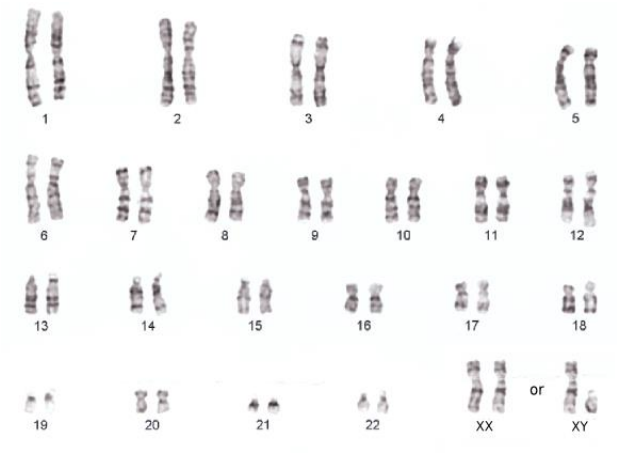
Chapter 14.1 & 14.2 Outline

Human Chromosomes



Humans have 46 chromosomes = 23 pairs of homologous chromosomes (2n=23). A *karyotype* image is photographed when a cell is in an early stage of mitosis because that is when chromatin is condensed and chromosomes are visible.

A human female has a pair of X chromosomes for the 23rd homologous pair. A human male is considered hemizygous, with XY instead of XX. The Y chromosome is much smaller than the X chromosome and mostly carries genes for male traits.

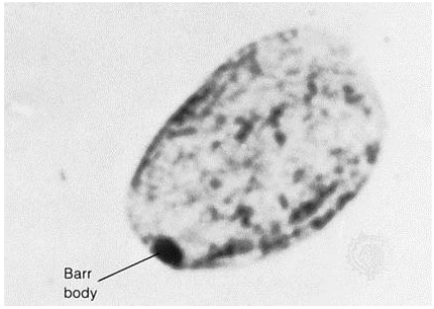
Chromosomes 1-22 are known as *autosomes*. Chromosome pair 23 is the *sex chromosomes*.

Genes carried on the X chromosome have a special pattern of inheritance. They are *sex-linked*. Recessive diseases or traits on the X chromosome show up much more often in men than in women because females have a second X chromosome that can mask the recessive allele with a dominant allele.

Punnett Square for Color Blindness

	X^B	X^b
X^B	X^BX^B	X^BX^b
Y	X^BY	X^bY

B = Normal
b = Color Blind

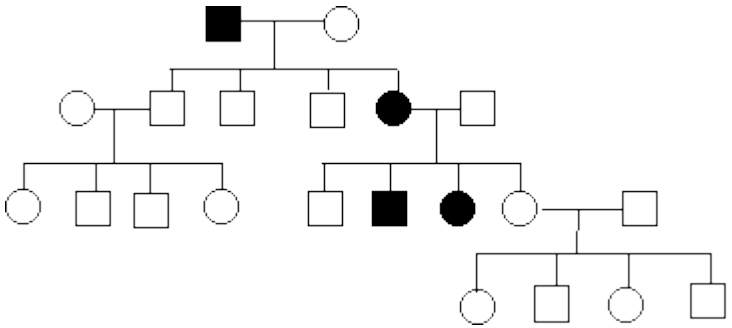


Strangely enough, in female cells one of the X chromosomes is inactivated, or turned off. X chromosome inactivation maintains the right amount of gene dosage. In female cells the inactivated X chromosome is a small mass offset in the nucleus, and is known as a Barr-body.

Human blood types exhibit both *codominance* and *multiple alleles*. Blood types are distinguished by antigens (glycoproteins) on the surface of the cell, I^A and I^B are codominant; both are fully expressed in a heterozygous individual. *i* is recessive to both I^A and I^B; the *i* does not produce a surface marker on the red blood cell.

