

Mendelian genetics

Asexual reproduction

- ✓ Offspring have genes from one parent
- ✓ Mitotic process
- ✓ Cloning
- ✓ Energy efficient

Sexual reproduction

- ✓ Offspring inherit genes from each parent
- ✓ Genetic recombination (diversity)
- ✓ Meiotic process
- ✓ Uses a lot of energy

Why genetics?

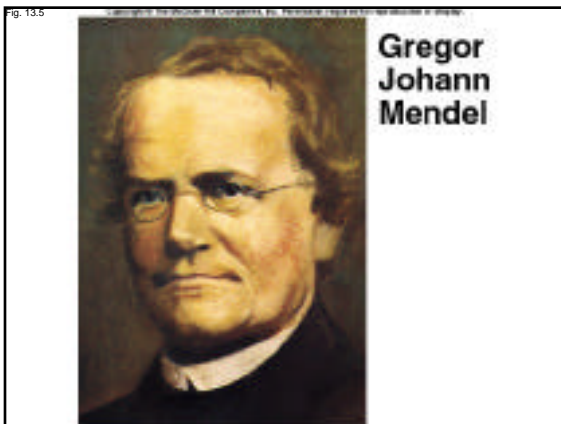
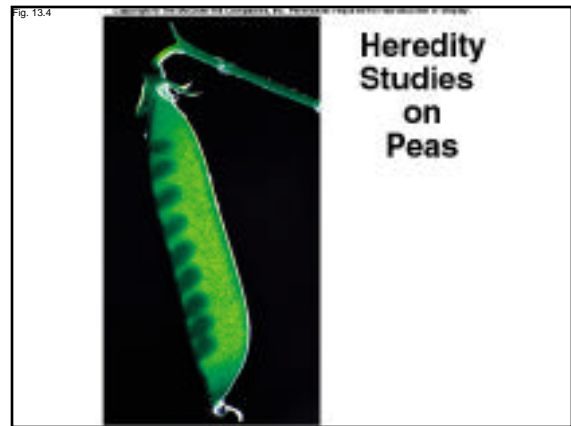
- ✓ Gene therapy, the future of medicine
 - 1. Locate the problem gene on a chromosome
 - 2. Isolate the gene to study its protein structure and function
 - 3. Design a biomolecule to correct for the defect

Definitions

- ✓ Transmission genetics
 - How is variation passed from one generation to the next
- ✓ Molecular genetics
 - How DNA and the cell carry these instructions
- ✓ Phenotype
 - Physical and behavior characteristics
- ✓ Genotype
 - Collection of genes (genome)
- ✓ Gene
 - A region of DNA that codes for a protein

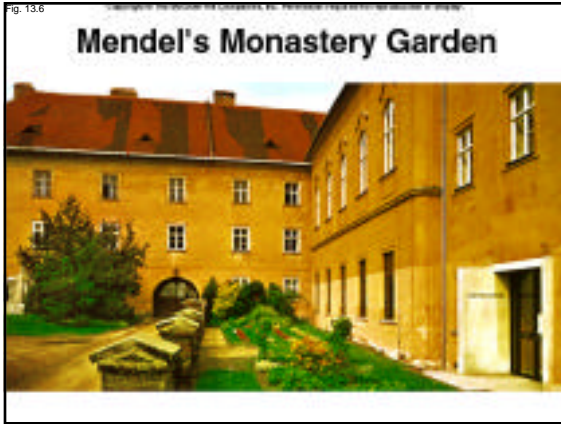
Crossing red and white snapdragons produces pink ones





The foundation of genetics

- ✓ A. Background on Gregor Mendel (1866)
 - Monk
 - Mathematical mind
- ✓ B. Mendel studied inherited traits in garden peas by making hybrids

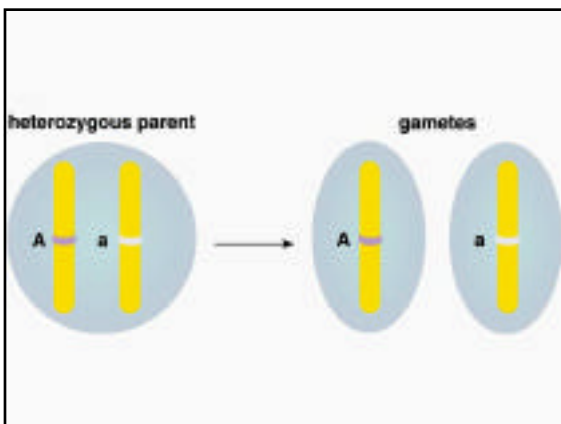
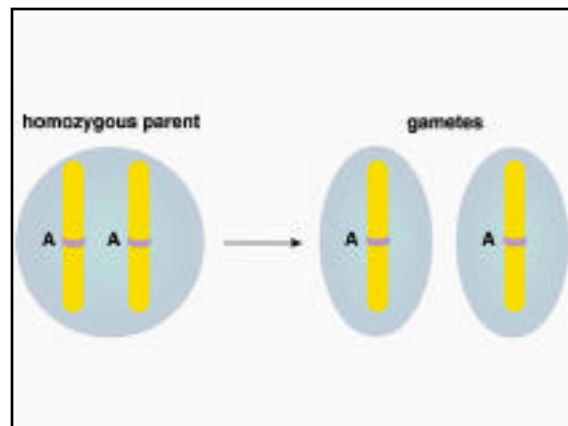


Laws of Inheritance

- ✓ All organisms use DNA as the genetic material.
- ✓ In eukaryotic organisms, DNA is organized into chromosomes.
- ✓ All chromosomes exist as pairs in sexual reproduction.
- ✓ In all eukaryotes, chromosomes behave in the same way during meiosis and fertilization.

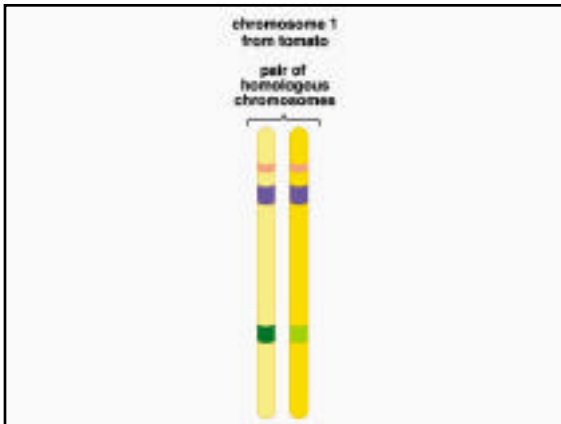
Diploid cells possess 2 copies of a gene

- ✓ Each gene is located on a homologous chromosome.
- ✓ Alternative versions of a gene are called alleles.
- ✓ Alleles are located on the chromosomes--one on each homolog.



Allele pairs

- ✓ If an allele pair are identical, or the cell contains 2 copies of the same allele, the organism is HOMOZYGOUS.
- ✓ If an allele pair are different, or the cell contains two different alleles of a gene, the organism is HETEROZYGOUS.



