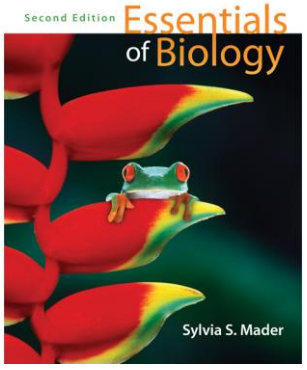
Name _____



Essentials of Biology

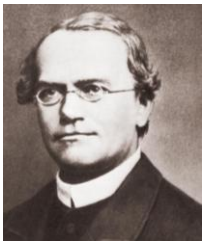
Sylvia S. Mader

Chapter 10
Patterns of Inheritance
Lecture Outline

10.1 Mendel's Laws

Gregor Mendel

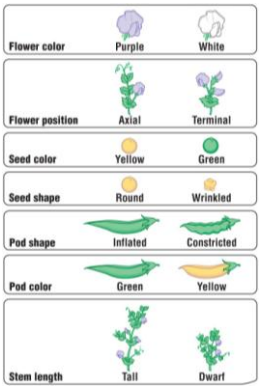
- Austrian monk
- Worked with pea plants in 1860's
- First person to analyze patterns of inheritance
- Deduced the fundamental principles of genetics.



Mendel working in his Abbey Garden

- **Studied garden peas**
  - Easily manipulated
  - Can self-fertilize
  - Many traits controlled by only one gene

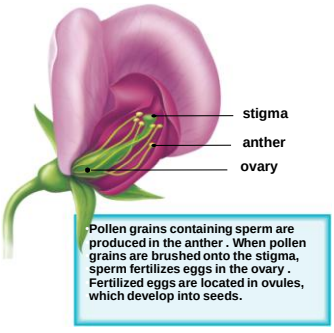
Figure 10.1



Mendel's Experimental Procedure

- Used garden pea, *Pisum sativa*
  - Easy to cultivate, short generation time
  - Normally self-pollinate but can be cross-pollinated by hand

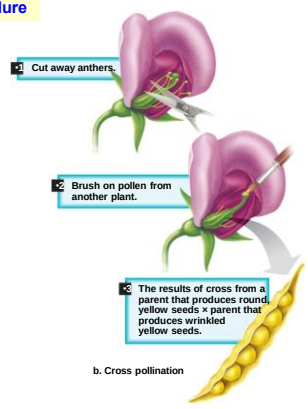
Figure 10.2
Garden pea anatomy & traits



a. Flower structure

Mendel's Experimental Procedure

- Used true-breeding varieties
  - offspring were like the parent plants and each other
- Kept careful records of large number of experiments
- Used mathematical laws of probability to interpret results to develop his theory of inheritance

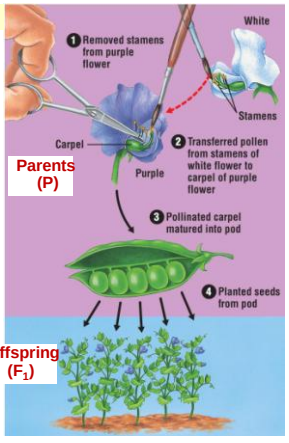


b. Cross pollination

Figure 10.2 (cont.)

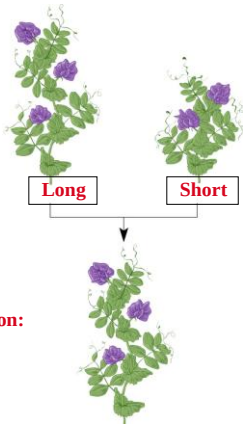
Mendel's Experimental Procedure

- Used true-breeding varieties
  - offspring were like the parent plants and each other
- Kept careful records of large number of experiments
- Used mathematical laws of probability to interpret results to develop his theory of inheritance



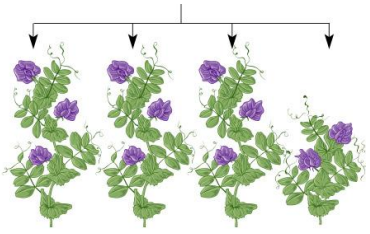
Cross between true breeding long and short pea plants

Which phenotype is dominant? Recessive?



Self-fertilization of F_1

How did Mendel Explain these results?

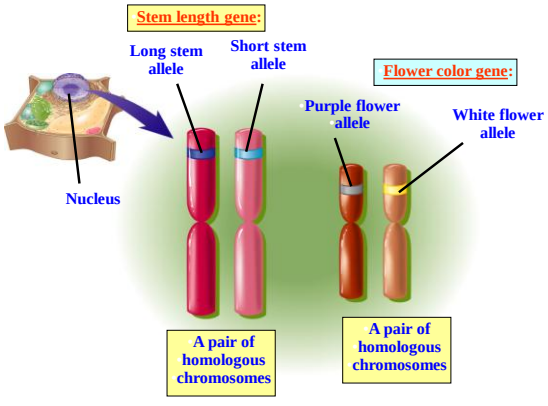


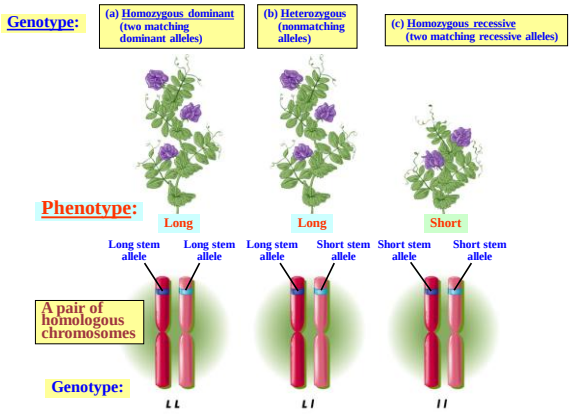
F_2 Long Long Long Short

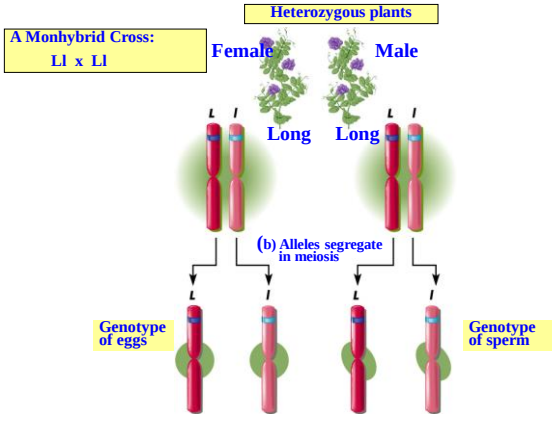
F_2 Phenotypic Ratio: (3 Long : 1 Short)

