Meiosis, Genes, and Alleles

- Genetics is the study of inheritance.
- Genetics can predict how genes may be passed on to future generations.
  - Requires an understanding of
    - How genes are organized on chromosomes
    - How chromosomes are passed on during meiosis

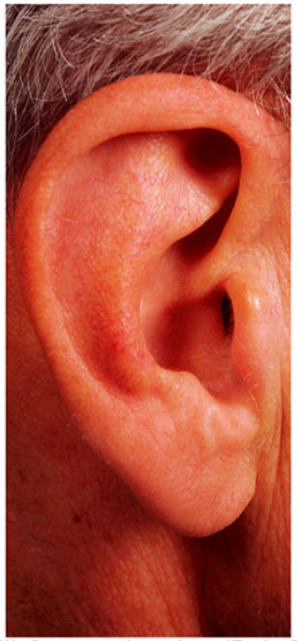
Different Ways to Study Genes

- A gene is…
  - A piece of DNA that has the necessary information to code for a protein and regulate its expression
  - Found on a chromosome
  - Related to a characteristic of an organism
    - These characteristics result from the work of a particular protein.
      - Eye color
      - Flower color
      - Pea shape

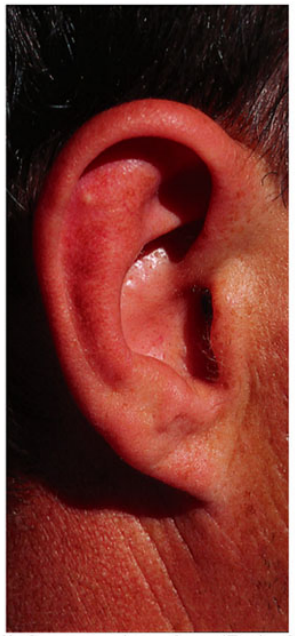
What is an allele?

- One particular gene may exist in multiple forms.
- An allele is
  - A specific version of a gene
- Example: Earlobe shaped gene
  - There are two different alleles for this gene.
    - Attached earlobe
    - Free earlobe
- Different alleles code for different forms of the same protein.
  - The different forms of the protein function differently.
    - Result in different characteristics

What is an allele?



© The McGraw-Hill Companies, Inc./Bob Coyle, photographer



© The McGraw-Hill Companies, Inc./Bob Coyle, photographer

Genomes and Meiosis

- The sum total of an organism's genes is called its *genome.*
  - In sexually reproducing organisms, the genome is *diploid*.
    - This means that they have two copies of every gene.
    - The copies may not be identical, so one individual could have two different alleles.
  - Single-celled organisms and sex-cells are *haploid.*
    - They only have one copy of each gene.
    - They only have one allele.