The foundation of genetics

✓ Mendel's laws

- Law of unit characters (particulate)
 - ↳ a. Paired alleles control the inheritance of traits
 - ↳ b. Homozygous—two of the same allele for a particular gene
 - ↳ c. Heterozygous—two different alleles for a particular gene

Dominance

- ✓ If one member of an allele pair is expressed over the other allele, it is considered **DOMINANT**.
- ✓ The allele that is either not expressed, or whose expression is masked by the expression of the dominant allele is considered **RECESSIVE**.

The foundation of genetics

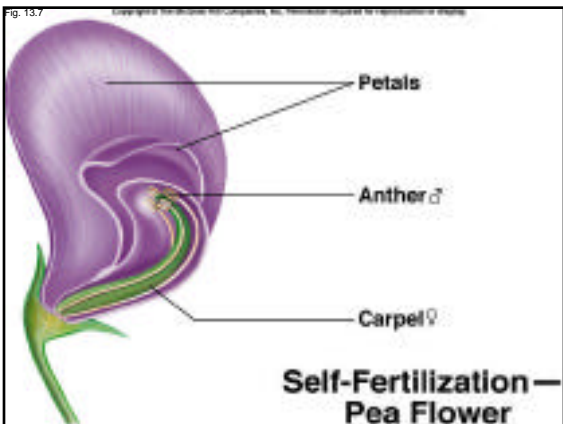
✓ Law of segregation

- ↳ Paired alleles of a gene separate during meiosis, resulting in gametes that contain only a single allele for each gene present in an organism
- ↳ Monohybrid cross is a cross of two parents differing by a single genetic trait
 - Genotype is the genetic makeup of an individual (the alleles present for particular genes)
 - Phenotype is the expressed trait or characteristic (may or may not be visible) resulting from gene expression

The foundation of genetics

✓ Mendel's laws (cont.)

- Law of independent assortment (cont.)
 - ↳ Pairs of alleles (genes) that control different traits segregate independently of each other during meiosis (given that the alleles are on separate chromosomes) *or*
 - ↳ b. Each homologous pair of chromosomes aligns independently of any other homologous pair during metaphase I of meiosis
 - ↳ c. Dihybrid cross is a cross of two parents differing by two genetic traits



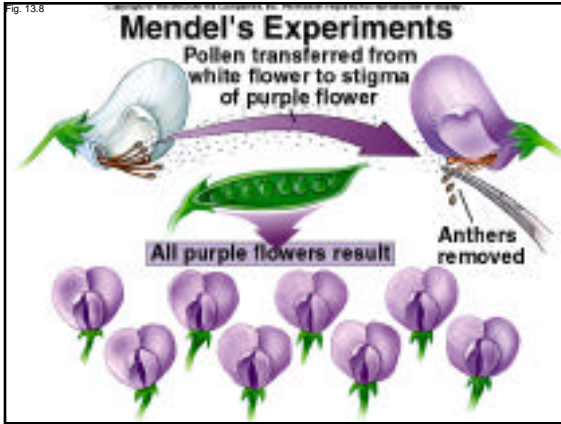
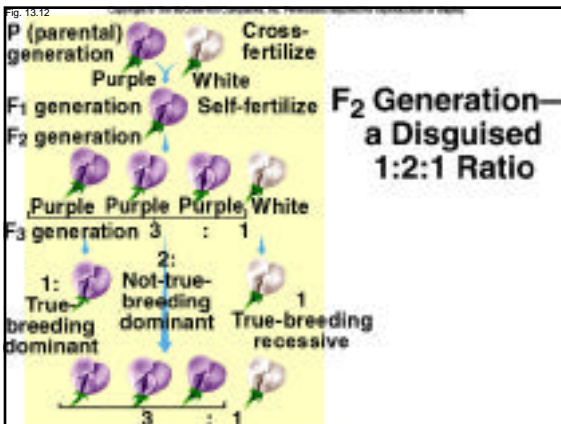
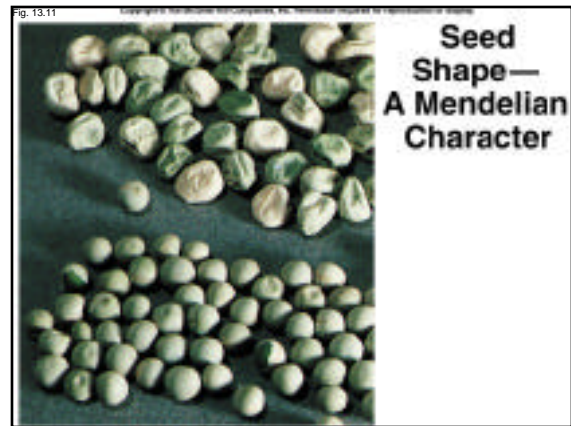
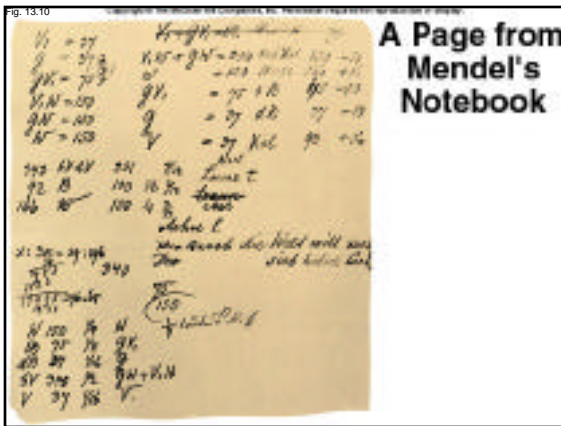


Fig. 13.9

Mendel's Experiment Results

Character	Dominant vs. recessive trait	F ₂ generation: Dominant form	F ₂ generation: Recessive form	Ratio
Flower color	Purple X White	705	224	3.15:1
Seed color	Yellow X Green	6022	2001	3.01:1
Seed shape	Round X Wrinkled	5474	1850	2.96:1
Pod color	Green X Yellow	428	112	3.82:1
Pod shape	Inflated X Constricted	882	299	2.95:1
Flower position	Axial X Terminal	651	207	3.14:1
Plant height	Tall X Dwarf	787	277	2.84:1



Genetic linkage and chromosome maps

- Genes on the same chromosome tend to be inherited together
- Degree of linkage between two genes is a function of the distance between the genes on the chromosome
- Crossing over (during meiosis) rearranges alleles of different genes that were previously linked, creating recombinants
- Recombinants are combinations of genes not found in either of the parents
- Linkage maps estimate relative distances between genes on a chromosome by observing the percentage of recombinant offspring resulting from experimental crosses

