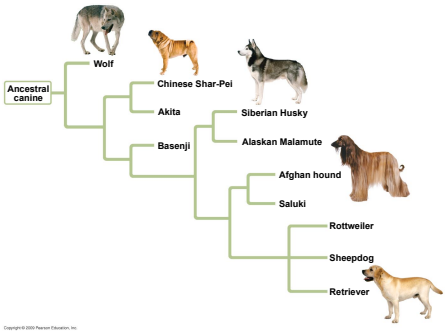
Genetics

Honors Biology 2012

1

Dog Breeds

- Result of artificial selection
  - Humans chose traits they found favorable for breeding



2

Old, Old School Ideas about Genetics

- Pangenesis
  - Particles called pangenes came from all parts of an organism to make sperm and eggs
  - Characteristics acquired in an organisms lifetime could be passed on to future generations
- Blending
  - Genetic material from both parents mixes to form a mixture of traits (like mixing paint)
- We now know both of these ideas are incorrect

3

Gregor Mendel

- Austrian Monk who worked with pea plants in the 1860s
- Proposed the idea that particles of heritable information (what we call genes) are passed on from generation to generation
- Chose to work with pea plants because he could observe many **characters** (features - ex. flower color) with several options of **traits** (purple, white, etc.)
- He could also strictly control mating
  - Self-fertilization
  - Cross-fertilization

4

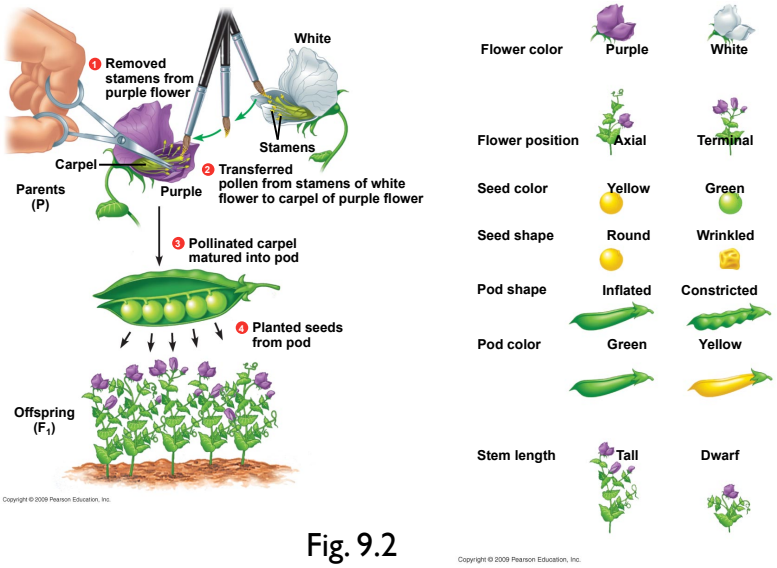


Fig. 9.2

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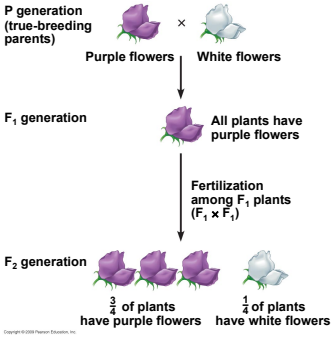
Law of Segregation

- Monohybrid Crosses (examines one character)



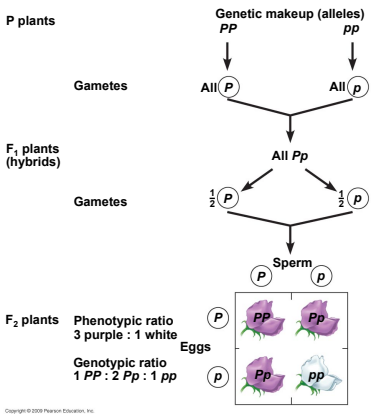


Fig. 9.3

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Four Hypotheses about the Law of Segregation

- Genes are found in alternative versions called **alleles**; the listing of alleles gives the **genotype** of an organism
- For each characteristic, an organism inherits two alleles (one from each parent) that can be the same of different
  - Homozygous - identical alleles
  - Heterozygous - different alleles
- If the alleles are different, the dominant allele determines the organism's **phenotype**
- Law of segregation - allele pairs separate from each other during the production of gametes so that sperm and egg has only one allele for each gene