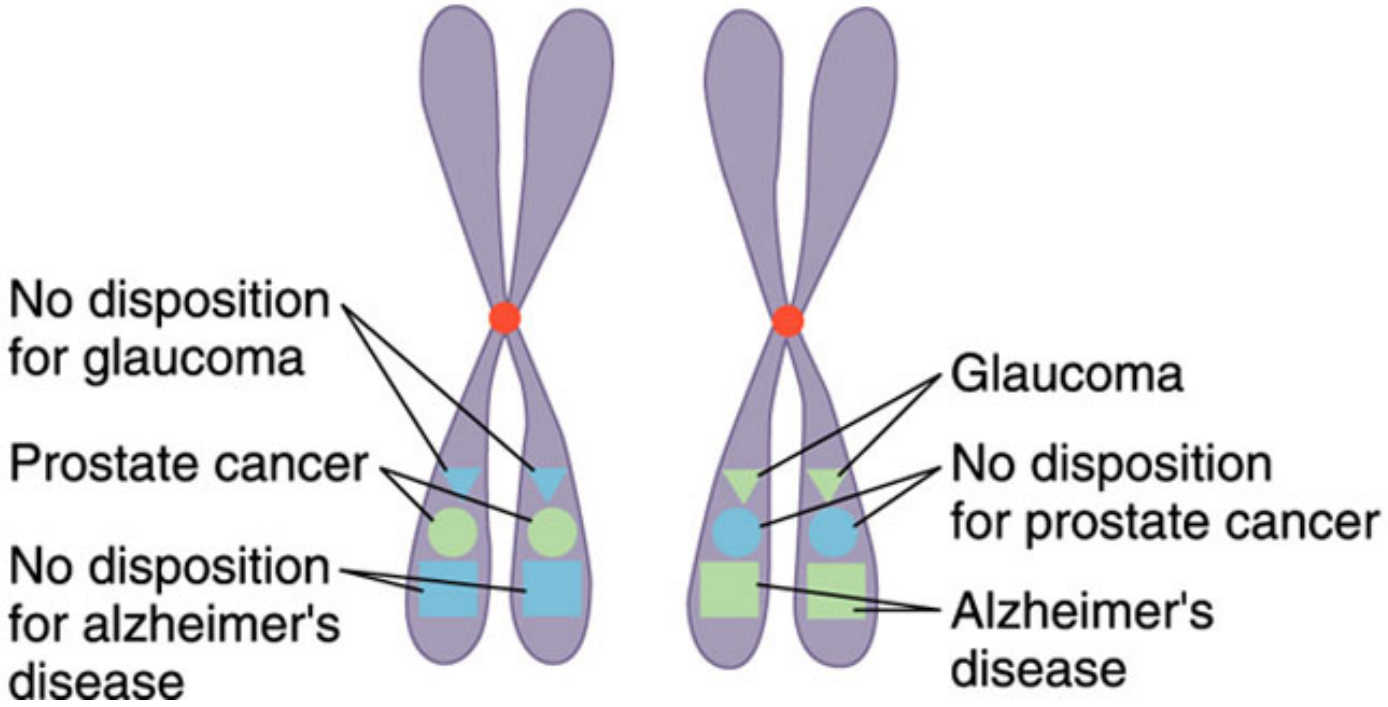
Genomes and Meiosis

- Sex cells are sperm and egg.
  - Sperm and egg only receive one set of that individual's genes.
  - When haploid egg joins with haploid sperm (fertilization), a diploid zygote results.
  - The zygote receives half of its genome from the sperm and half of its genome from the egg.
    - Has a unique set of genes, different from the parents

Genomes and Meiosis

- Meiosis is the process by which egg and sperm are made.
  - Homologous chromosomes can carry different alleles.
  - When the homologous chromosomes separate during meiosis, the alleles are delivered to different sex cells.

Homologous Chromosomes



Fundamentals of Genetics

- Three questions allow us to predict how a trait will be inherited:
  - What alleles do the parents have for that trait?
  - What alleles will be present in the gametes that the parents produce?
  - What is the likelihood that gametes with specific combinations of alleles will be fertilized?

Phenotype vs. Genotype

- Diploid organisms have two copies of every gene.
  - This means that one individual can have two different versions of a gene.
  - The term allele is used to identify different versions of a gene.
- *Genotype* describes the combination of alleles present in the organism's cells.
- *Phenotype* describes the organism's appearance.
  - This is a result of its genotype.

Example: Earlobe Shape

- Phenotypes: free or attached
- Genotypes:
  - EE (two alleles for free earlobes)
    - Earlobes will be free
  - ee (two alleles for attached earlobes)
    - Earlobes will be attached
  - Ee (one allele for free and one allele for attached)
    - Earlobes will be free
- The free earlobe allele is dominant.
  - It out-performs the attached earlobe allele.
- The attached earlobe allele is recessive.
  - Masked by the dominant allele when present together
  - Only expressed when two copies are present

Homozygous vs. Heterozygous

- Homozygous
  - Two copies of the same allele
    - EE is homozygous dominant.
    - ee is homozygous recessive.
- Heterozygous
  - Two different alleles
  - Ee

Predicting Genotype from Phenotype

- An individual with the dominant phenotype
  - Could be homozygous dominant
  - Could be heterozygous
  - A person with free earlobes could be
    - EE or Ee
- An individual with the recessive phenotype
  - Is always homozygous recessive
  - A person with attached earlobes is ee.

Predicting Gametes from Meiosis

- The Law of Segregation:
  - Alleles will separate during meiosis.
    - Each gamete will receive one allele.
- An EE individual will make gametes that have E.
- An ee individual will make gametes that have e.
- An Ee individual will make gametes that have either E or e.