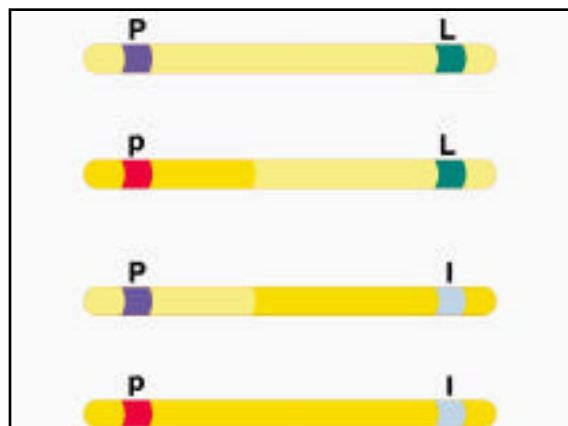
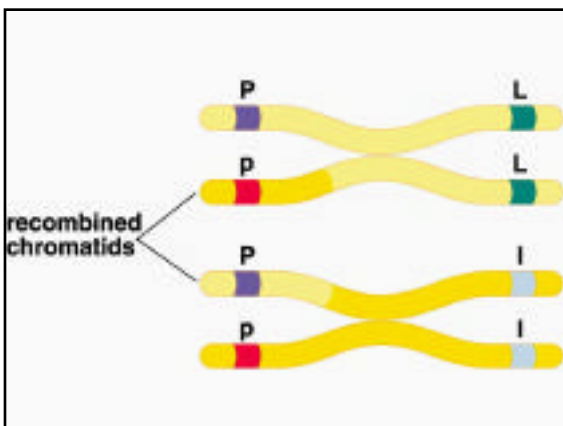
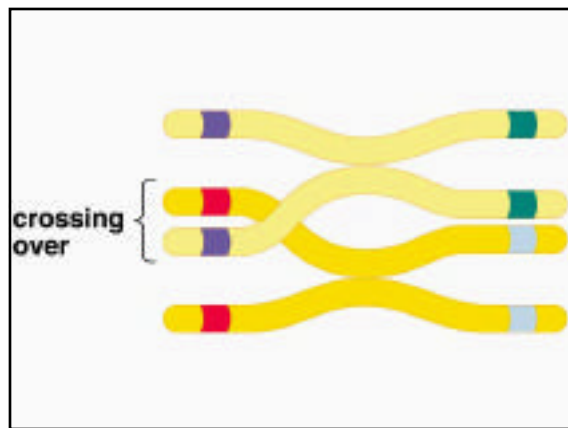
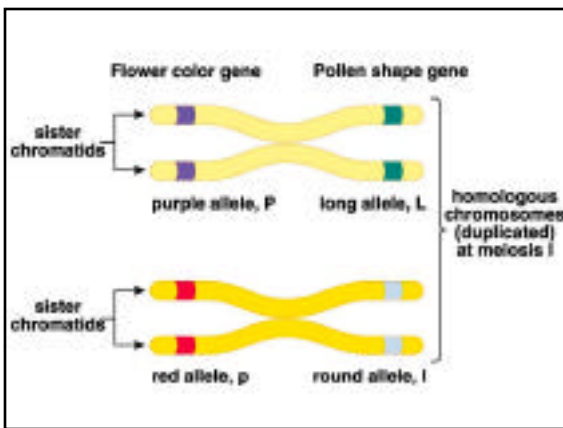
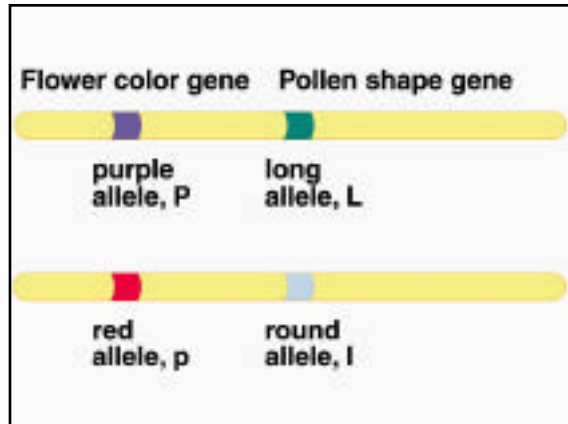
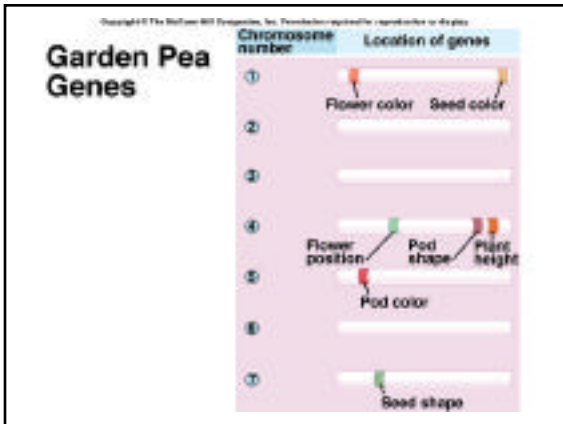
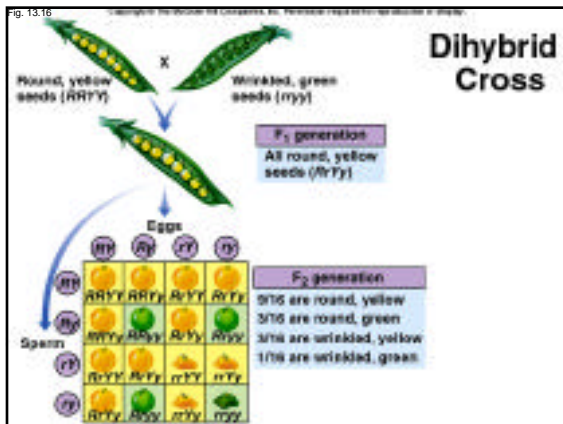
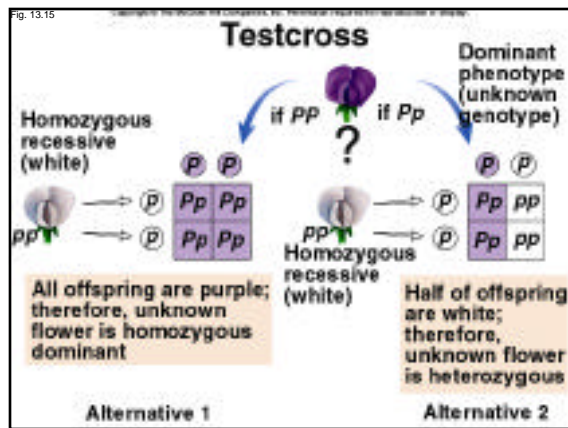
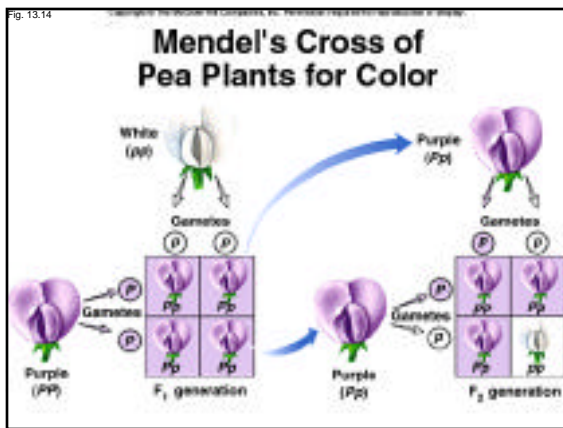
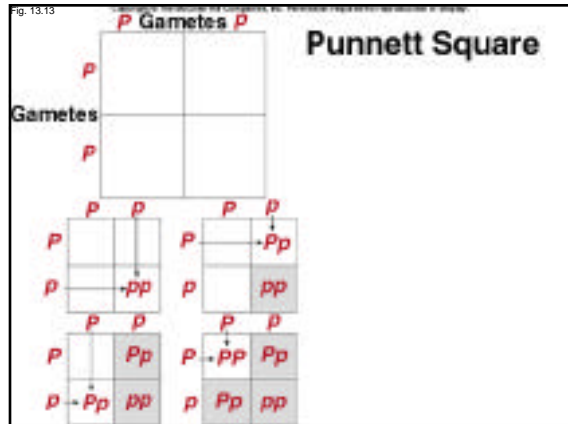


