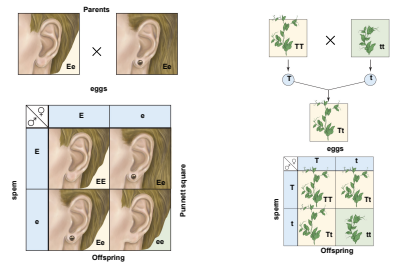
Chapter 11: pp. 189 - 210

Mendelian Patterns of Inheritance

BIOLOGY
10th Edition

Sylvia S. Mader



Outline

- Blending Inheritance
- Monohybrid Cross
  - Law of Segregation
- Modern Genetics
  - Genotype vs. Phenotype
  - Punnett Square
- Dihybrid Cross
  - Law of Independent Assortment
- Human Genetic Disorders

Gregor Mendel

- Austrian monk
  - Studied science and mathematics at University of Vienna
  - Conducted breeding experiments with the garden pea *Pisum sativum*
  - Carefully gathered and documented mathematical data from his experiments
- Formulated fundamental laws of heredity in early 1860s
  - Had no knowledge of cells or chromosomes
  - Did not have a microscope

Gregor Mendel

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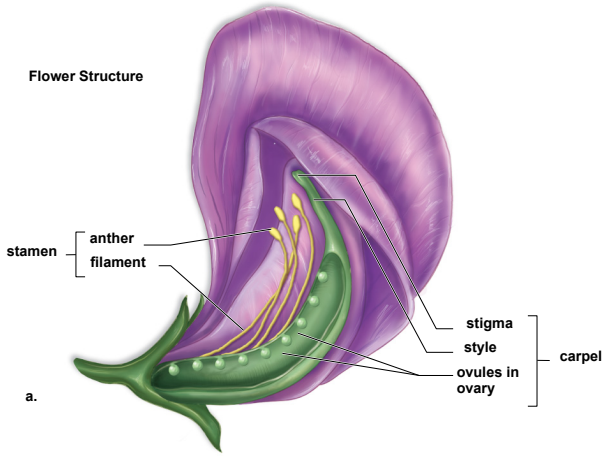
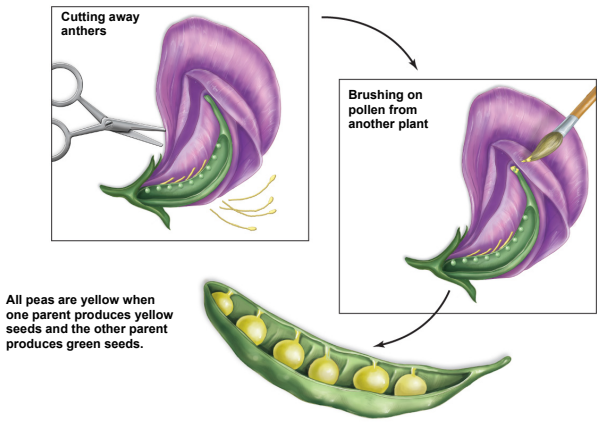
Fruit and Flower of the Garden Pea



Garden Pea Traits Studied by Mendel



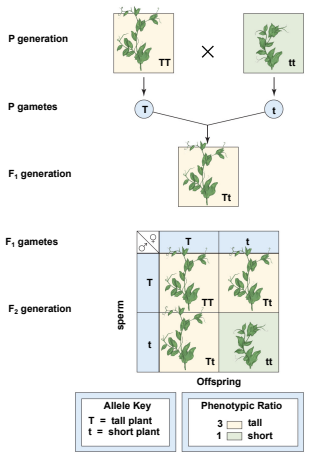
Blending Inheritance

- Theories of inheritance in Mendel's time:
  - Based on blending
  - Parents of contrasting appearance produce offspring of intermediate appearance
- Mendel's findings were in contrast with this
  - He formulated the particulate theory of inheritance
  - Inheritance involves reshuffling of genes from generation to generation

One-Trait Inheritance

- Mendel performed cross-breeding experiments
  - Used "true-breeding" (homozygous) plants
  - Chose varieties that differed in only one trait (monohybrid cross)
  - Performed reciprocal crosses
    - Parental generation = P
    - First filial generation offspring = F_1
    - Second filial generation offspring = F_2
  - Formulated the Law of Segregation