Predicting Offspring from Fertilization

- Fertilization is the process of two haploid sex cells joining to form a diploid zygote.
  - The genotype of the offspring will be determined by the alleles carried by the gametes.
- A genetic cross is a planned mating between two organisms.
  - The outcome of a given cross is predicted by a Punnett Square.
- Single-factor crosses track the inheritance of one trait.
  - Also called monohybrid crosses
- Double-factor crosses track the inheritance of two traits.
  - Also called dihybrid crosses

Punnett Square

Cross: *Ee* × *Ee*

Gametes Possible: 50% *E* and 50% *e* 50% *E* and 50% *e*

Punnett Square:

	E	*e*
E	*EE*	*Ee*
e	*Ee*	*ee*

Probability vs. Possibility

- Probability is the mathematical chance that an event will happen.
  - Expressed as a percent, or a fraction
    - Probability = the # of events that can produce a given outcome/the total # of possible outcomes.
- The probability of two or more events occurring simultaneously is the product of their individual probabilities.
- Possibility states that an event *can* happen; probability states how likely the event is to happen.

The First Geneticist: Gregor Mendel

- Mendel was a monk who was the first to describe the basic patterns of inheritance.
  - Studied inheritance in garden pea plants
  - Studied several different phenotypes
  - Identified the concepts of dominance and recessiveness
  - Didn't know about genes or chromosomes
  - Identified patterns by mathematical analysis of the data

Mendel's Experiment

- Parental (P) generation
  - A pure-breeding purple-flowered plant mated with a pure-breeding white-flowered plant.
  - CC x cc
- First filial generation (F1)
  - All offspring had purple flowers (Cc).
  - They were allowed to self-pollinate.
  - Cc x Cc
- Second filial generation (F2)
  - ¾ of the offspring were purple
  - ¼ of the offspring were white
  - 3:1 ratio, purple: white
- Mendel saw this pattern with any of the traits he studied.

Dominant and Recessive Traits in Pea Plants

TABLE 10.1

Dominant and Recessive Traits in Pea Plants

Gene	Dominant Allele Phenotype	Recessive Allele Phenotype
Plant height	Tall	Dwarf
Pod shape	Full	Constricted
Pod color	Green	Yellow
Seed surface texture	Round	Wrinkled
Seed color	Yellow	Green
Flower color	Purple	White

Mendel's Conclusions

- Organisms have two pieces of genetic information for each trait.
  - We know these as alleles.
- The Law of Dominance
  - Some alleles mask other alleles.
- Gametes fertilize randomly.
- The Law of Segregation
  - Alleles separate into gametes during meiosis.

Solving Genetics Problems: Single-Factor Crosses

- The pod color of some pea plants is inherited so that green pods are dominant to yellow pods.
- A pea plant that is heterozygous for green pods is crossed to a pea plant that produces yellow pods.
- What proportion of the offspring will have green pods?

Step I: Make a Gene Key

Gene Key
Gene or Condition: pod color

Allele Symbols	Possible Genotypes	Phenotype
G = green	GG	Green
	Gg	Green
g = yellow	gg	Yellow

Step 2: Identify Information in the Problem

- A green plant is crossed with a yellow plant.
- The green pod plant is heterozygous.
  - Gg
- The yellow pod plant is homozygous.
  - gg
- The cross is Gg x gg.

Step 3: Determine Possible Gametes from Each Parent

- Heterozygous green pod parent (Gg)
  - Could make gametes with G or g
- Homozygous yellow pod parent (gg)
  - Could make gametes with g