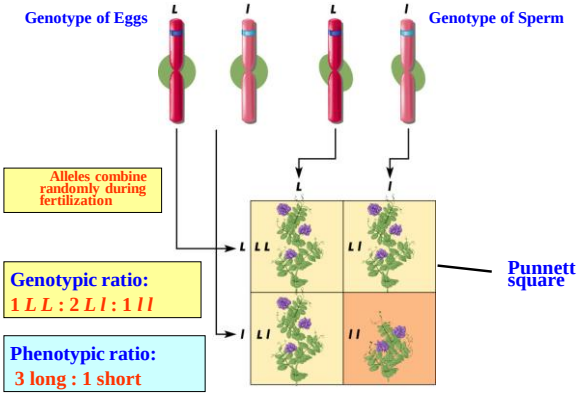
¾ of offspring are long

¼ of the offspring are short









Mendel's Law of Segregation

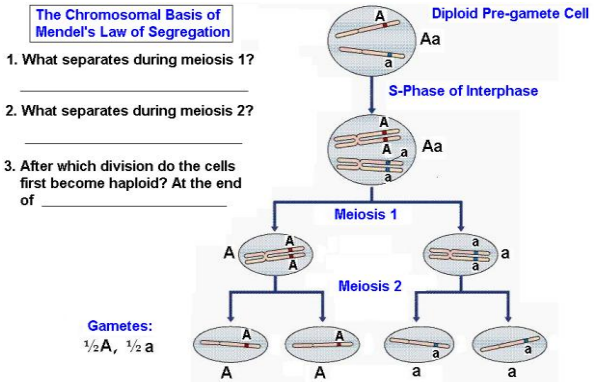
- Alleles separate from each other during meiosis
- Results in gametes with one or the other allele, but never both

Formation of gametes from a pre-gamete cell

Genotype of Pre-gamete cell: Aa

meiosis

Genotype of Gametes: ½ A ½ a



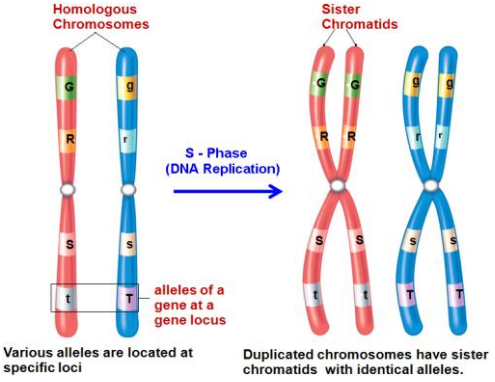
Genetic Terms

1. Phenotype Vs. Genotype—what's the difference.
2. What are Alleles?
   - Alternate forms of a gene… e.g.'s?
3. Where are the alleles of a gene located?
4. How many alleles can a person inherit for any one trait?
   - How many alleles are there in a population for a particular trait such as hair color?
5. What's the relationship between alleles and homologous chromosomes?
6. Dominant vs. recessive alleles—what's the difference? E.g.'s?
7. How can you determine if an allele is dominant or recessive?

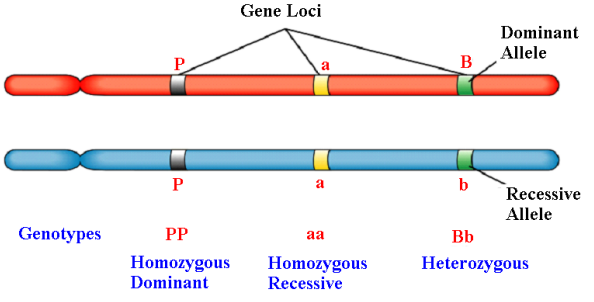
Genotype vs. Phenotype

- **Genotype**: alleles individual receives at fertilization
  - **Homozygous**: **2 identical alleles**
    - Homozygous dominant
    - Homozygous recessive
  - **Heterozygous: 2 different alleles**
- **Phenotype**: physical appearance of individual
  - Determined by genotype and environmental factors

Figure 10.5 Alleles on Homologous chromosomes



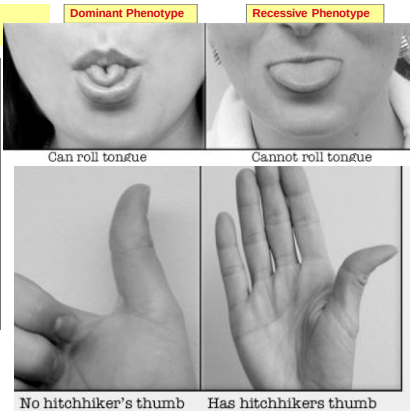
Homologous Pair of Chromosomes with 3 linked genes



Dominant vs. Recessive Phenotype

Family Pedigrees

- Shows the history of a trait in a family from one generation to another
- Allows researchers to determine **if a phenotype is dominant or recessive**



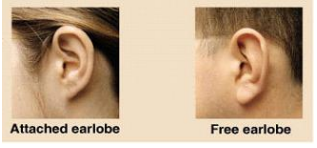
Types of Genotypes and their resulting Phenotypes

Genotype		Phenotype
RR = _____	→	_____
Rr = _____	→	_____
rr = _____	→	_____

Allele Symbols:
R = Tongue Roller
r = Nonroller

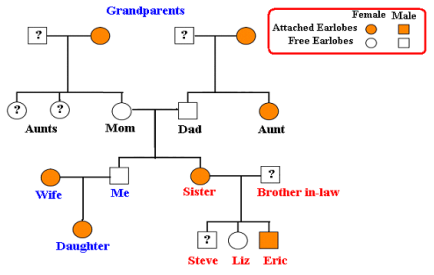
Attached vs. Free Earlobes

1. Due to a recessive or dominant allele?
2. Sex-linked or Autosomal?
3. Must examine a pedigree to answer these questions



Marr Family Pedigree for Earlobe Attachment

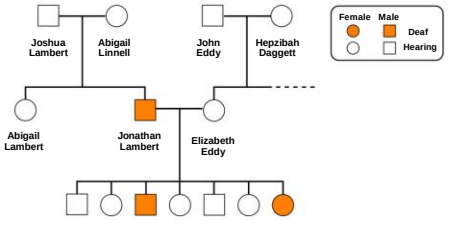
1. Which allele is dominant? Recessive?
- Is the allele for earlobe attachment sex-linked (X-linked) or autosomal?
- What are the genotypes of all family members?