Mendel's Monohybrid Crosses: An Example



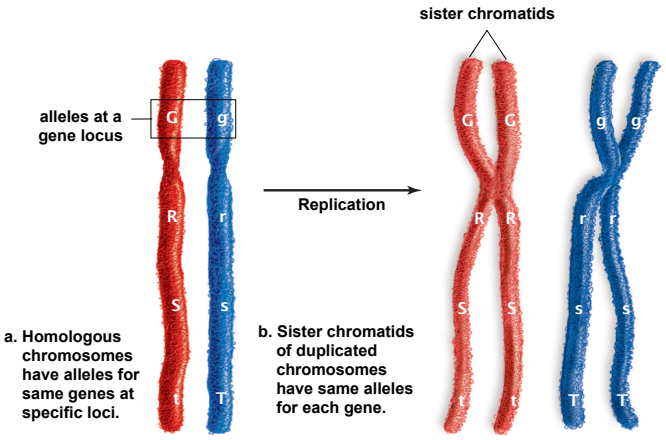
Law of Segregation

- Each individual has a pair of factors (alleles) for each trait
- The factors (alleles) segregate (separate) during gamete (sperm & egg) formation
- Each gamete contains only one factor (allele) from each pair
- Fertilization gives the offspring two factors for each trait

Modern Genetics View

- Each trait in a pea plant is controlled by two alleles (alternate forms of a gene)
- Dominant allele (capital letter) masks the expression of the recessive allele (lower-case)
- Alleles occur on a homologous pair of chromosomes at a particular gene locus
  - Homozygous = identical alleles
  - Heterozygous = different alleles

Homologous Chromosomes



Genotype versus Phenotype

- Genotype
  - Refers to the two alleles an individual has for a specific trait
  - If identical, genotype is homozygous
  - If different, genotype is heterozygous
- Phenotype
  - Refers to the physical appearance of the individual

Genotype versus Phenotype

TABLE 11.1

Genotype Versus Phenotype

Genotype	*Genotype*	*Phenotype*
TT	Homozygous dominant	Tall plant
Tt	Heterozygous	Tall plant
tt	Homozygous recessive	Short plant

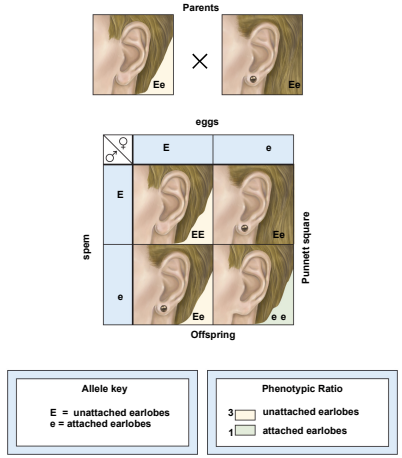
Punnett Square

- Table listing all possible genotypes resulting from a cross
  - All possible sperm genotypes are lined up on one side
  - All possible egg genotypes are lined up on the other side
  - Every possible zygote genotypes are placed within the squares

Punnett Square

- Allows us to easily calculate probability, of genotypes and phenotypes among the offspring
- Punnett square in next slide shows a 50% (or ½) chance
  - The chance of *E* = ½
  - The chance of *e* = ½
- An offspring will inherit:
  - The chance of *EE* =½!½=¼
  - The chance of *Ee* =½!½=¼
  - The chance of *eE* =½!½=¼
  - The chance of *ee* =½!½=¼

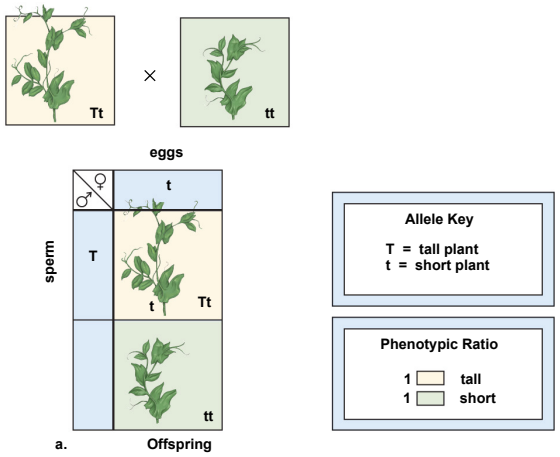
Punnett Square Showing Earlobe Inheritance Patterns