Step 4: Create a Punnett Square

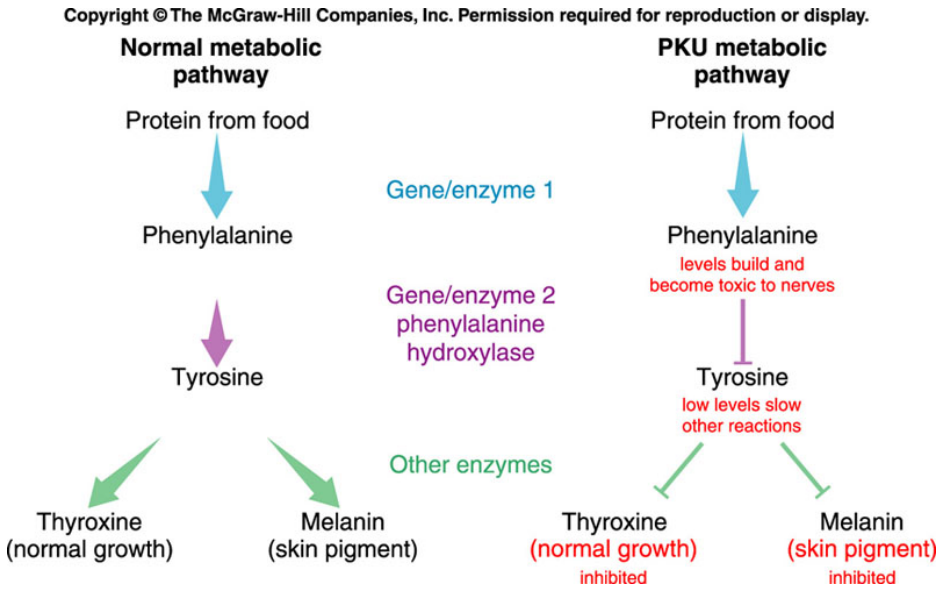
- Put the gametes from one parent on one side.
- Put the gametes from the other parent on the other side.
- Simulate random fertilization by crossing the possible gametes.
  - This will determine offspring phenotypes.

Step 5: Determine Offspring Phenotypes and Calculate Probability

- Use the gene key to determine the phenotype of the offspring you predicted.
- Revisit the question to calculate the answer to the question.
  - What proportion of offspring will produce green pods?
    - The answer is 50%.

Cross #2: PKU

- The normal condition is to convert phenylalanine to tyrosine. It is dominant over the condition for PKU.
- If both parents are heterozygous for PKU, what is the probability that they will have
  - A child that is normal?
  - A child with PKU?



Solution Pathway

TABLE 10.3

Solution Pathway

Steps in Information Flow	The Problem
Parental phenotypes	Father × Mother Normal × Normal
Parental genotypes	*Pp* × *Pp*
Possible sex cells	*P* *P* *p* *p*
Offspring genotype	*P* | *p* *P* | *PP* | *Pp* *P* | *Pp* | *pp*
Offspring phenotype	Normal 25% *PP* 50% *Pp* 75% Total Phenylketonuria 25% Total

Double-factor Crosses

- Dihybrid crosses track the inheritance of two traits.
- Mendel used dihybrid crosses to identify the law of independent assortment.
  - States that alleles of one character separate independently of alleles of another character
  - Only true when the genes for the two characters are on different chromosomes

Solving Double-factor Crosses

- When solving a double-factor cross, you must obey the law of segregation and the law of independent assortment.
  - Each gamete must receive *only one* copy of each gene.
  - All combinations of alleles for A and B must be considered.
- Consider an individual whose genotype is AaBb.
  - Gametes could receive AB, Ab, aB or ab.

A Sample Double-factor Cross

- In humans the allele for free earlobes is dominant over the allele for attached earlobes.
- The allele for dark hair dominates the allele for light hair.
- If both parents are heterozygous for earlobe shape and hair color, what types of offspring can they produce, and what is the probability for each type?

Solving the Double-factor Cross

- Start by creating a gene key for each gene.

Gene Key
Gene or Condition: earlobe type

Allele Symbols	Possible Genotypes	Phenotype
E = free	EE	Free earlobes
	Ee	Free earlobes
e = attached	ee	Attached earlobes

Solving the Double-factor Cross

TABLE 10.4

Solution Pathway

Steps in Information Flow	The Problem
Parental phenotypes	Father × Mother Free earlobes Free earlobes Dark hair Dark hair
Parental genotypes	The problem states that both parents are heterozygous for both characteristics. *EeHh* × *EeHh*
Possible sex cells	Notice that the Law of Independent Assortment has been added as a skill that should be used for a double-factor cross. Both parents have the same genotypes, so they each produce the same types of gametes. *EH* *EH* *Eh* *Eh* *eH* *eH* *eh* *eh*

Solving the Double-factor Cross

Offspring genotype

Set up a Punnett square to show the possible fertilization events.

	EH	*Eh*	*eH*	*eh*
EH	*EEHH*	*EEHh*	*EeHH*	*EeHh*
Eh	*EEHh*	*EEhh*	*EeHh*	*Eehh*
eH	*EeHH*	*EeHh*	*eeHH*	*eeHh*
eh	*EeHh*	*Eehh*	*eeHh*	*eehh*

Offspring phenotype

Count up the different genotypes and then combine them by similar phenotype using the information in the Gene Key. The Punnett square is 4 × 4, so each box counts for 1/16 of the possible offspring.