A family pedigree for Deafness

- Is deafness a dominant or recessive trait?
- How can you tell?

Allele Symbols: D = _____ d=_____



Common Monogenic Human Traits

	Dominant Allele	Recessive Allele
1.	Free Earlobes	Attached Earlobes
2.	Straight Thumb	Hitchhiker's thumb
3.	Long eyelashes	Short eyelashes
4.	Normal health	Cystic Fibrosis
5.	Normal health	Tay-Sac's Disease
6.	Normal R.B.C's	Sickle cell anemia
7.	Huntington's Disease	Normal Health

Common Polygenic Human Traits

	Dominant	Recessive
1.	Dark-colored hair	Light-colored hair
2.	Curly hair	Straight hair
3.	Dark eyes	Light eyes (blue or gray)
4.	Hazel or green eyes	Blue or gray eyes
5.	Tall	Short
6.	Dark skin	Light skin

Common Sex-linked Recessive Human Traits: X-linked

X-linked recessive traits

- **Uncommon in females—why?**
- **Father must have disease and mother must be a carrier for a daughter to have the disease.**

1. **Color Vision**

   X^N = Normal color vision; X^n = Red/Green Colorblind

2. **Hemophilia:**

   X^N = Normal blood clotting; X^n = bleeder

3. **Duchenne Muscular Dystrophy**

   X^N = Normal muscles; X^n = muscular dystrophy

Laws of Probability – application to inheritance

1. **The results of one trial of a *chance event* do not affect the results of later trials of that same chance event**
   - **E.g. Tossing of a coin, gender of children, etc.**

Laws of Probability – application to inheritance

2. **The Multiplication Rule: The chance that two or more <u>independent chance events</u> will occur together is equal to the *product* of their chances occurring separately**
   a. **What are the chances of a couple having 9 girls?**
   b. **E.g. What are the chances of a couple having a boy with the following characteristics:**
      - **Brown hair (3/4), Non-tongue roller (1/4), Blue eyes (1/4), Attached earlobes (1/4)**

How to Solve Genetics Problems

<u>Sample Problem</u>: Mom and dad are heterozygous for tongue rolling where tongue rolling is dominant to non-rolling. What is the chance that the couple will produce a girl that is a non-roller?

<u>Use the following steps as a general guide to solve this and other problems:</u>

1. **Select a letter to represent the gene involved**
   - **Use upper case for the dominant allele, lower case for the recessive allele.**
2. **Write the genotypes of the parents.**
3. **Determine all possible gametes for each parent.**
   - **Alleles for a trait segregate into separate gametes during meiosis**
4. **Determine the genotypes of the offspring.**
   - **Make a Punnett square to represent all possible gamete combinations between the two parents**
5. **Use the genotypes found in the Punnett Square to determine the possible phenotypes of the offspring to answer the question.**

Types of Genetics Problems

<u>Monohybrid Crosses</u>

- **Involve only one trait such as …??**

<u>Sample Problem #1</u>: True breeding parental pea plants were crossed to produce the F_1 generation, below. The F_1 generation was inbreed to produce an F_2 generation.

a.) Which allele is dominant? How do you know?

b.) Determine the genotypes and phenotypes for all 3 generations

c.) Predict the genotypic and phenotypic ratios for the F_2.

P: Purple flowered pea plant x White flowered Pea Plant

F_1: 100% Purple Flowered

F_2: ???

Monohybrid Cross Sample Problem #2

A true breeding black mouse was crossed with a true breeding brown mouse to produce the F_1 generation, below. The F_1 generation was then inbred to produce an F_2 generation.

a.) Which allele is dominant? How do you know?

b.) Determine the genotypes and phenotypes for all 3 generations

c.) Predict the genotypic and phenotypic ratios for the F_2.

P: Black mouse x Brown mouse

F_1: 100% black mice

F_2: ???

Monohybrid Cross Sample Problem #3

A mouse with black fur was crossed with a mouse with brown fur to produce the F_1 generation, below. The F_1 generation was then inbred to produce the F_2 generation. Dominance is the same as in sample problem #2.

a.) Determine the genotypes and phenotypes for all 3 generations

b.) Predict the genotypic and phenotypic ratios for the F_2.

P: Black mouse x Brown mouse

F_1: ½ black mice; ½ Brown

F_2: ???

Monohybrid Cross Sample Problem #4

Use the information below to answer the following questions. Dominance is the same as in the preceding problems involving mice.

a.) Calculate the phenotypic ratio of the F_2.

b.) Determine the genotypes and phenotypes for all 3 generations

c.) Determine the expected phenotypic and genotypic ratios for the F_2.

d.) Explain why the expected phenotypic ratio is *different* than the actual phenotypic ratio for the F_2.

P: ?????????? x ??????????

F_1: 100% black mice

F_2: 27 Black mice + 10 Brown mice

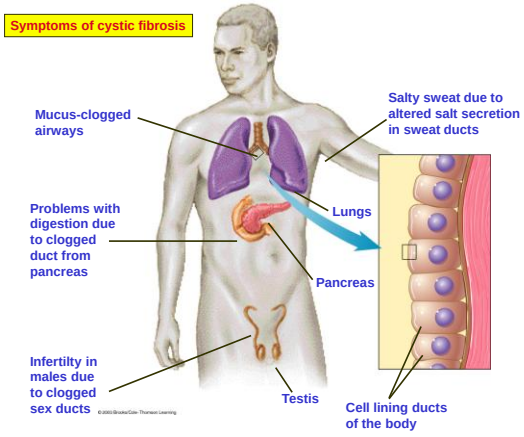
Monohybrid Cross Sample Problem #5

A couple, Jack and Jill, is concerned about having a child with cystic fibrosis. Although both of Jack's and both of Jill's parents are healthy and show no signs of cystic fibrosis, both Jack and Jill each had a sister die of the disease. The couple went to a clinic to be genetically tested for cystic fibrosis and were each found to be heterozygous for cystic fibrosis. What are the chances of Jack and Jill having a….

a.) phenotypically healthy child?

b.) child that is homozygous dominant? Heterozygous? Homozygous recessive?

c.) girl with cystic fibrosis? Boy with cystic fibrosis?