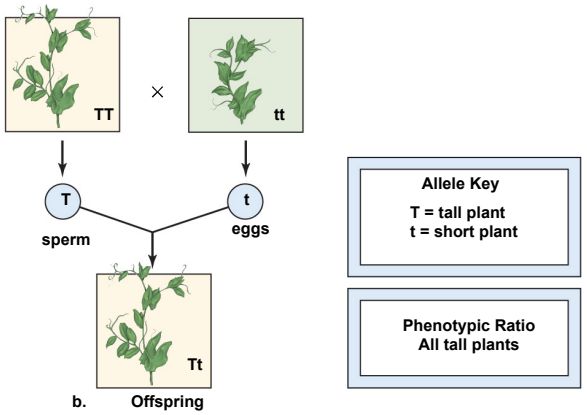
Monohybrid Test cross

- Individuals with recessive phenotype always have the homozygous recessive genotype
- However, individuals with dominant phenotype have indeterminate genotype
  - May be homozygous dominant, or
  - Heterozygous
- Test cross determines genotype of individual having dominant phenotype

One-Trait Test Cross



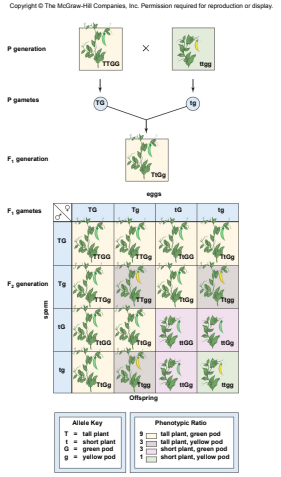
One-Trait Test Cross



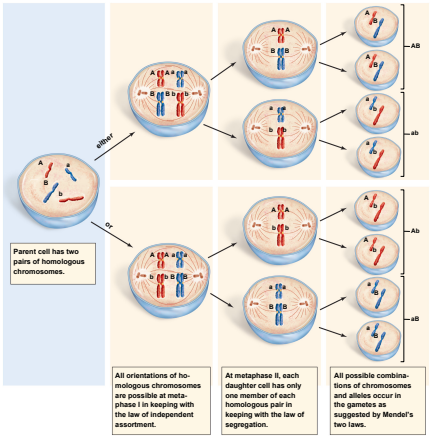
Two-Trait Inheritance

- Dihybrid cross uses true-breeding plants differing in two traits
  - Observed phenotypes among F_2 plants
  - Formulated Law of Independent Assortment
    - The pair of factors for one trait segregate independently of the factors for other traits
    - All possible combinations of factors can occur in the gametes
    - Mendel tracked each trait through two generations.
  - **P generation** is the parental generation in a breeding experiment.
  - **F1 generation** is the first-generation offspring in a breeding experiment.
  - **F2 generation** is the second-generation offspring in a breeding experiment

Two-Trait (Dihybrid) Cross



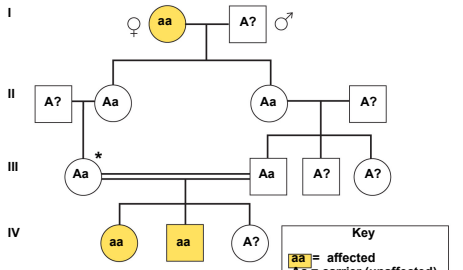
Independent Assortment and Segregation during Meiosis



Human Genetic Disorders

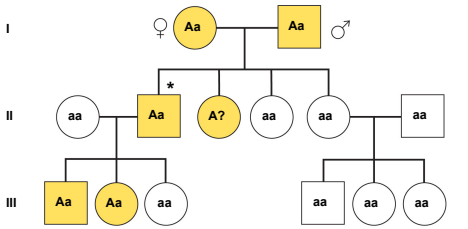
- Genetic disorders are medical conditions caused by alleles inherited from parents
- Autosome - Any chromosome other than a sex chromosome (X or Y)
- Genetic disorders caused by genes on autosomes are called autosomal disorders
  - Some genetic disorders are autosomal dominant
    - An individual with AA has the disorder
    - An individual with Aa has the disorder
    - An individual with aa does NOT have disorder
  - Other genetic disorders are autosomal recessive
    - An individual with AA does NOT have disorder
    - An individual with Aa does NOT have disorder, but is a carrier
    - An individual with aa DOES have the disorder