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Homologous Chromosomes

- Alleles of a gene reside at the same locus on homologous chromosomes

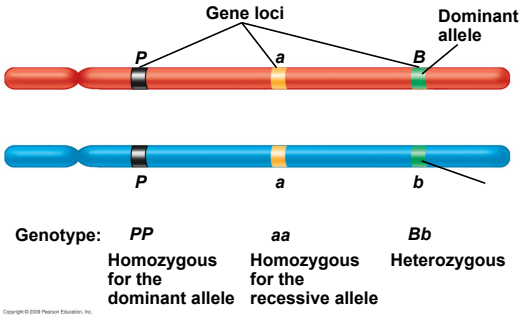

Fig. 9.4

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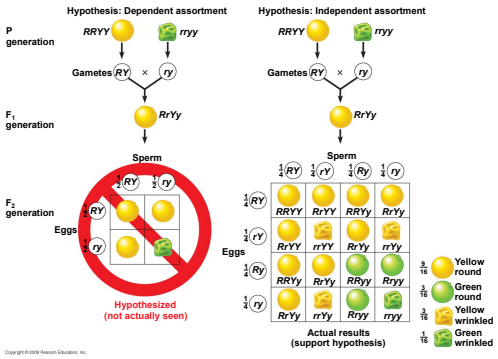
Law of Independent Assortment

- Seen in a dihybrid cross
- Mendel needed to explain: why combinations not found in the parents were observed and why a 9:3:3:1 ratio was observed



Law of Independent Assortment - each pair of alleles segregates independently of the other pair during gamete formation (this means that all combinations are possible)

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Test Crosses

- Help to determine the genotype of an individual
- Involves crossing the unknown individual with a homozygous recessive individual

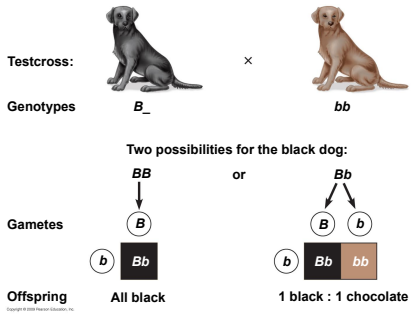
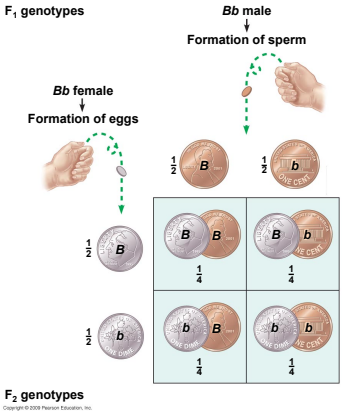


Fig. 9.6

Probability Rules and Genetics

- Rule of Addition - add the probabilities of events that can happen in multiple ways (OR)
- Rule of Multiplication - multiply the probabilities of events that must occur together (BOTH)



Pedigree

- Shows inheritance of a trait through multiple generations
- Shows dominant and recessive inheritance
- Can be used to determine the genotypes of family members

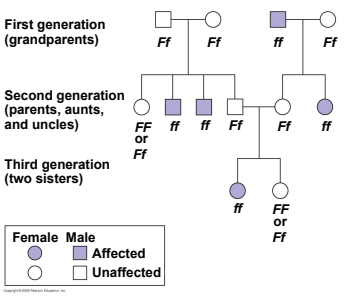


Fig. 9.8

Dominance

- Recessive inheritance
  - Require two recessive alleles to show disease
  - Heterozygous parents are carriers
- Dominant inheritance
  - Require one dominant allele to show disease

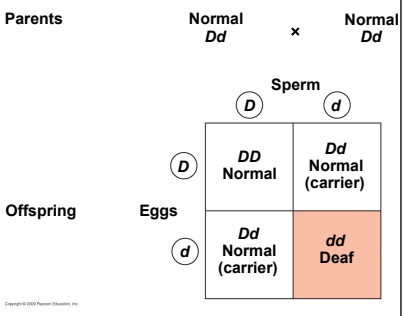
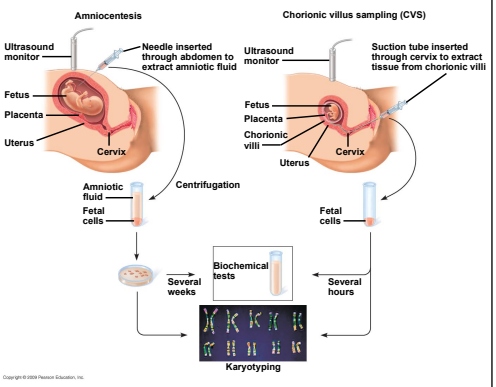