Free Earlobes and Dark Hair	Free Earlobes and Light Hair	Attached Earlobes and Dark Hair	Attached Earlobes and Light Hair
1/16—*EEHH*			
2/16—*EEHh*			
2/16—*EeHH*	1/16—*EEhh*	1/16—*eeHH*	
4/16—*EeHh*	2/16—*Eehh*	2/16—*eeHh*	1/16—*eehh*
9/16	3/16	3/16	1/16

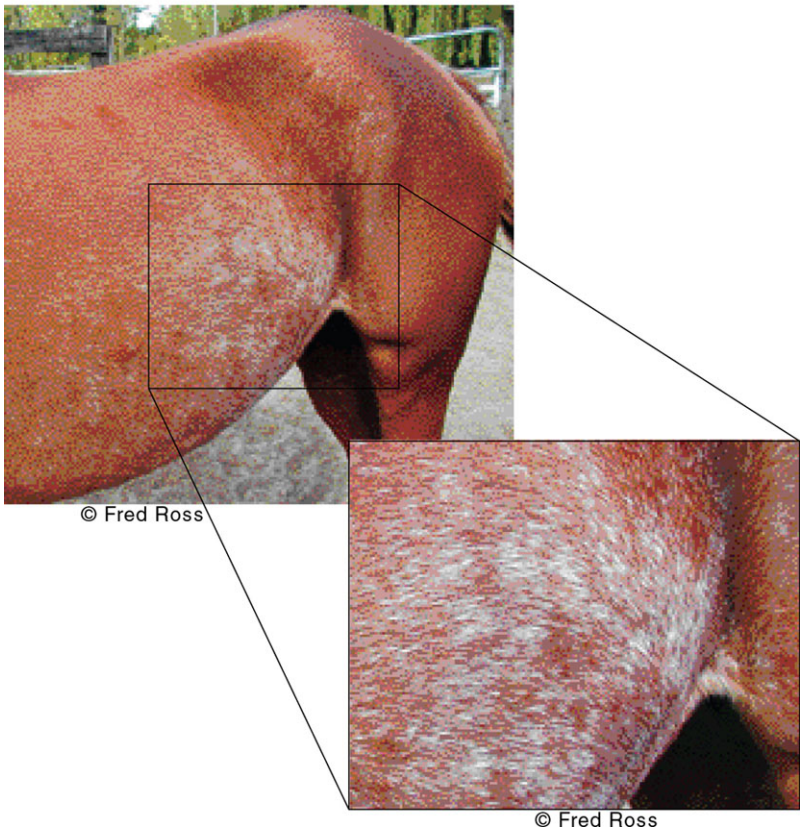
Modified Mendelian Patterns

- Some alleles have consistent dominant/ recessive patterns like Mendel observed.
- However, many traits are not inherited following these patterns.
- Several other types of inheritance patterns exist.

Codominance

- Some alleles are codominant.
  - Both phenotypes are expressed together in a heterozygote.
  - This will result in three phenotypes.
  - Horse color
    - $D^R D^R$ is chestnut color
    - $D^R D^W$ is white color
    - $D^W D^W$ is palomino-colored (chestnut with white mane and tail)

Codominance



Incomplete Dominance

- Occurs when the phenotype of the heterozygote is intermediate between the two homozygotes
  - Appears as if the heterozygotes are blends of the homozygotes
- Snapdragons
  - F^wF^w=white flower
  - F^rF^r=red flower
  - F^wF^r=pink flower

Incomplete Dominance

© John Cunningham/Visuals Unlimited

Sample Problem: Incomplete Dominance

- If a pink snapdragon is crossed with a white snapdragon, what phenotypes can result?
- What is the probability of each phenotype?