Autosomal Recessive Pedigree Chart



Autosomal recessive disorders
- Most affected children have unaffected parents.
- Heterozygotes (Aa) have an unaffected phenotype.
- Two affected parents will always have affected children.
- Close relatives who reproduce are more likely to have affected children.
- Both males and females are affected with equal frequency.

Autosomal Dominant Pedigree Chart

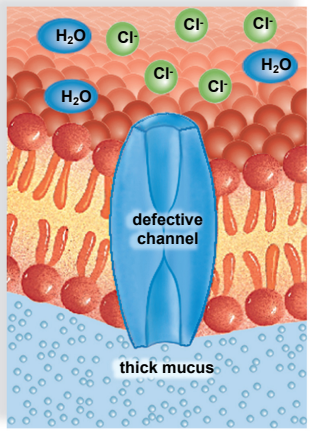


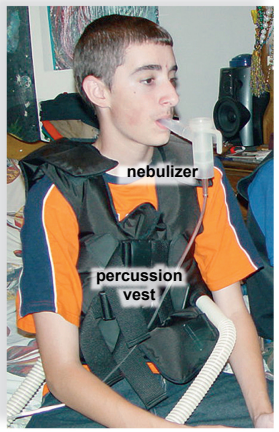
Autosomal dominant disorders
- affected children will usually have an affected parent.
- Heterozygotes (Aa) are affected. Two affected parents can produce an unaffected child.
- Two unaffected parents will not have affected children.
- Both males and females are affected with equal frequency.

Autosomal Recessive Disorders

- Tay-Sachs Disease
  - Progressive deterioration of psychomotor functions
- Cystic Fibrosis
  - Mucus in bronchial tubes and pancreatic ducts is particularly thick and viscous
- Phenylketonuria (PKU)
  - Lack enzyme for normal metabolism of phenylalanine

Cystic Fibrosis





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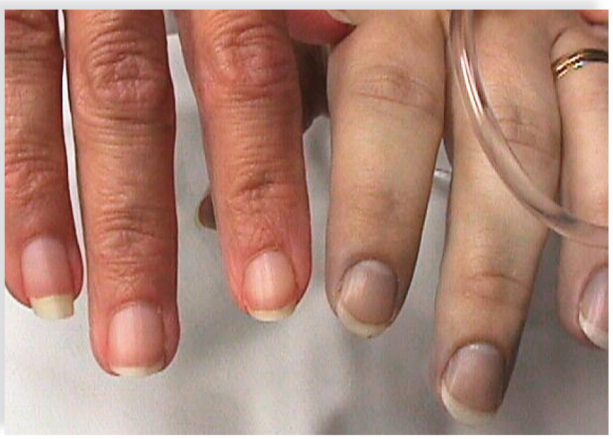
Methemoglobinemia



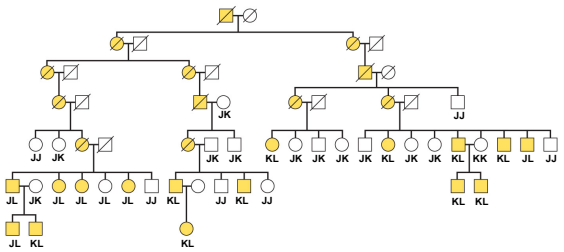
Courtesy Division of Medical Toxicology, University of Virginia

Autosomal Dominant Disorders

- Neurofibromatosis
  - Tan or dark spots develop on skin and darken
  - Small, benign tumors may arise from fibrous nerve coverings
- Huntington Disease
  - Neurological disorder
  - Progressive degeneration of brain cells
    - Severe muscle spasms
    - Personality disorders

A Victim of Huntington Disease

a.