TABLE 9.9 SOME AUTOSOMAL DISORDERS IN HUMANS

Disorder	Major Symptoms	Incidence	Comments
Recessive disorders			
Albinism	Lack of pigment in skin, hair, and eyes	$\frac{1}{22,000}$	Prone to skin cancer
Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated	$\frac{1}{2,500}$ Caucasians	See Module 9.9
Galactosemia	Accumulation of galactose in tissues; mental retardation; eye and liver damage	$\frac{1}{100,000}$	Treated by eliminating galactose from diet
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation	$\frac{1}{10,000}$ in U.S. and Europe	See Module 9.10
Sickle-cell disease	Sickled red blood cells; damage to many tissues	$\frac{1}{400}$ African-Americans	See Module 9.13
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood	$\frac{1}{3,500}$ Jews from central Europe	See Module 4.11
Dominant disorders			
Achondroplasia	Dwarfism	$\frac{1}{25,000}$	See Module 9.9
Alzheimer's disease (one type)	Mental deterioration; usually strikes late in life	Not known	
Huntington's disease	Mental deterioration and uncontrollable movements; strikes in middle age	$\frac{1}{25,000}$	See Module 9.9
Hypercholesterolemia	Excess cholesterol in blood; heart disease	$\frac{1}{500}$ are heterozygous	See Module 9.11

Genetic Testing

- Testing of parents
- Fetal Testing: biochemical and kareotyping
  - Amniocentesis
  - Chorionic villus sampling (CVS)
- Newborn screening



Incomplete Dominance

- Neither allele is dominant over the other
- Expression of both alleles (heterozygous) observed as an intermediate phenotype

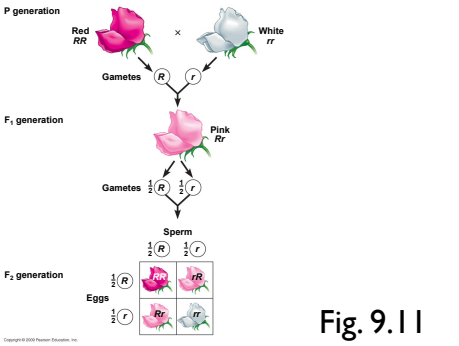
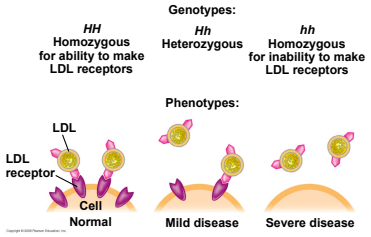


Fig. 9.11



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Multiple Alleles

- More than two alleles found in the population
- Diploid individual can carry any two of the alleles
- Most common example is ABO blood type
- Codominance - neither allele is dominant over the other (see both phenotypes)

Blood Group (Phenotype)	Genotypes	Red Blood Cells	Antibodies Present in Blood	Reaction When Blood from Groups Below Is Mixed with Antibodies from Groups at Left			
				O	A	B	AB
O	ii		Anti-A Anti-B				
A	I^AI^A or I^Ai	Carbohydrate A	Anti-B				
B	I^BI^B or I^Bi	Carbohydrate B	Anti-A				
AB	I^AI^B		—				

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Pleiotropy

- One gene influencing many factors
- Ex. Sickle-Cell gene

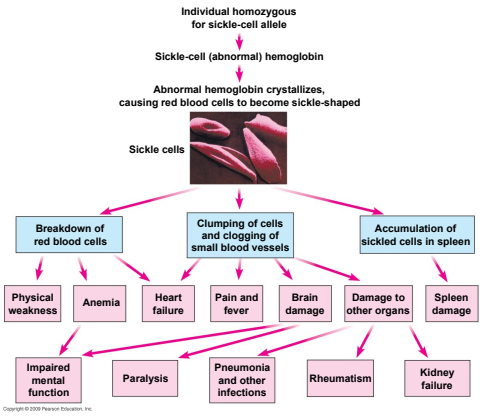

Fig. 9.13

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Polygenic Inheritance

- Many genes influence one trait
- Ex. Skin color

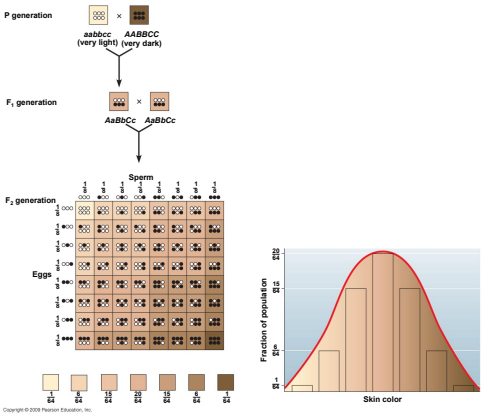

Fig. 9.14

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Phenotype can be Influenced by the Environment

- Skin color can be impacted by sunlight
- Susceptibility to diseases (like cancer) has hereditary and environmental components