Monohybrid Cross Sample Problem #6

Gaucher disease is an *autosomal recessive disorder*. What are the chances of a phenotypically normal and healthy couple having a child with Gaucher disease if *each partner has a brother with GD and the parents of the couple are phenotypically healthy*?

Hints: (This problem is *more complex* than you may think!)

1.) Neither couple knows their genotype.

2.) Being phenotypically healthy eliminates one of the possible genotypes for the couple.

Test Cross

- Used to determine if an organism with the dominant phenotype is homozygous dominant or heterozygous
- Involves the cross of an organism with the dominant phenotype with _____.

e.g. Free earlobes is dominant to attached earlobes in humans. How could your instructor determine if he is homozygous or heterozygous for free earlobes?

Instructor (Free Earlobes) X Wife (_____)

↓

Daugher (_____)

Conclusion??

Types of Genetics Problems

Dihybrid Crosses

- Involve two traits. Such as …??

Dihybrid Cross Sample Problem #1: True breeding parental pea plants were crossed to produce the F_1 generation, below. The F_1 generation was inbreed to produce an F_2 generation.

a.) Which alleles are dominant? How do you know?

b.) Determine the genotypes and phenotypes for all 3 generations

P: Long & Purple flowered pea plant x Short & White flowered Pea Plant

F_1: All Long & Purple Flowered pea plants

F_2: 9 Long & Purple : 3 Long & White : 3 Short & Purple : 1 Short & White

In general, the F_2 of a Dihybrid Cross: 9 D_1 & D_2 : 3 D_1 & R_2 : 3 R_1 & D_2 : 1 R_1 & R_2

Illustration of the Dihybrid Cross in Sample Problem #1

(slide 1 of 2)

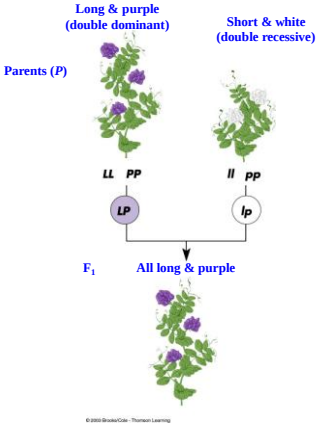
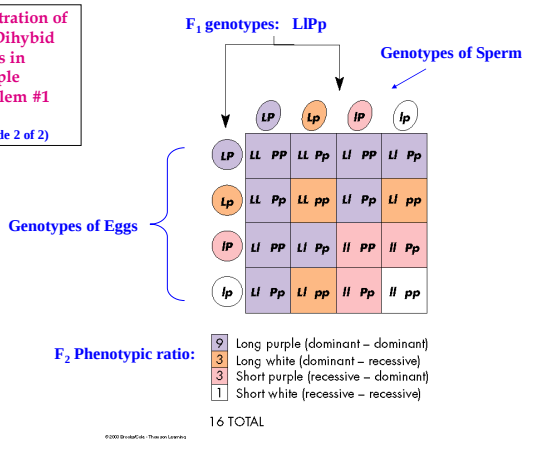


Illustration of the Dihybrid Cross in Sample Problem #1

(slide 2 of 2)



Explaining Dihybrid Crosses

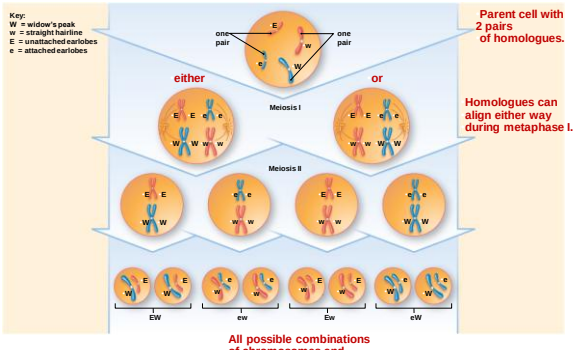
Mendel's Law of Independent Assortment

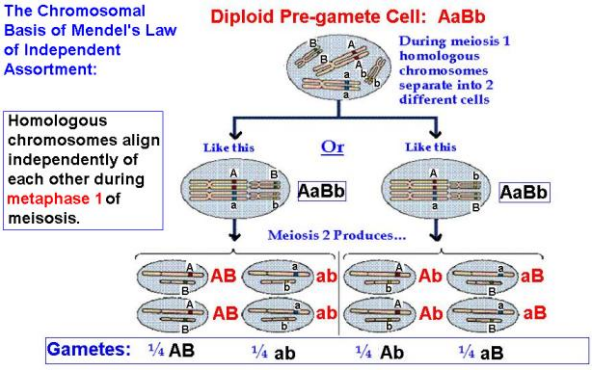
- Each pair of alleles separates (segregates) independent from other pairs of alleles during gamete formation unless the genes for these alleles are found on the same chromosome
- Results in a 9 : 3 : 3 : 1 Phenotypic Ratio in the F_2!

Figure 10.8 Mendel's laws and meiosis

- Gene for earlobes and hairline on different chromosomes
- Gametes have all possible combination of alleles.

Figure 10.8 Potential gametes produced by a person who is *EeWw*





Determining Gametes for traits that assort independently

- **Traits that assort independently are on different homologous pairs of chromosomes—I.e. the traits are not linked.**
- **Number of genetically different gametes possible = 2^n (where n = the number of heterozygous traits)**
- **Practice Problems**
  - **How many genetically unique gametes are possible for the following genotypes? List the genotypes of all possible gametes for #'s 1-5, below.**

1. **AaBb**
2. **AABb**
3. **AABBCC**
4. **AaBbCc**
5. **AaBBCc**
6. **AaBbCcddEe**

Using the Probability Method to Solve "Multi-hybrid" Problems

- From the crosses below, what are the chances of producing an organism with all
  - dominant phenotypes?
  - recessive phenotypes?
  - homozygous dominant genotypes?