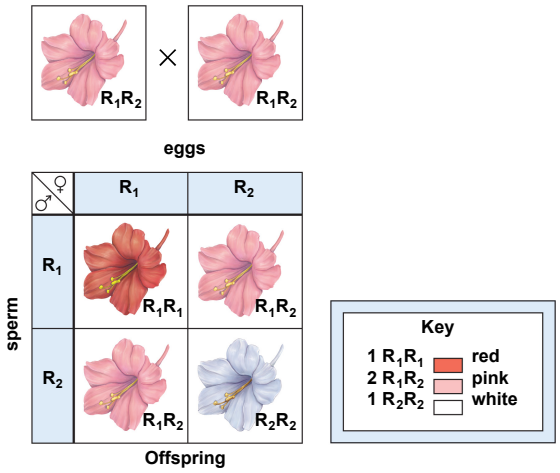
b.

a: © Steve Uzzell

Incomplete Dominance

- Heterozygote has phenotype intermediate between that of either homozygote
  - Homozygous red has red phenotype
  - Homozygous white has white phenotype
  - Heterozygote has pink (intermediate) phenotype
- Phenotype reveals genotype without test cross

Incomplete Dominance



Multiple Allelic Traits

- Some traits controlled by multiple alleles
- The gene exists in several allelic forms (but each individual only has two)
- ABO blood types
- The alleles:
  - I^A = A antigen on red cells, anti-B antibody in plasma
  - I^B = B antigen on red cells, anti-AB antibody in plasma
  - I = Neither A nor B antigens, both antibodies

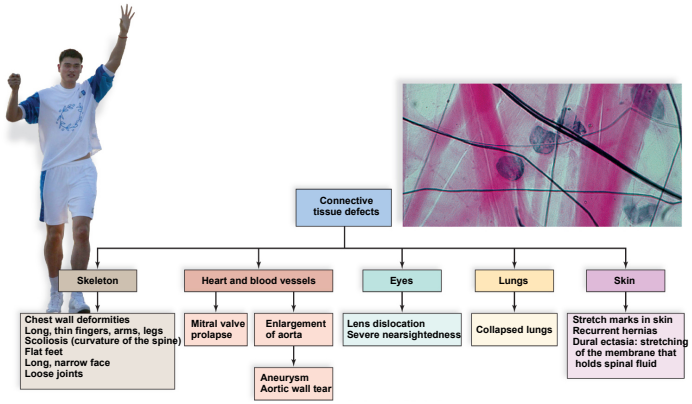
Multiple Allelic Traits

Phenotype	*Genotype*
A	I^AI^A, I^Ai
B	I^BI^B, I^Bi
AB	I^AI^B
O	ii

Pleioptropic Effects

- **Pleiotropy** occurs when a single mutant gene affects two or more distinct and seemingly unrelated traits.
  - Marfan syndrome have disproportionately long arms, legs, hands, and feet; a weakened aorta; poor eyesight

Marfan Syndrome

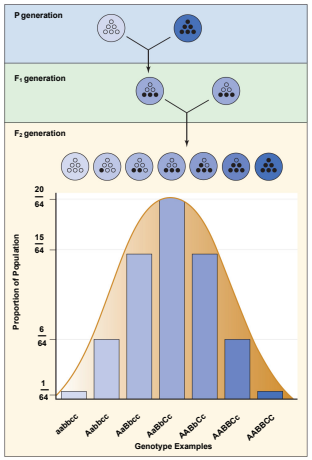


(Left): © AP/Wide World Photos; (Right): © Ed Reschke

Polygenic Inheritance

- Occurs when a trait is governed by two or more genes having different alleles
- Each dominant allele has a quantitative effect on the phenotype
- These effects are additive
- Result in continuous variation of phenotypes

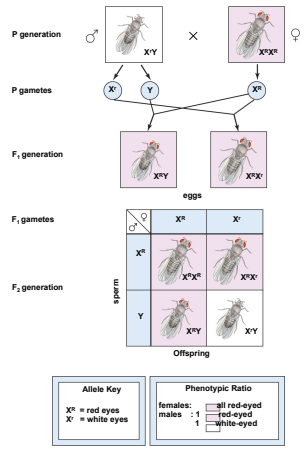
Frequency Distributions in Polygenic Inheritance



