
I^AI^A or I^Ai	A	
I^BI^B or I^Bi	B	
I^AI^B	AB	
ii	O	

Table 14.2

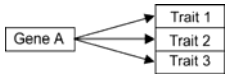
Codominance: the ABO blood group

- Occurs when each allele produces a unique product (ABO blood group).
- Each allele of codominant genes is thus fully expressed in the heterozygote
- The ABO blood group as an example
  - Type A blood has A antigens and B antibodies
  - Type B blood has B antigens and A antibodies
  - Type AB blood has both A and B antigens and no A or B antibodies
  - Type O blood has neither A or B antigens an both A an B antibodies in the plasma



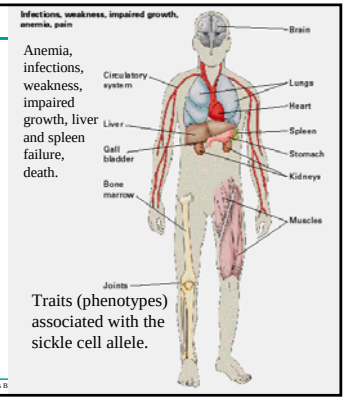
Pleiotropy

- A gene has multiple phenotypic effects



Pleiotropy:

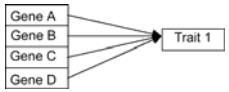
The sickle cell allele



Traits (phenotypes) associated with the sickle cell allele.

Extending Mendelian Genetics for Two or More Genes

- Some traits
  - May be determined by two or more genes



Epistasis

- In epistasis
  - A gene at one locus alters the phenotypic expression of a gene at a second locus

- An example of epistasis

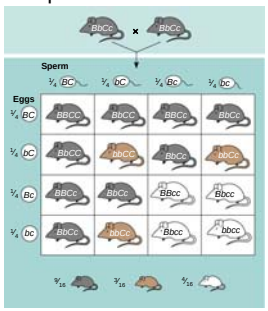


Figure 14.11

Polygenic Inheritance

- Many human characters
  - Vary in the population along a continuum and are called quantitative characters
  - Human skin, hair and eye colour

- Quantitative variation usually indicates polygenic inheritance
  - An additive effect of two or more genes on a single phenotype

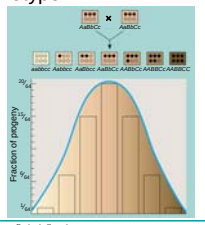


Figure 14.12

Polygenic Inheritance: Eye Color

- Eye color is likely to be a polygenic trait. The early view that blue is a simple recessive has been repeatedly shown to be wrong by observation of brown-eyed offspring of 2 blue-eyed parents.
- Blue-eyed offspring from 2 brown-eyed parents is a more frequent finding.
- At least two separate genes, each with two incompletely dominant alleles, govern human eye color.
- A man and a woman, each heterozygous for both genes, could have children with five different eye colors, ranging from light blue (no dominant alleles) through light brown (two dominants) to almost black (all four alleles dominant)."

Nature and Nurture: The Environmental Impact on Phenotype

- Another departure from simple Mendelian genetics arises
  - When the phenotype for a character depends on environment as well as on genotype
- Multifactorial characters
  - Are those that are influenced by both genetic and environmental factors

- The norm of reaction is the phenotypic range of a particular genotype that is influenced by the environment

Figure 14.13

Integrating a Mendelian View of Heredity and Variation

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

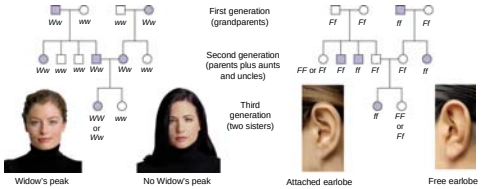
- Even in more complex inheritance patterns
  - Mendel's fundamental laws of segregation and independent assortment still apply

Pedigree Analysis

- A pedigree
  - Is a family tree that describes the interrelationships of parents and children across generations

Pedigrees – family tree of traits (analysis)

- Pedigrees can be used to predict the traits of future offspring (genetic disorders)



(a) Dominant trait (widow's peak) (b) Recessive trait (attached earlobe)

Figure 14.14 A, B

Genetic disorders

- May be recessive or dominant
- Recessively inherited disorders
  - Show up only in individuals homozygous for the allele
- Carriers
  - Are heterozygous individuals who carry the recessive allele but are phenotypically normal

Mating of Close Relatives

- Matings between relatives
  - Can increase the probability of the appearance of a genetic disease
  - Are called consanguineous matings

Huntington's disease

- Is a degenerative disease of the nervous system
- Has no obvious phenotypic effects until about 35 to 40 years of age

Figure 14.16

Multifactorial Disorders

- Many human diseases
  - Have both genetic and environment components
- Examples include
  - Heart disease and cancer

Genetic Testing and Counseling

- Genetic counselors
  - Can provide information to prospective parents concerned about a family history for a specific disease

Counseling Based on Mendelian Genetics and Probability Rules

- Using family histories
  - Genetic counselors help couples determine the odds that their children will have genetic disorders

Fetal Testing

- In amniocentesis
  - The liquid that bathes the fetus is removed and tested
- In chorionic villus sampling (CVS)
  - A sample of the placenta is removed and tested
- For a growing number of diseases
  - Tests are available that identify carriers and help define the odds more accurately

- Fetal testing

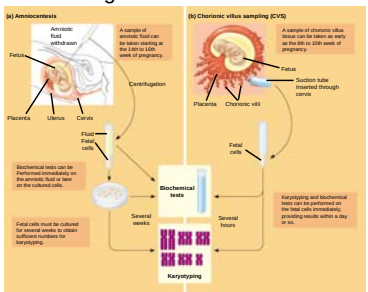


Figure 14.17 A, B