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Chromosomal Inheritance

- Mendel's Laws correlate with chromosome separation in meiosis
  - Law of Segregation depends on separation of homologous chromosomes in anaphase I
  - Law of Independent Assortment depends on alternative orientations of chomosomes in metaphase I

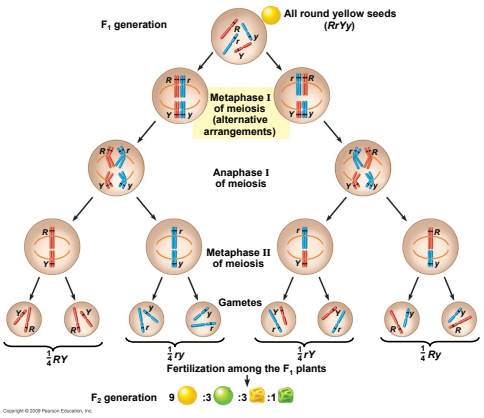

Fig. 9.16

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Linked Genes

- Genes on the same chromosome tend to be inherited together
- Example: Plants with purple flowers and long pollen was crossed with red flowers and pollen
- F_2 generation did not show 9:3:3:1 ratio
  - Most F_2 had purple flowers and long pollen or red flowers and round pollen
- Linked alleles can be separated by crossing over

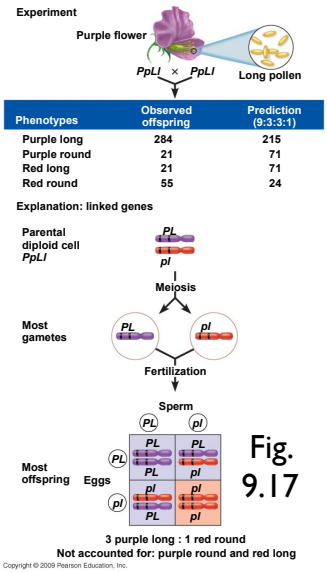

Fig. 9.17

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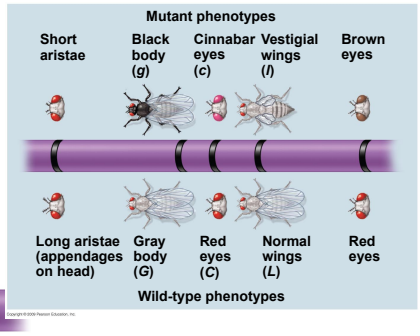
Genetic Maps

- Show the order of genes on a chromosome



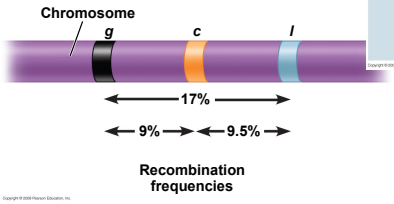


Fig. 9.19B

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Chromosomes Determine Sex in Most Species

- X-Y system in mammals and fruit flies
- X-O system in grasshoppers and roaches
- Z-W system in birds, butterflies, and some fish
- In ants and bees
  - Diploid = female; Haploid = male

Fig. 9.20

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Sex-linked Genes

- X-linked genes are passed from mother to son and mother to daughter
- X-linked genes are passed from father to daughter
- Y-linked genes are passed from father to son

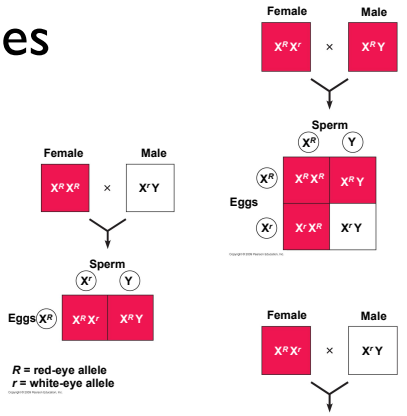

Fig. 9.21

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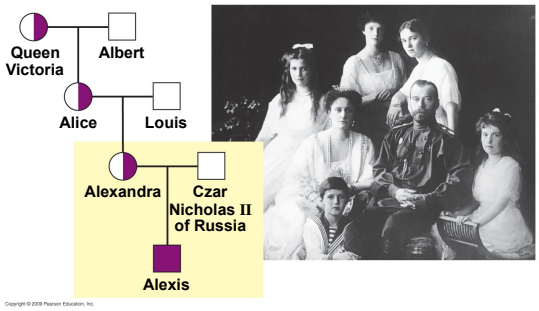
Hemophilia



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Y Chromosome and Evolution

- Similarities in Y chromosome sequences show a significant percentage of men are related to the same male parent
- This demonstrates a connection between people living in distant locations

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