Solution Pathway: Incomplete Dominance

TABLE 10.5

Solution Pathway

Steps in Information Flow	The Problem
Parental phenotypes	Pink × White
Parental genotypes	$F^RF^W \times F^WF^W$
Possible sex cells	F^R F^W F^W
Offspring genotype	<table><tr><td></td><td>F^W</td></tr><tr><td>F^R</td><td>F^RF^W</td></tr><tr><td>F^W</td><td>F^WF^W</td></tr></table>
Offspring phenotype	50% pink 50% white

Multiple Alleles

- Some traits have more than two possible alleles for a single trait.
- Each person can only have two alleles for a given trait because diploid organisms have only 2 copies of each gene.
- Example: ABO blood types
  - 3 alleles for blood type antigens on red blood cells
    - I^A = blood type A antigens
    - I^B = blood type B antigens
    - i = blood type O, neither type A or type B antigens
  - Six possible genotypes; each individual can only have two alleles
    - I^AI^A, I^Ai = Type A blood
    - I^BI^B, I^Bi = Type B blood
    - I^BI^A = Type AB blood
    - Ii = Type O blood

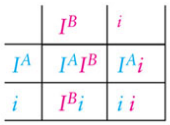
Sample Problem: Multiple Alleles

- Allele A and allele B are codominant.
- Allele A and allele B are both dominant to O.
- A male heterozygous with blood type A and a female heterozygous with blood type B have a child.
- What are the possible phenotypes of their offspring?

Solution Pathway: Multiple Alleles

TABLE 10.6

Solution Pathway

Steps in Information Flow	The Problem
Parental phenotypes	Type A × Type B
Parental genotypes	$I^Ai \times I^Bi$
Possible sex cells	I^A I^B i i
Offspring genotype	
Offspring phenotype	25% Type AB (I^AI^B) 25% Type A (I^Ai) 25% Type B (I^Bi) 25% Type O (ii)

Polygenic Inheritance

- Some characteristics are determined by the interaction of several genes.
- A number of different pairs of alleles combine their efforts to determine a characteristic.
- Polygenic inheritance is common with characteristics that show great variety within the population.
  - Height, eye color, intelligence, etc.

Skin Color is a Polygenic Trait

- Skin color is governed by at least 3 different genes.
  - Therefore, a wide variety of skin colors exist in the human population.

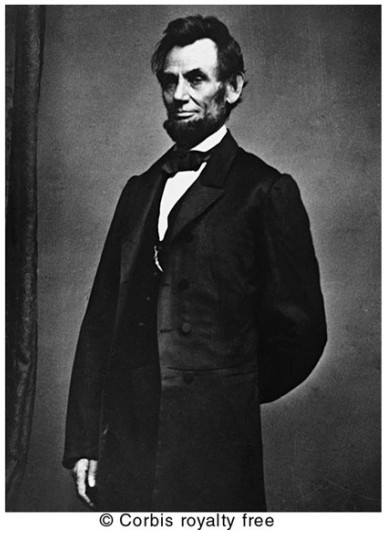
Gene 1	d^1d^1	d^1D^1	d^1D^1	D^1D^1	D^1d^1	D^1d^1	D^1D^1
Gene 2	d^2d^2	d^2d^2	d^2D^2	D^2d^2	D^2d^2	D^2D^2	D^2D^2
Gene 3	d^3d^3	d^3d^3	d^3d^3	d^3d^3	D^3D^3	D^3D^3	D^3D^3
Total number of dark-skin genes	0	1	2	3	4	5	6
	Very light			Medium			Very dark
# of light "d" alleles	6	5	4	3	2	1	0
# of dark "D" alleles	0	1	2	3	4	5	6

Pleiotropy

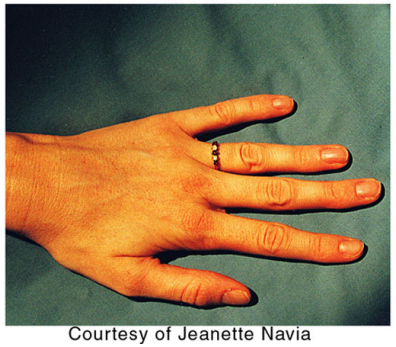
- Some genes affect a variety of phenotypes.
  - These genes are called pleiotropic.
- The disease PKU results from a mutation in one gene.
  - The one defective protein leads to several phenotypes.
    - Mental retardation, abnormal growth, pale skin pigmentation