Table 13.1: Some Dominant and Recessive Traits in Humans.

Recessive Trait	Phenotype	Dominant Trait	Phenotype
Albinism	Lack of melanin pigmentation	Midnight hair	Presence of hair on middle segment of finger
Alcaptonuria	Inability to metabolize homogentisic acid	Dark hair/eyes	Short fingers
Red-green color blindness	Inability to distinguish between green and red light	Hemophilia/bleeding	Supernumeration of nipples (extra nipples, arising in middle age)
Cystic fibrosis	Abnormal gland secretion, leading to lung degeneration and/or pancreatic failure	Phenylketonuria (PKU) excretion	Ability to taste PTC as bitter
Chadwick's muscle dystrophy	Tearing away of muscle during childhood	Carotid sinus	Inability to complete the limbic loop
Hemophilia	Inability to form blood clots	Hypertension (often the most serious form)	Downward kink of blood cholesterol and fat in heart attack
Sickle cell anemia	Defective hemoglobin that causes red blood cells to clump and clog	Polidactyly	Extra fingers and toes



- ### Investigating human genetic disorders
- ✓ Pedigree analysis
 - Most human genetic disorders are caused by recessive alleles
 - Sickle-cell anemia, Tay-Sachs, Albinism, cystic fibrosis, PKU
 - A few human genetic disorders are caused by dominant alleles
 - Normally die before reproducing—usually no phenotypic carriers
 - Exception is Huntington's disease—does not appear until later in life
 - Achondroplasia, Marfan syndrome, polydactyly

Pedigree symbols

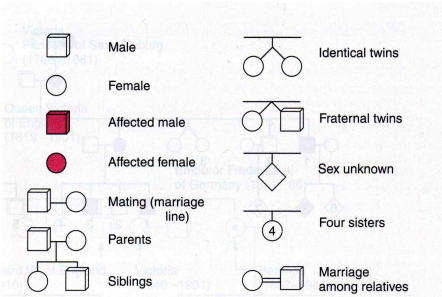


Figure 5.18 Symbols used in a pedigree.

TABLE 13.2 Some Inherited Genetic Disorders

Disorder	Symptoms	Defect	Disinfect/Excessive	Frequency among Human Births
Cystic fibrosis	Mucus plugs lungs, low salt levels	Defect of chloride transport molecule	Recessive	1:3,500
Wick's syndrome	Free blood clotting	Abnormal hemoglobin molecule	Recessive	1:25
Tay-Sachs disease	Elimination of neural enzyme (enzyme in retina)	Defective enzyme	Recessive	1:10,000
Phenylketonuria	Brain cells accumulating in infant	Defective enzyme (phenylalanine hydroxylase)	Recessive	1:10,000
Hemophilia	Blood fails to clot	Defect in blood-clotting factor VIII	Sex-linked recessive	1:10,000
Huntington's disease	Brain cells gradually die; depression in middle age	Presence of an inactive form of gene	Dominant	1:10,000
Hemochromatosis (hemochromatosis)	Excess iron in blood, leading to heart disease	Abnormal form of hemochromatosis	Dominant	1:10,000

Fig. 13.22

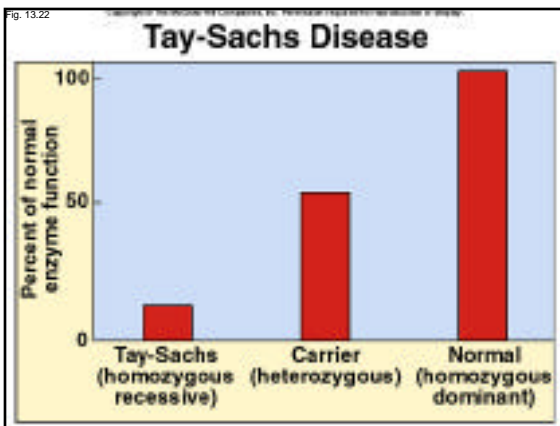
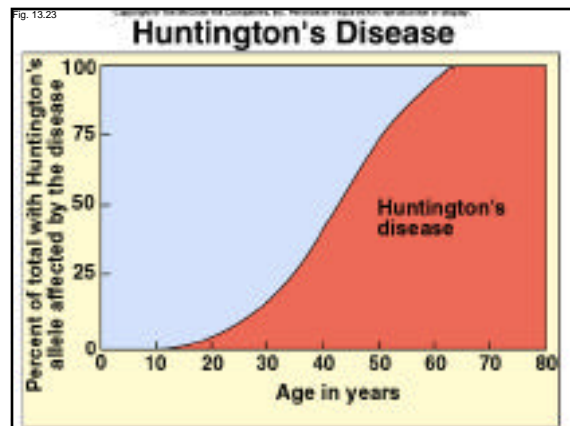


Fig. 13.23



Albinism

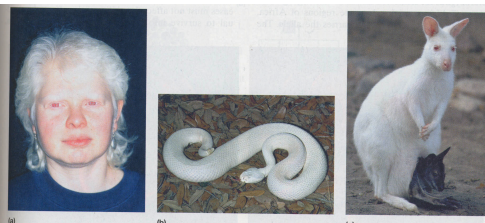
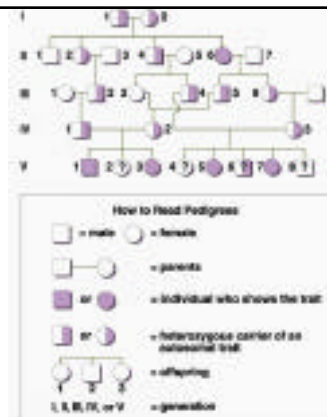
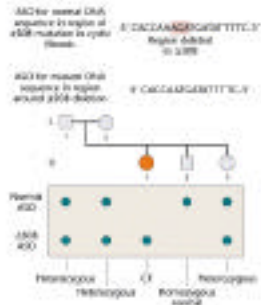


Figure 12-15 Albinism
Albinism is controlled by a single, recessive allele. Melanin is found throughout the animal kingdom, and albinos of many species have been observed: (a) human, (b) rattlesnake, and (c) rabbit. The female vulture, having mated with a normally pigmented male, carries a normally colored offspring in her pouch.