1. AaBb x AaBb
2. AaBbCc x AaBbCc
3. AaBBCc x aabbcc

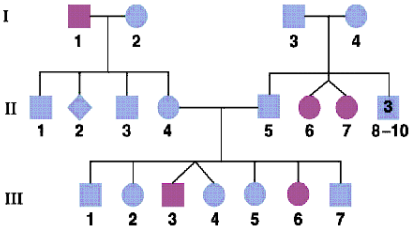
How to use the probability method

1. Treat the problem as if it consisted of several monohybrid crosses
2. Determine the gametes for each of these monohybrid crosses
3. Make a Punnett square for each of the monohybrid crosses
4. Use the information from each Punnett square and the "multiplication rule" to solve the problem

Analysis of Pedigrees

1. **Is the disease dominant or recessive? How can you tell?**
2. **Autosomal or Sex-linked inheritance? How can you tell?**
3. **Can you determine the genotypes of all individuals?**
   - **For which phenotype do we always know the genotype?**

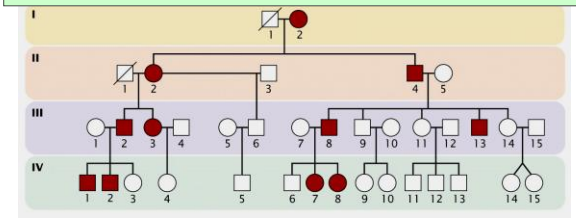
Pedigree #1 (Purple shading indicates genetic disease)



Analysis of Pedigrees

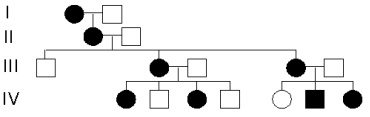
1. Is the disease dominant or recessive? How can you tell?
2. Autosomal or Sex-linked inheritance? How can you tell?
3. Can you determine the genotypes of all individuals?
   - For which phenotype do we always know the genotype?

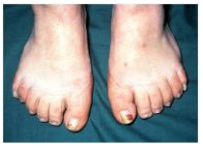
Pedigree #2 (shading indicates genetic disease)

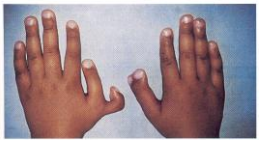


Human Polydactyly: Extra Finger or Toe

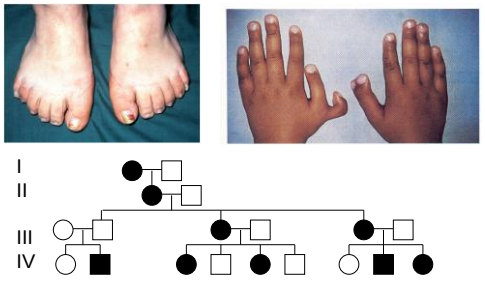
1. Is the disease dominant or recessive? How can you tell?
2. Autosomal or Sex-linked inheritance? How can you tell?
3. Can you determine the genotypes of all individuals?





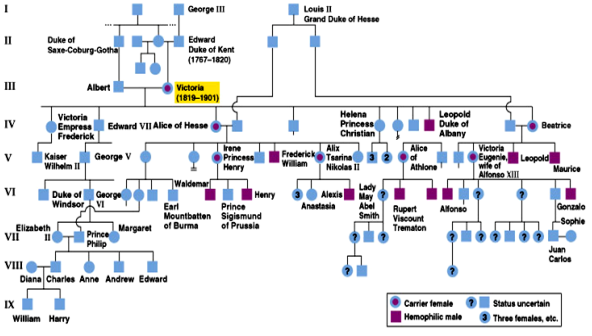


Human Polydactyly



Analysis of British Royal Family Pedigree

1. Is the disease dominant or recessive? Autosomal or sex-linked?
2. Determine genotypes



10.2 – 10.3 Beyond Mendel's Laws

- Sex-linked recessive inheritance
  - Recessive on X-chromosome
    - e.g. Hemophilia, colorblindness, Androgen Insensitivity Syndrome (e.g. Jamie Lee Curtis?) → http://www.medhelp.org/www/ais/ ; http://www.youtube.com/watch?v=ETIxoQGVjos
- Incomplete dominance
  - e.g. Snapdragons
    - red flower x white flower → pink flower
  - Sickle cell anemia

    NN = healthy; nn = sickle cell anemia (deadly); Nn = sickle cell trait
- Co- dominance: ABO Blood Groups
  - Blood types: A, B, AB, O

Is it possible to be XY and female? XX and male?

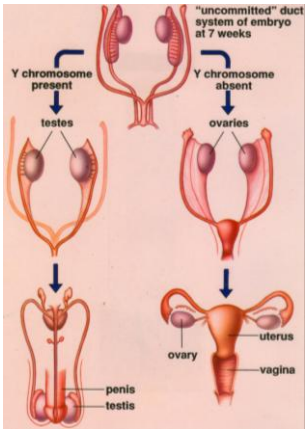
The Maria Patino Story

- Maria Patino couldn't sleep before her 1st race at the 1985 World University Games in Japan. She was the Spanish National Champion and scheduled to perform in the 60m hurdles the next day but she wasn't sure if she would be able to compete. Earlier that day she reported to ***"Sex Control"*** which scraped cells from her cheek to test for sex chromosomes. She had passed the test in 1983 in Helsinki but had forgotten to bring her "Certificate of Femininity". A few hours after the test officials told her the test was abnormal but not to worry. But she worried all night. Did she have leukemia that killed her brother? Did she have AIDS? The next morning they did a follow up check and **she failed the sex test! She had male sex chromosomes, XY!** Sports officials decided Maria should fake an injury in warm-up so no one would suspect why she withdrew. Spanish officials told her she had to drop out of sports.
- Maria was aghast:
  - "I knew I was a women in the eyes of medicine, God and most of all, in my own eyes."
  - It came out in the newspapers. Her boyfriend left her and other friends also. Spanish sports officials took her records out of the record books.
- **Marias phenotype**: female genitalia, female body proportions, sexually attracted to males, but no uterus, sterile and no pubic hair.
- Let's investigate how sex is determined to try to figure out what is happening.