Marfan's Syndrome is Pleiotropic

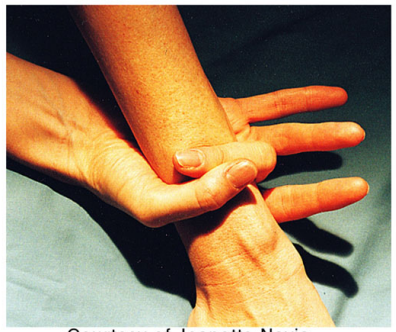
Courtesy of Jeanette Navia



© Corbis royalty free



Courtesy of Jeanette Navia



Courtesy of Jeanette Navia

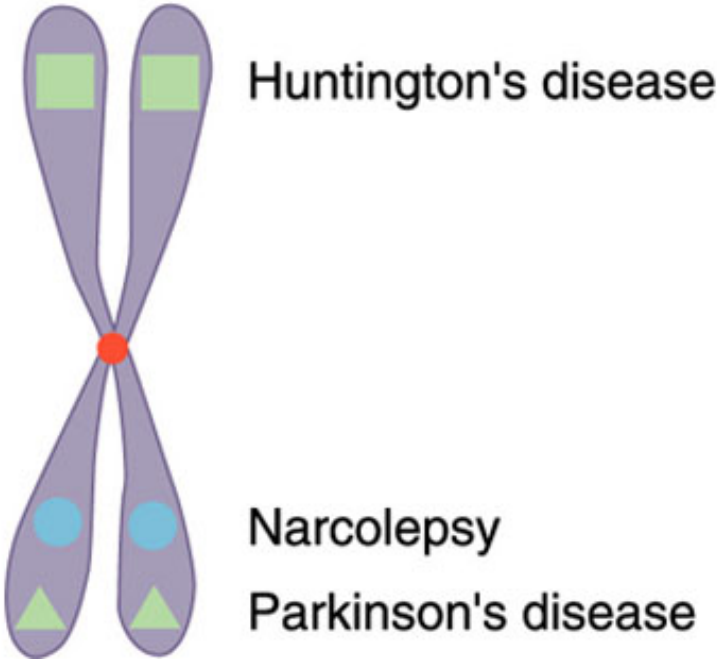
Linkage

- Genes that are on the same chromosome are linked.
- Linked genes are inherited together more often than would be predicted by probability.
- All of the genes on a given chromosome represent a *linkage group.*
  - All of the genes in a linkage group will be inherited together.
  - Crossing-over can separate linked genes and mix allele combinations.
    - The closer genes are to one another on a chromosome, the less likely they will be separated by crossing-over, and the more likely they will be inherited together.

Autosomal Linkage

- Autosomes are the chromosomes that are not involved in sex determination.
- Of the 23 pairs of human chromosomes, #1-22 are autosomes.
  - Genes on the same autosomal chromosome are autosomally linked.
- #23 are sex chromosomes.
  - Called X and Y