Albinism pedigree



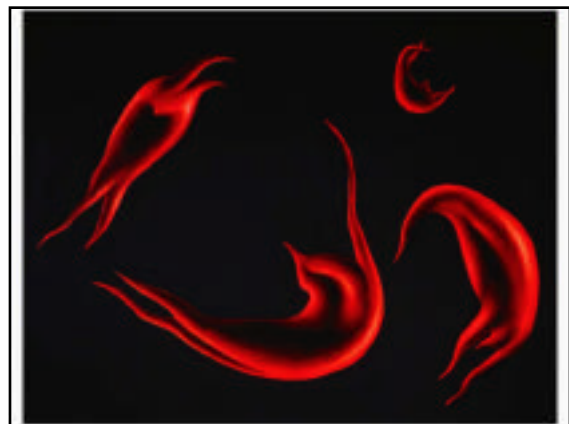
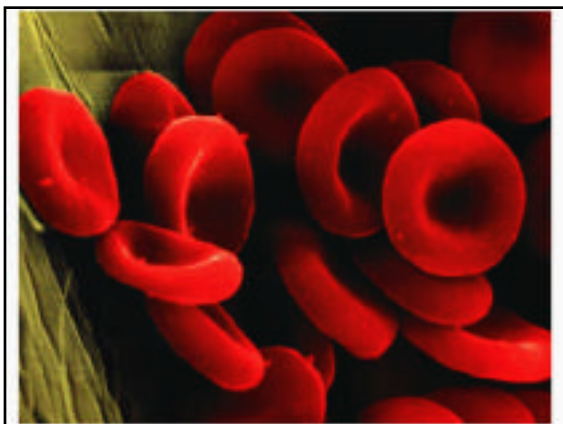
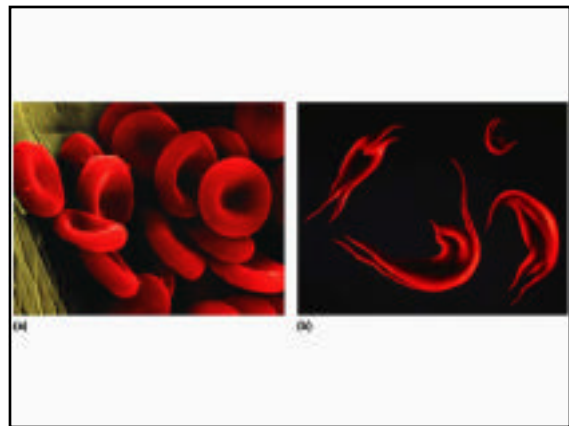
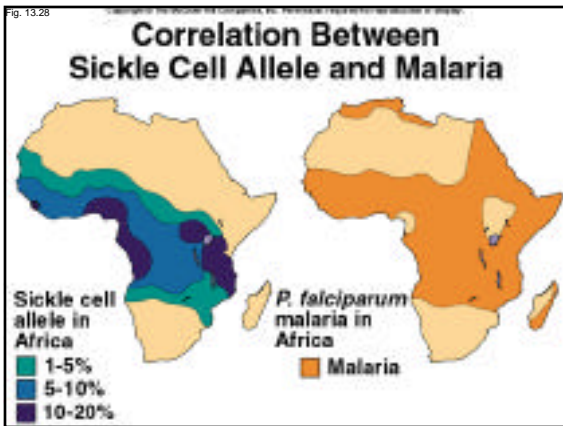
Cystic fibrosis



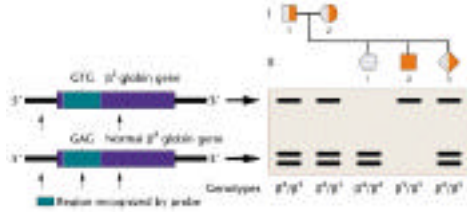
Phenylketonuria (PKU)



Fig. 13.28

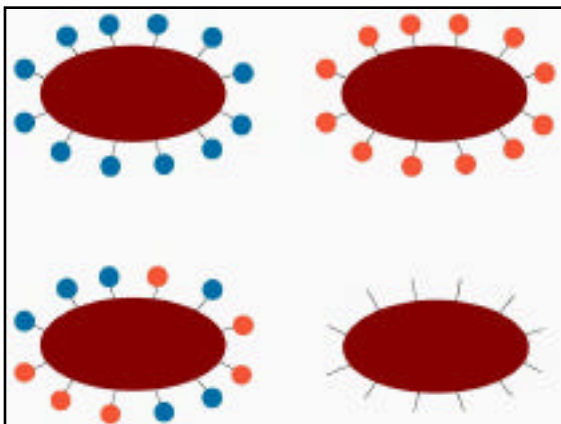
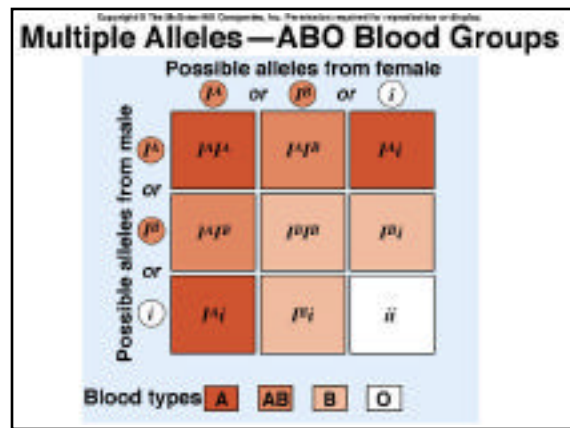
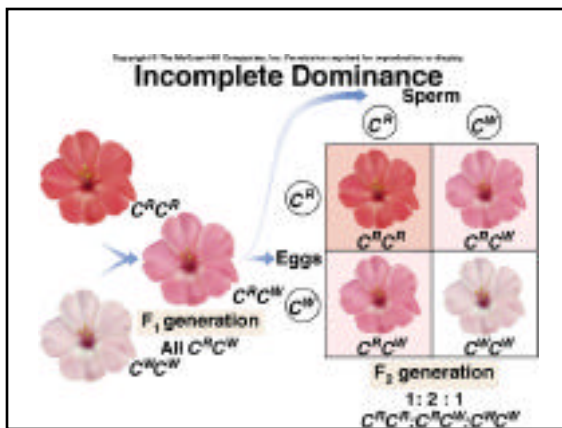