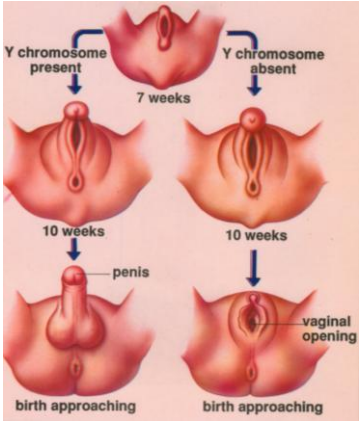
Human Sex Chromosomes

- **Sex chromosomes in humans**
  - » Female Genotype = XX
  - » Male Genotype = XY
- **Sex-linked Alleles are carried on the X-chromosome**
  - » **~1000 genes on X-chromosome**
- **Y-chromosome**
  - » **Only ~20 genes on Y-chromosome**
    - – Mostly involved with male fertility
  - » **SRY gene** on Y chromosome activated around the 7th week of pregnancy
    - – **Gene product stimulates gonads to differentiate into male sex organs.**
    - – **SRY** = Sex-determining Region, Y-chromosome

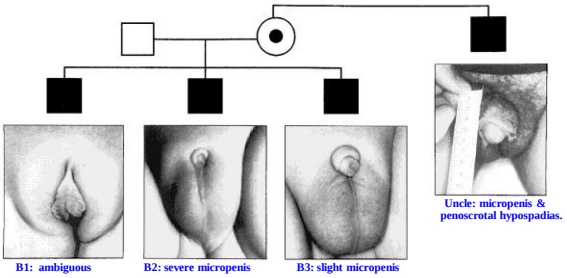
Effect of SRY Gene Activation on the Development of the Internal Sex Organs



Effect of SRY Gene Activation on the Development of the External Sex Organs



Pedigree of a family with varying degrees of Androgen Insensitivity



A family with four affected individuals, three brothers (B1–3) and their uncle, displaying strikingly different external genitalia.

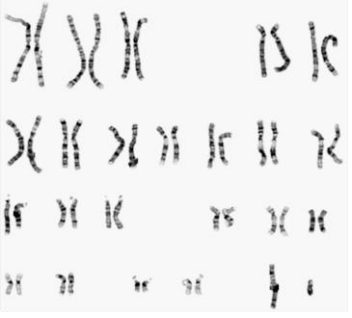
Holterhus, P. et al. 2000. ***Journal Clinical Endocrinol Metab.*** **85: 3245-3250** (Avail. http://jcem.endojournals.org/cgi/content/full/85/9/3245)

Abstract

Molecular causes of phenotypic diversity in androgen insensitivity syndrome, occurring even in the same family, have rarely been identified. We report on a family with four affected individuals, three brothers (B1–3) and their uncle, displaying strikingly different external genitalia: B1, ambiguous; B2, severe micropenis; B3, slight micropenis; and uncle, micropenis and penoscrotal hypospadias. All had been assigned a male gender. We detected the same L712F mutation of the androgen receptor (AR) gene in each subject. Methyltrienolone binding on cultured genital skin fibroblasts of B2 suggested moderate impairment of the ligand-binding domain [maximal binding capacity, 38.2 fmol/mg protein (normal); K_d, 0.21 nmol/L; normal range, 0.03–0.13 nmol/L]. In *trans*-activation assays, the mutant 712F-AR showed considerable deficiency at low concentrations of testosterone (0.01–0.1 nmol/L) or dihydrotestosterone (0.01 nmol/L). Remarkably, this could be fully neutralized by testosterone concentrations greater than 1.0 nmol/L. Hence, the 712F-AR could switch its function from subnormal to normal within the physiological concentration range of testosterone. This was reflected by an excellent response to testosterone therapy in B1, B2, and the uncle. Taking into account the well documented individual and time-dependent variation in testosterone concentration in early fetal development, our observations clearly illustrate the potential impact of varying ligand concentrations for distinct cases of phenotypic variability in androgen insensitivity syndrome.

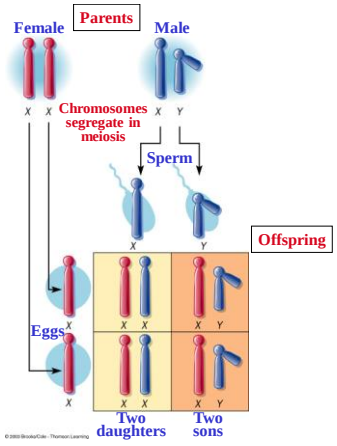
Normal Karyotype of Human Chromosomes

- **What are homologous chromosomes?**
- **What gender?**
- **Sex vs. autosomal chromosomes?**



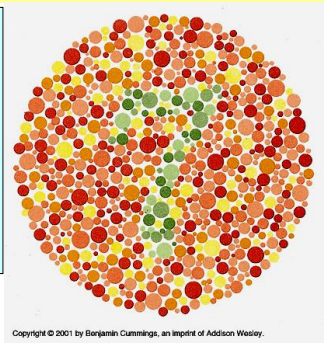
Gender Determination in Humans

- Gender is determined by the presence of the SRY-gene on the Y-chromosome
- SRY gene is turned on around the 7th week of gestation.
- XY females = Androgen Insensitivity Syndrome
  - X-linked recessive
  - Androgen receptor doesn't recognize testosterone
  - Consequences?



Test for Red-Green Colorblindness

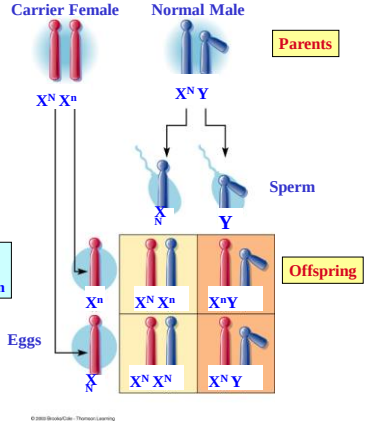
- Colorblindness is caused by a malfunction of light-sensitive cells in the retina of the eyes
- What number do your see?
- Like all X-linked recessive traits, colorblindness is very rare in women. Why??



Colorblindness: X-linked Recessive Inheritance

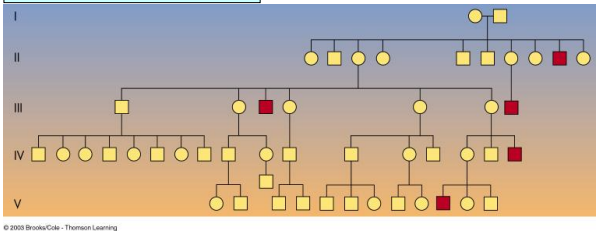
Carrier Female $X^N X^n$ × Normal Male $X^N Y$

2 Healthy daughters + 1 healthy son + 1 color blind son



Pedigree Duchenne Muscular Dystrophy

- Mode of inheritance?
- Autosomal or Sex-linked?
- Genotypes?