X – Linked Inheritance

- In mammals
  - The X and Y chromosomes determine gender
  - Females are XX
  - Males are XY
- The term **X-linked** is used for genes that have nothing to do with gender
  - Carried on the X chromosome.
  - The Y chromosome does not carry these genes
  - Discovered in the early 1900s by a group at Columbia University, headed by Thomas Hunt Morgan.
    - Performed experiments with fruit flies
      - They can be easily and inexpensively raised in simple laboratory glassware
      - Fruit flies have the same sex chromosome pattern as humans

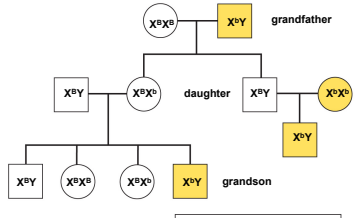
X – Linked Inheritance



Human X-Linked Disorders

- Several X-linked recessive disorders occur in humans:
  - Color blindness
    - The allele for the blue-sensitive protein is autosomal
    - The alleles for the red- and green-sensitive pigments are on the X chromosome.
  - Menkes syndrome
    - Caused by a defective allele on the X chromosome
    - Disrupts movement of the metal copper in and out of cells.
  - Muscular dystrophy
    - Wasting away of the muscle
  - Adrenoleukodystrophy
    - X-linked recessive disorder
    - Failure of a carrier protein to move either an enzyme or very long chain fatty acid into peroxisomes.
  - Hemophilia
    - Absence or minimal presence of a clotting factor VIII, or clotting factor IX
    - Affected person's blood either does not clot or clots very slowly.

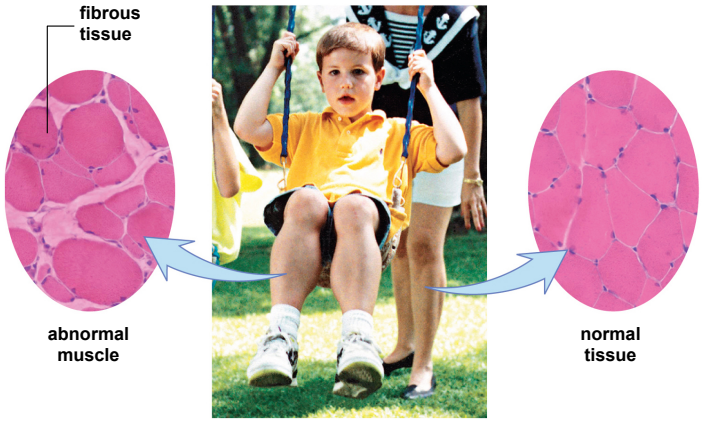
X-Linked Recessive Pedigree



X-Linked Recessive Disorders

- More males than females are affected.
- An affected son can have parents who have the normal phenotype.
- For a female to have the characteristic, her father must also have it. Her mother must have it or be a carrier.
- The characteristic often skips a generation from the grandfather to the grandson.
- If a woman has the characteristic, all of her sons will have it.

Muscle Dystrophy



(Abnormal): Courtesy Dr. Rabi Tawil, Director, Neuromuscular Pathology Laboratory, University of Rochester Medical Center; (Boy): Courtesy Muscular Dystrophy Association; (Normal): Courtesy Dr. Rabi Tawil, Director, Neuromuscular Pathology Laboratory, University of Rochester Medical Center.

Terminology

- Pleiotropy
  - A gene that affects more than one characteristic of an individual
  - Sickle-cell (incomplete dominance)
- Codominance
  - More than one allele is fully expressed
  - ABO blood type (multiple allelic traits)
- Epistasis
  - A gene at one locus interferes with the expression of a gene at a different locus
  - Human skin color (polygenic inheritance)

Review

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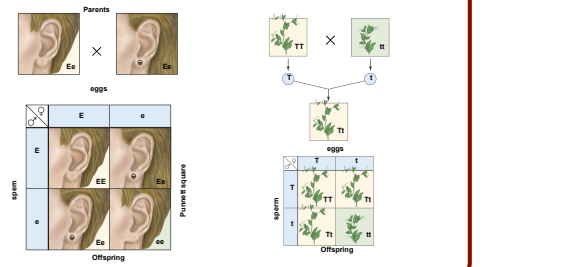

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