Sickle cell anemia



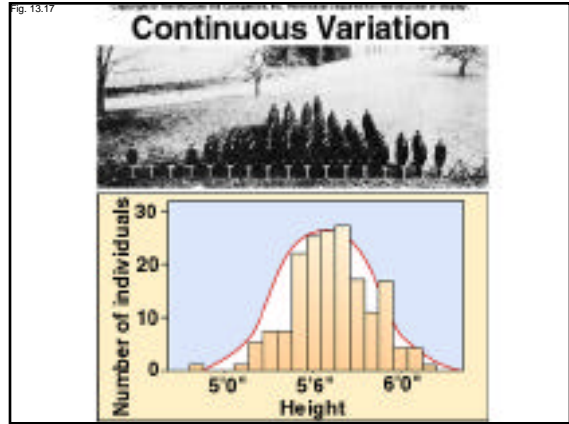
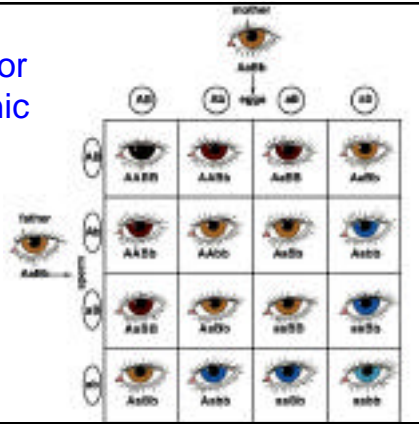
V. Variations on the Mendelian theme

- ✓ A. Incomplete dominance
- ✓ B. Multiple alleles
- ✓ C. Codominance
- ✓ D. Polygenic inheritance
- ✓ E. Gene interactions
- ✓ F. Multiple effects of a single gene
 - 1. Pleiotropy—one gene having multiple effects

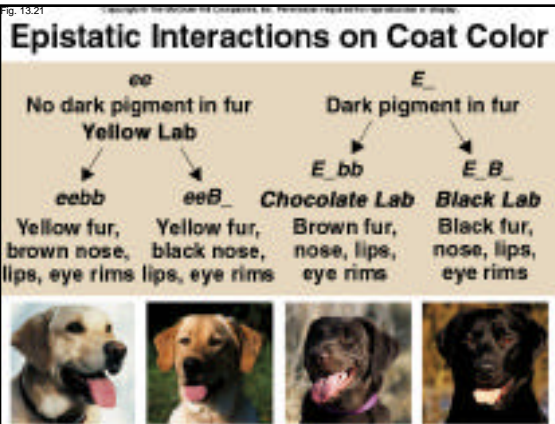
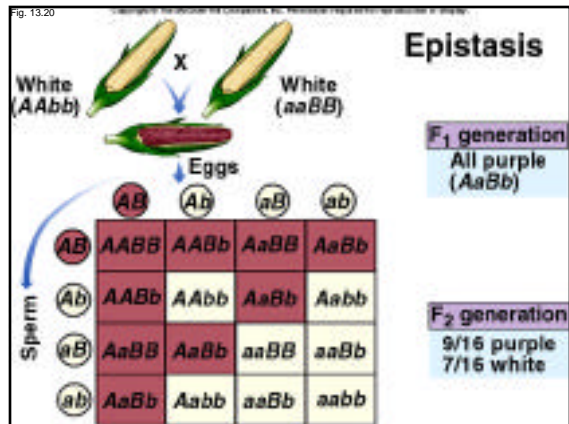
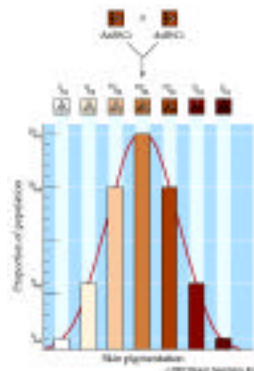


Blood type of cells	Genotype	Antibodies made by body	Reaction to added antibodies	
			Anti-A	Anti-B
A	$I^A I^A$ or $I^A I^O$	Anti-B	Agglutination	No agglutination
B	$I^B I^B$ or $I^B I^O$	Anti-A	No agglutination	Agglutination
AB	$I^A I^B$	Neither anti-A nor anti-B	Agglutination	Agglutination
O	$I^O I^O$	Both anti-A and anti-B	No agglutination	No agglutination

Eye color polygenic



Skin Pigmentation



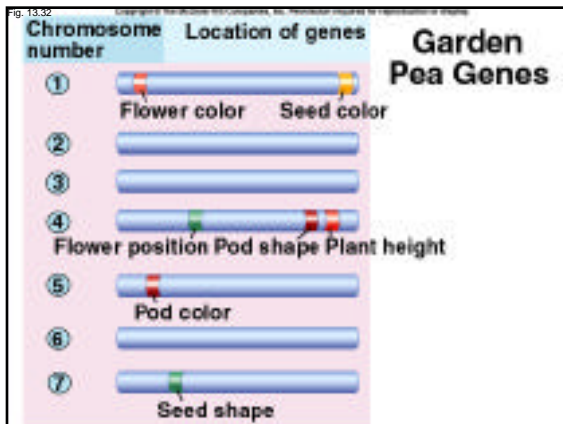
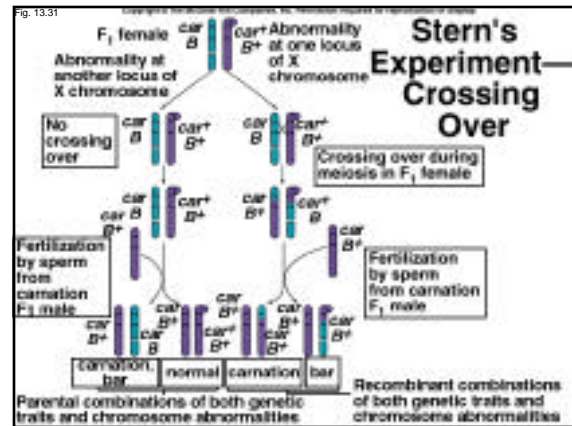
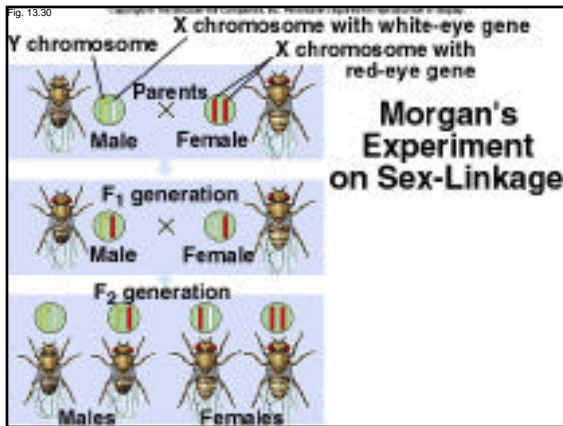
Environmental effects