Blood Type	Genotype		Can Receive Blood From:
A	i^Ai i^Ai^A	AA AO	A or O
B	i^Bi i^Bi^B	BB BO	B or O
AB	i^Ai^B	AB	A, B, AB, O
O	ii	OO	O

Human Pedigrees are similar to a family tree, but with the oldest (often deceased) individuals shown at the top of the diagram, and the youngest below. Females are shown with a circle, and males are squares. An individual who has a disease or trait is shaded. You can determine if an allele is dominant, recessive, autosomal or sex linked by analyzing the pattern of inheritance in a pedigree chart.



Genetic Disorders

Cystic fibrosis—caused by a deletion of three consecutive nucleotide bases. The *Cf* allele shows an autosomal recessive pattern of inheritance.

Sickle Cell disease—caused by a nucleotide substitution, which changes the shape of the hemoglobin protein. Sickle cell disease is autosomal recessive, but heterozygous individuals are resistance to the malaria parasite.

Huntington's disease—caused by duplication of a stretch of CAG nucleotides. The disease is autosomal dominant and usually fatal by age 40.

Down's syndrome—caused by a trisomy of chromosome 21. Because of nondisjunction during meiosis (failure of homologous chromosomes to separate in anaphase I or II), the zygote inherited 3 copies of chromosome 21 instead of 2.

1. A color blind father and heterozygous mother have a child. Draw the Punnett square.

    a. What is the chance that they have a color blind daughter?

    b. What is the chance that they have a color blind son?

2. A father with normal vision and a heterozygous mother have a child. Draw the Punnett square.

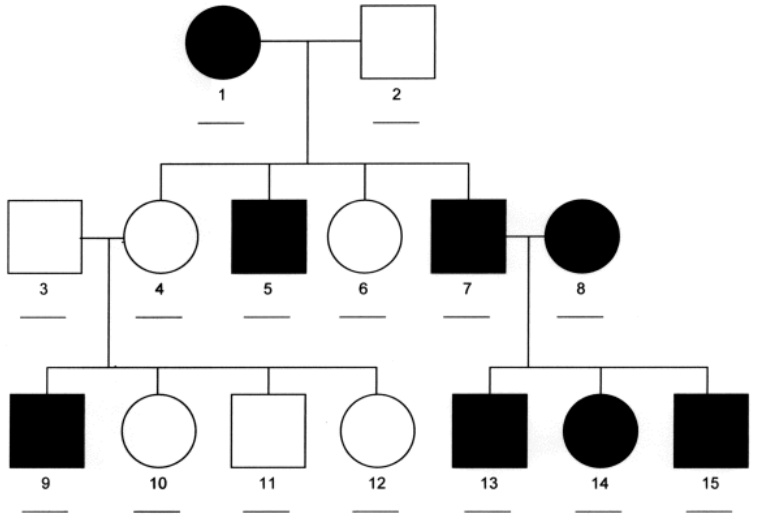
    a. What is the chance that they have a color blind daughter?

    b. What is the chance that they have a color blind son?

3. A woman with type *A* blood is claiming that a man with type *AB* blood is the father of her child, who is also type *AB*. Could this man be the father? Show the possible crosses; remember the woman can have *AO* or *AA* genotypes.

4. A man with type *AB* blood is married to a woman with type *O* blood. They have two natural children, and one adopted child. The children's blood types are: *A*, *B*, and *O*. Which child was adopted?

5. Determine whether this pedigree shows a dominant/recessive trait, and whether it is autosomal or sex linked. Include the genotype of every individual.



6. Coat color in Labrador retrievers shows epistasis. The *B* allele produces a black pigment; the *b* allele produces a brown pigment; the *E* allele produces a protein that is necessary for either color; *ee* dogs are yellow.

    A black coat father and a yellow mother have a litter of puppies. 3 puppies are black, 1 is brown, and 4 are yellow. What are the genotypes of the parents?

7. In mice, the black agouti coat color is recessive. The dominant allele produces a yellow coat color. If an agouti mouse mates with a yellow mouse, 50% of the offspring are yellow and 50% are agouti. In an experiment, a research breeds two yellow mice. 8 of the offspring are yellow and 4 are black. Explain why (a Punnett square might help?)