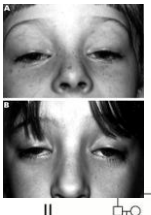


Sample Problem

Mary's paternal and maternal grandfathers are both colorblind. There is no evidence of colorblindness in either grandmother's family histories.

a. What is Mary's genotype? Phenotype?

b. What are the chances that Mary's brother is colorblind?



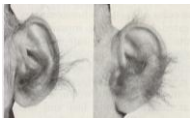
X-Linked Dominant Example

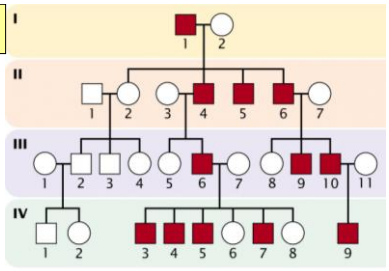
Congenital Bilateral Ptosis: **Droopy Eyelids**

Locus: Xq24-Xq27.1

Hairy ears, Y-LINKED?

HYPERTRICHOSIS PINNAE AURIS



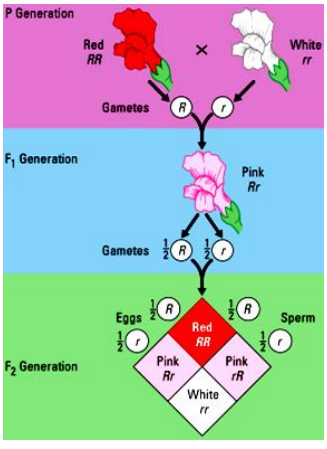


C.Stern *et al.* (1964) Am J Hum Gen. **16**:467.

Incomplete Dominance

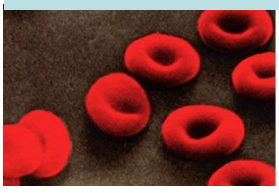
- The dominant allele is incompletely dominant over the recessive allele
- Phenotype of heterozygous individuals is in-between that of the homozygous dominant and homozygous recessive phenotypes
  - » E.g. Snapdragons, sickle cell anemia

Incomplete Dominance in Snapdragons



Sickle Cell Anemia—an example of incomplete dominance

- Uncommon in U.S.A. (~1 in 60,000)
- Common in West Africa (~1 in 50) and African Americans (~1 in 400)
  - Lethal in the homozygous recessive condition
  - What is the adaptive value of heterozygous condition in West Africa?



Normal Red Blood Cells

Sickled R.B.C.'s clump together and clog blood vessels

P: Male with Sickle Cell Trait (Hh) x Female with Sickle Cell Trait (Hh)

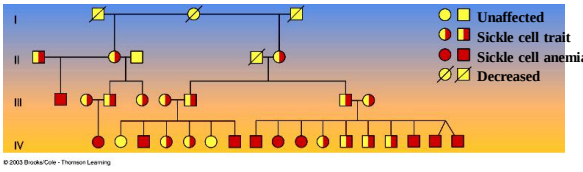
Gametes of Male with sickle cell trait

H = healthy hemoglobin allele
h = sickle cell allele

Gametes of Female with sickle cell trait

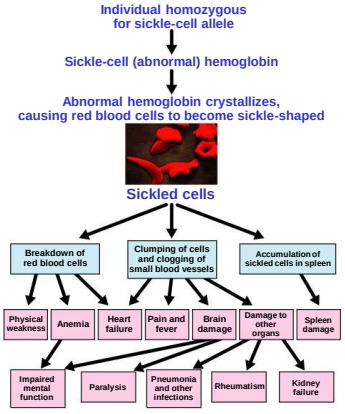
	H	h
H	HH = normal RBC Non-sickling	Hh = RBC sickle when levels are O_2 Low Sickle cell trait
h	Hh = RBC sickle when levels are O_2 Low Sickle cell trait	hh = Sickled RBC's Sickle cell anemia

Sickle Anemia Pedigree: An example of incomplete dominance



Pleiotrophy

- **The impact of a single gene on more than one characteristic**
- **Examples of Pleiotrophy:**
  - **Sickle-Cell Anemia**
  - **Gaucher Disease**
  - **Cystic Fibrosis**



Codominance: Blood Types

Blood Type (Phenotype)	Surface Molecule on R.B.C.	Possible Genotypes
A	Red blood cell	I^AI^A or I^Ai
B		I^BI^B or I^Bi
AB		I^AI^B
O	(Neither A nor B)	*ii*

- **Alleles**
  - **I^A = Allele for Type A**
  - **I^B = Allele for Type B**
  - ***i*** **= Allele for Type O**
- **I^A is dominant to *i***
- **I^B is dominant to *i***
- **I^A and I^B are codominant**
- **What do these alleles code for?**
- **How many alleles can you inherit?**

ABO and Rh Blood Type Frequencies in the United States

(Source: Stanford School of Medicine: Blood Center)

ABO Type	Rh Type	How Many Have It	
O	positive	37.4%	44%
O	negative	6.6%	
A	positive	35.7%	42%
A	negative	6.3%	
B	positive	8.5%	10%
B	negative	1.5%	
AB	positive	3.4%	4%
AB	negative	.6%	

Blood Group (Phenotype)	Genotypes	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	Anti-B				
B	I^BI^B or I^Bi	Anti-A				
AB	I^AI^B	—				

Blood Types: Sample Problem #1

A couple has the type A and Type B, respectively. Is it possible for them to have a child with the following blood types? If so, what is the genotype of each parent?

a. Type O
b. Type A
c. Type B
d. Type AB

Blood Types: Sample Problem #2

A couple has the type A and Type AB, respectively. Is it possible for them to have a child with the following blood types? If so, what is the genotype of each parent?

a. Type O
b. Type A
c. Type B
d. Type AB

Rhesus Factor—a RBC surface molecule

- **Rh factor is inherited independently from the ABO system**
- **Rh positive people:**
  - » **R.B.C's have the Rhesus factor surface molecule**
- **Rh Negative people:**
  - » **R.B.C's w/o the Rhesus factor surface molecule**
- **Alleles**
  - » **R = Rh factor**
  - » **r = no Rh factor**