Investigating human genetic disorders

✓ Sex-linked human disorders

- Baldness?
- Red-green color blindness
- Hemophilia



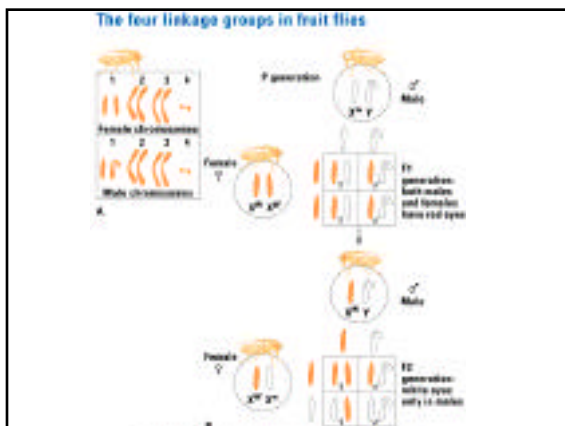
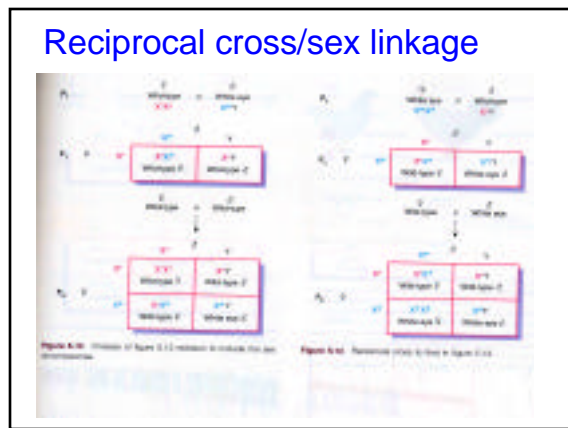
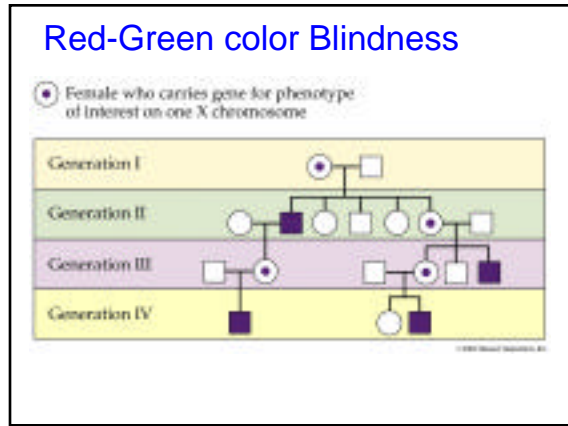
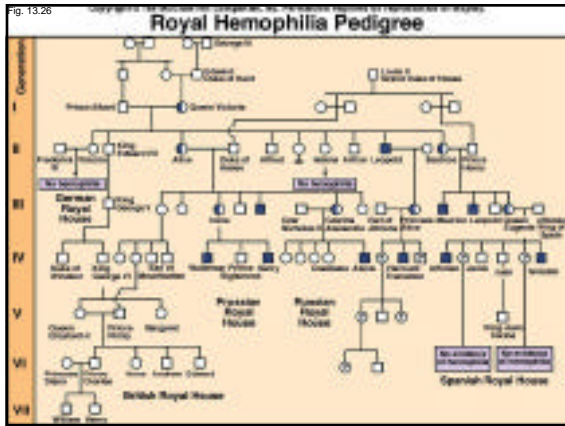
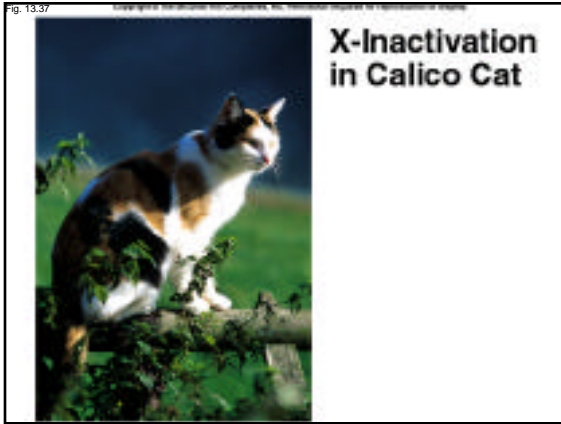


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First Genetic Map

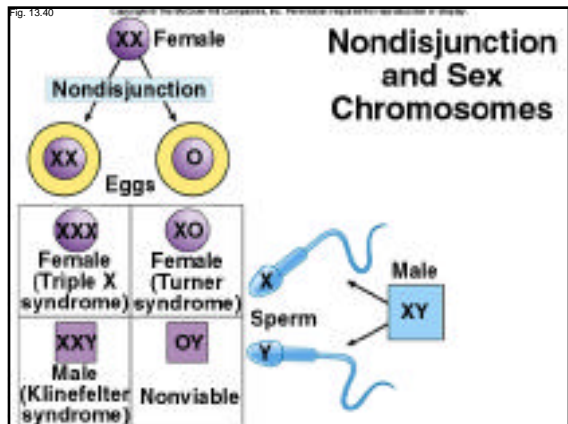
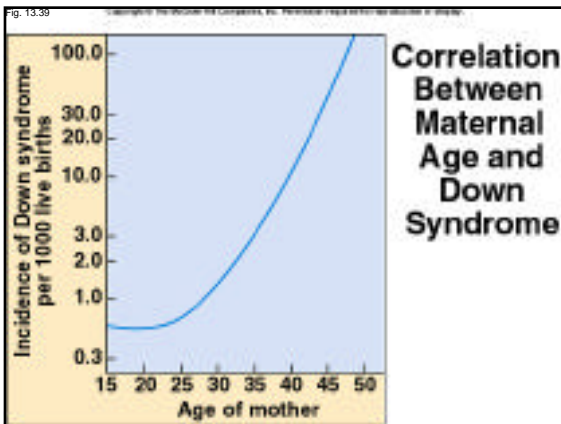
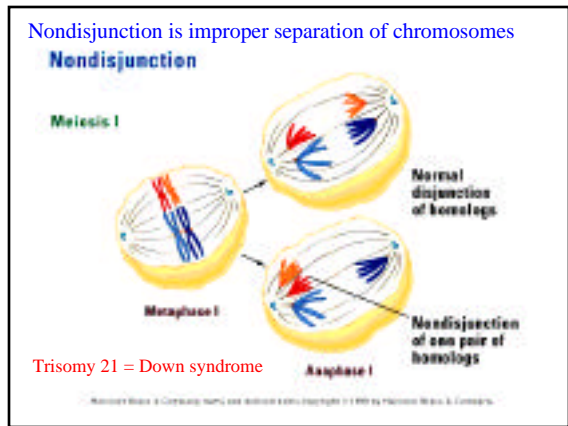
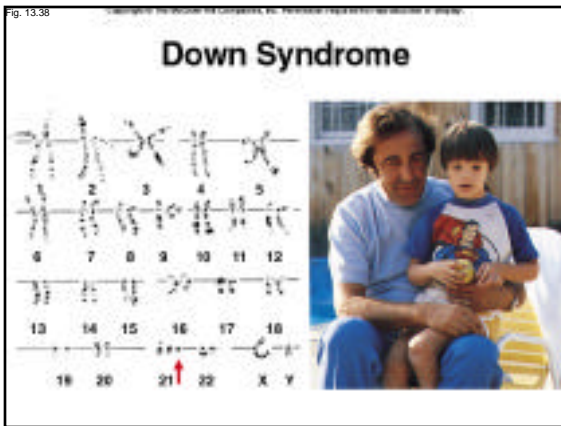
Five traits	Recombination frequencies	Genetic map
<i>y</i> Yellow body color	<i>y</i> and <i>w</i> 0.010	
<i>w</i> White eye color	<i>v</i> and <i>m</i> 0.030	
<i>v</i> Vermilion eye color	<i>v</i> and <i>r</i> 0.269	
<i>m</i> Miniature wing	<i>v</i> and <i>w</i> 0.300	
<i>r</i> Rudimentary wing	<i>v</i> and <i>y</i> 0.322	
	<i>w</i> and <i>m</i> 0.327	
	<i>y</i> and <i>m</i> 0.355	
	<i>w</i> and <i>r</i> 0.450	