Linked Genes are Found on the Same Chromosome



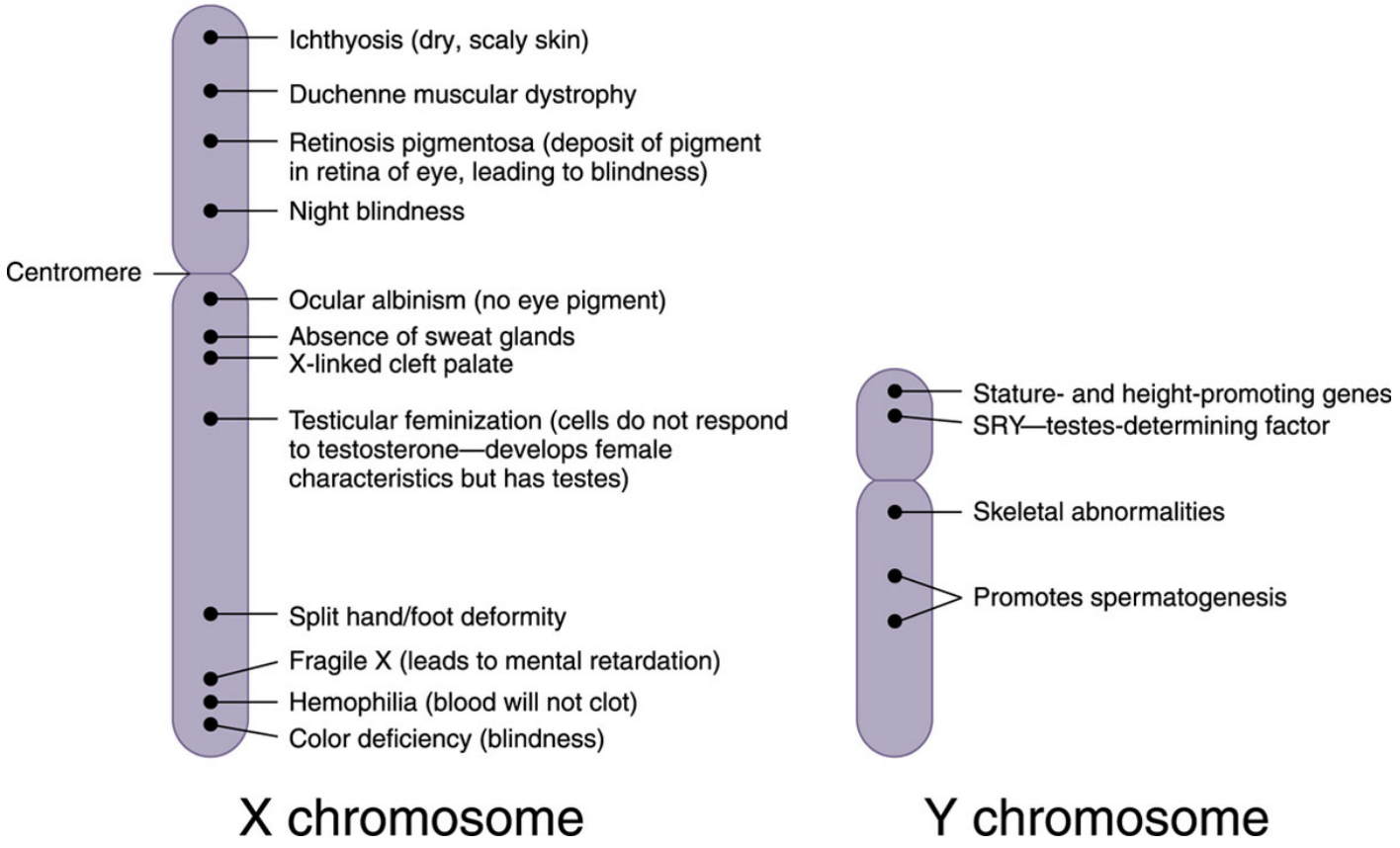
Sex Determination

- The sex chromosomes, X and Y, are a homologous pair.
  - This pair is unique because X and Y carry different sets of genes.
  - The Y chromosome has genes that determine maleness.
  - The X chromosome has a variety of genes on it, many of which are not involved in gender determination.

Sex Linkage

- Genes on the X or Y chromosomes are called sex-linked.
  - Genes on the X chromosome are called X-linked.
    - Males only have one X chromosome, so one copy of a recessive allele will result in the recessive phenotype in men.
    - Women have two copies of X, so they can be heterozygous or carriers of a recessive trait without showing the phenotype.
    - Hemophilia, color-blindness, muscular dystrophy

Sex Chromosomes



X-linked Inheritance Patterns

- In humans, the allele for normal color vision is dominant and the allele for color deficiency is recessive.
- Both alleles are X-linked.
- People who cannot detect the difference between certain colors such as green and red are described as having color-deficient vision.
- A male who has normal color vision mates with a female who is heterozygous for normal color vision.
- What type of children can they have in terms of these traits?
- What is the probability for each type?

Solution Pathway: X-linked Inheritance

TABLE 10.7

Solution Pathway

Steps in Information Flow	The Problem
Parental phenotypes	Father: normal vision × Mother heterozygote for color vision
Parental genotypes	$X^BY \times X^BX^b$
Possible sex cells	X^B X^B Y X^b
Offspring genotype	X^B | X^b X^B | X^BX^B | X^BX^b Y | X^BY | X^bY
Offspring phenotype	50% normal females (½ of these are carriers) 25% normal males 25% color-deficient males

Other Influences on Phenotype

- Variable expressivity
  - Some dominant traits are not expressed equally in all individuals with the trait.
  - Polydactylism
- Environmental factors
  - Can influence the expression of a trait
  - Freckles and sunlight
  - Diabetes and diet

© Renee Lynn/Photo Researchers

Courtesy of Mary Drapeau

Courtesy of Mary Drapeau