Phenotype	Possible Genotypes
Rh + (Rh positive)	RR or Rr
Rh- (Rh negative)	rr

Blood Types: Sample Problem #3

A couple has the type A+ and Type AB+, respectively. What are the chances of the couple having a child with the following phenotypes.

a. Type O+ **b. Type O-**

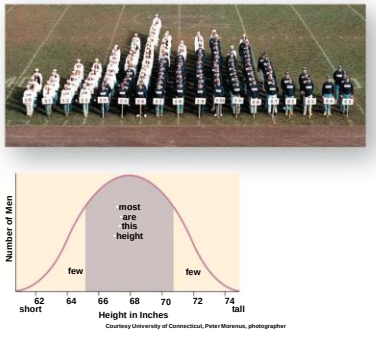
c. Type A+ **d. Type A-**

e. Type B+ **f. Type B-**

g. Type AB+ **h. Type AB-**

- Polygenic inheritance
  - Trait is governed by 2 or more sets of alleles.
  - Each dominant allele has a quantitative effect on phenotype and effects are additive.
  - Result in continuous variation – bell-shaped curve
  - Multifactorial traits – polygenic traits subject to environmental effects
    - Cleft lip, diabetes, schizophrenia, allergies, cancer
    - Due to combined action of many genes plus environmental influences

Figure 10.11 Height in humans, a polygenic trait



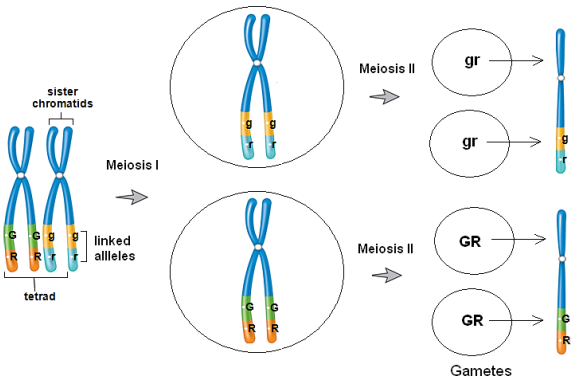
- **Environment and the phenotype**
  - Relative importance of each can vary.
  - Temperature can effect coat color.
    - Rabbits homozygous for *ch* have black fur where the skin temperature is low.
    - Enzyme encoded by gene is active only at low temperatures.

Figure 10.12 Coat color in Himalayan rabbits

10.4 Inheritance of Linked Genes

- Some fruit fly crosses violated the law of independent assortment ☹
  - Offspring simply resembled one of the parents
- **Gene linkage**
  - 2 or more traits on same chromosome
  - Traits on same chromosome do NOT segregate independently

Figure 10.16 Linked alleles Usually Stay Together



- Occasionally crossing-over produces new combinations.
  - Nonsister chromatids exchange genes.
  - Recombinant gametes have a new combination of alleles.

Crossing-over results in recombination of alleles

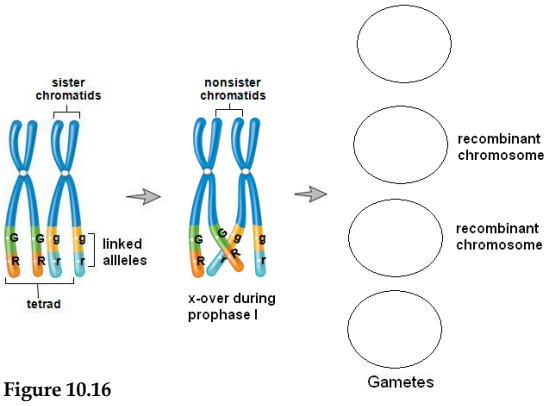
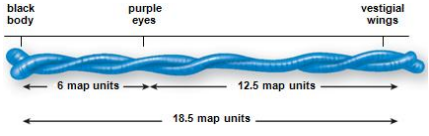


Figure 10.16

- Distance between genes
  - The closer 2 genes are on a chromosome, the less likely they are to cross-over.
  - You can use the percentage of recombinant phenotypes to determine the distance between genes.
  - 1% crossing-over = 1 map unit.
  - In a black-body and purple-eye cross, 6% of offspring are recombinant = genes are 6 map units apart.
  - Results can make a chromosome map.



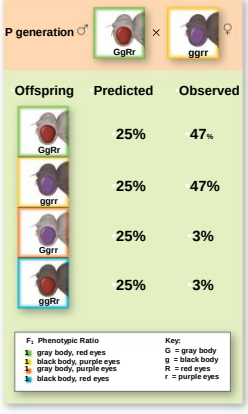


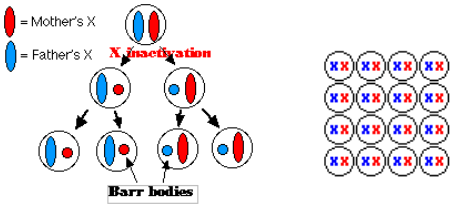
Figure 10.17 Linked alleles do not assort independently

Why are Calico Cats females, not males?

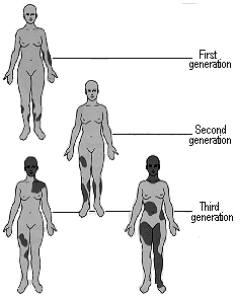
Gene for white is on an autosomal chromosome and unrelated to the alleles on the X-chromosome

- **Genes for fur color in cats on the X chromosome:**
  - X^B = Black
  - X^b = Yellowish-orange
- **Calico cats are heterozygous: X^BX^b**
  - » Why calico and not black?
  - » Due to X-inactivation—what's that?

X-inactivation in females



Anhidrotic Ectodermal Dysplasia



Calico Cats: X^BX^b

- **Classic example of X-inactivation**
- **Different fur producing cells randomly inactivate one of the X chromosomes**
  - **Happens during embryonic development**
- **Gives the patchy calico fur pattern:**
  - **Black patches have cells with the X^B chromosome active**
  - **Yellow patches have cells with the X^b chromosome active**

Genotype	X^BX^B	X^BX^b	X^bX^b	X^BY	X^bY
Phenotype					