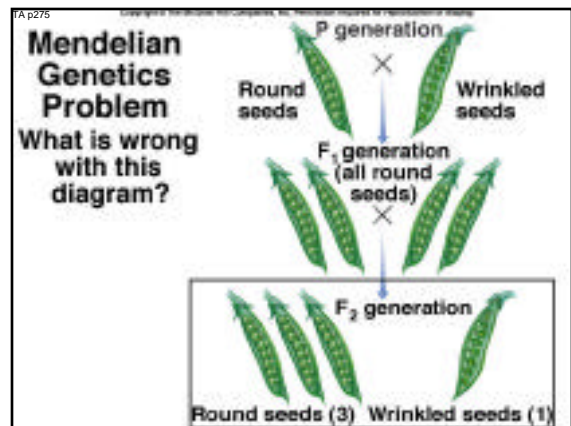
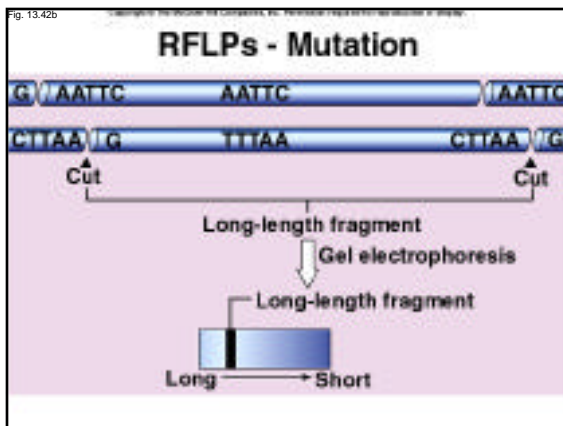
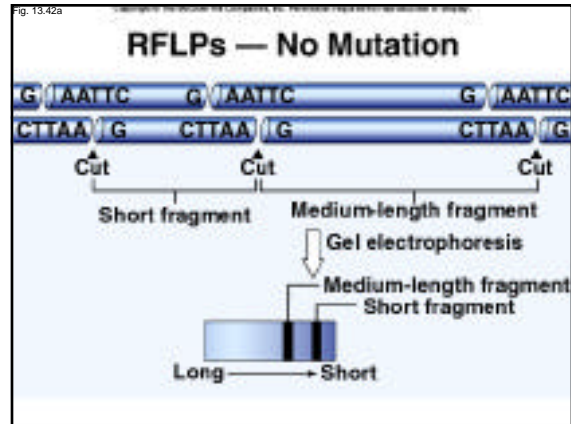
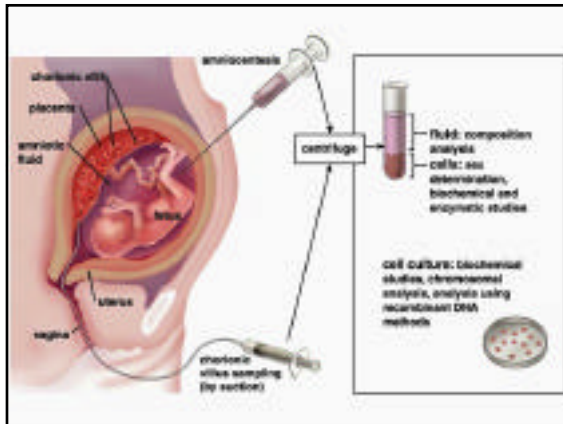


Investigating human genetic disorders

✓ Disorders resulting from nondisjunction during meiosis

- Abnormal number of sex chromosomes
 - ↳ a. Turner's syndrome (XO): sterile, short female; lacks Barr bodies
 - ↳ Trisomy X (XXX): fertile female; no detectable defects; decreased intelligence
 - ↳ Klinefelter's syndrome (XXY): mixed secondary sex characteristics; sterile male
 - ↳ XYY males: decreased intelligence; increased height, increased predisposition for violence
 - ↳ Trisomy 21 (Down syndrome): changes increase with increasing age of mother or possibly father





The End.

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Table 13.A Biocidal Distribution of the Sexes of Children in Human Families

Composition of Family	Order of Birth	Calculation	Probability
3 boys	bbb	$p \times p \times p$	p^3
2 boys and 1 girl	bbg	$p \times p \times q$	p^2q
	bgb	$p \times q \times p$	p^2q
	gbb	$q \times p \times p$	p^2q
1 boy and 2 girls	ggb	$q \times q \times p$	pq^2
	gbg	$q \times p \times q$	pq^2
	bgg	$p \times q \times q$	pq^2
4 girls	gggg	$q \times q \times q \times q$	q^4

9.1 Numbers of Pairs of Chromosomes in Some Plant and Animal Species

COMMON NAME	SPECIES	NUMBER OF CHROMOSOME PAIRS
Mosquito	<i>Culex pipiens</i>	3
Housefly	<i>Musca domestica</i>	6
Toad	<i>Bufo americanus</i>	11
Rice	<i>Oryza sativa</i>	12
Frog	<i>Rana sylvatica</i>	13
Alligator	<i>Alligator mississippiensis</i>	16
Rhesus monkey	<i>Macaca mulatta</i>	21
Wheat	<i>Triticum aestivum</i>	21
Human	<i>Homo sapiens</i>	23
Potato	<i>Solanum tuberosum</i>	24
Donkey	<i>Equus asinus</i>	31
Horse	<i>Equus caballus</i>	32
Dog	<i>Canis familiaris</i>	39
Carp	<i>Cyprinus carpio</i>	52

9.2 Two Different Ways for Cells to Die

	NECROSIS	APOPTOSIS
Stimuli	Low O ₂ , toxins, ATP depletion, damage	Specific, genetically programmed physiological signals
ATP required	No	Yes
Cellular pattern	Swelling, organelle disruption, tissue death	Chromatin condensation, membrane blebbing, single-cell death
DNA breakdown	Random fragments	Nucleosome-sized fragments
Plasma membrane	Burst	Blebbled
Fate of dead cells	Ingested by phagocytes	Ingested by neighboring cells
Reaction in tissue	Inflammation	

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