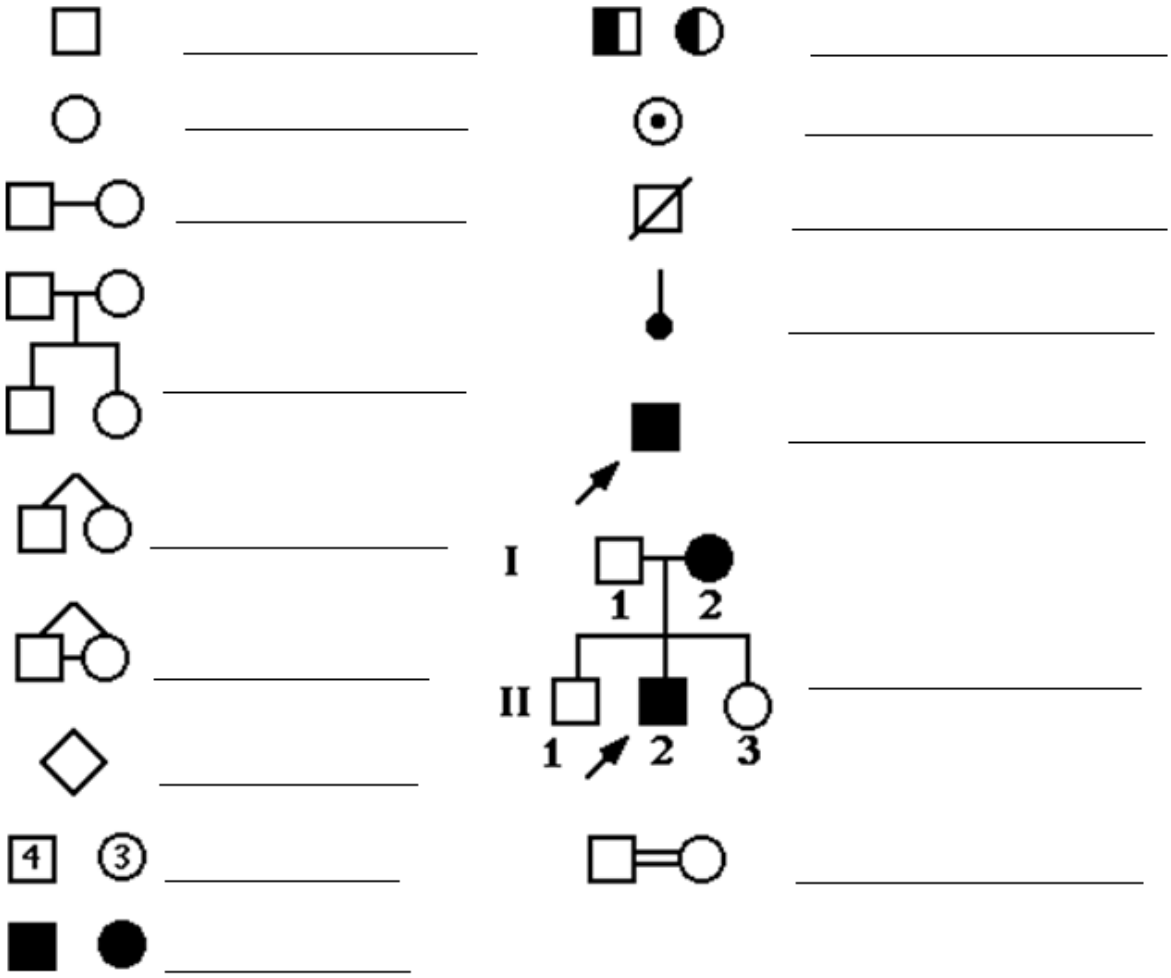
Name _____ Date _____

# Pedigree Construction Notes

GO TO → Mendelian Inheritance (http://www.uic.edu/classes/bms/bms655/lesson3.html)

When human geneticists first began to publish family studies, they used a variety of symbols and conventions. Now there are agreed upon standards for the construction of pedigrees.

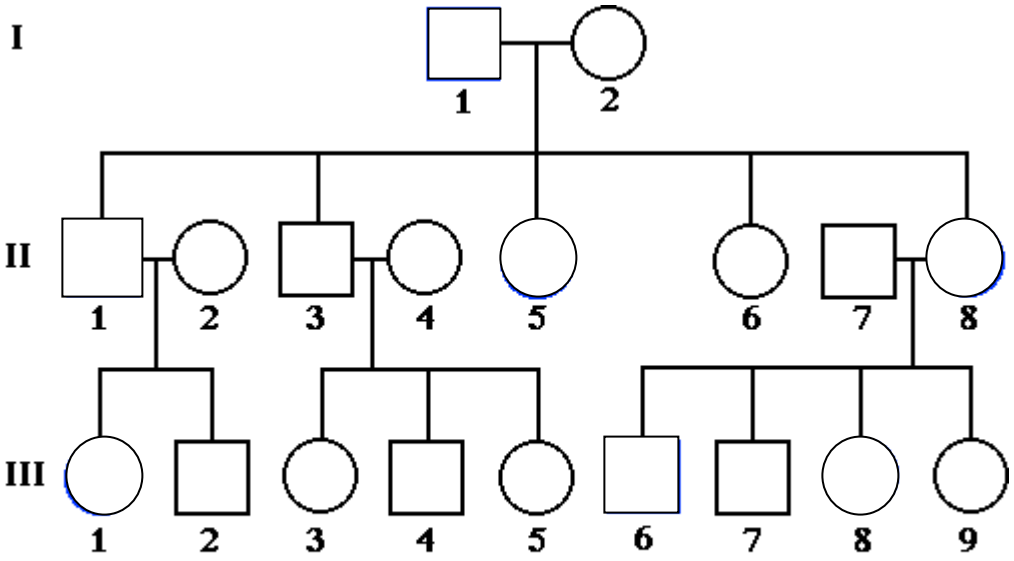



Remember:

1. **Males** are always represented by _____ symbols, **females** with _____symbols.
2. A _____ drawn between a square and a circle represents a mating of that male and female.
3. Two lines drawn between a square and a circle indicate a _____, the two individuals are _____, usually second cousins or closer relatives.
4. When possible, the square should be placed on the _____ and the circle on the _____ of the mating line.
5. Generations are connected by a _____ extending down from the mating line to the next generation.

6. Children of a mating are connected to a _____ line, called the _____, by short vertical lines.
7. The children of a sibship are always listed in _____, the oldest being on the _____.
8. Sometimes to simplify a pedigree only one parent is shown, the other is _____. This neither signifies parthenogenic development nor does it signify divinely inspired conception, it merely means the parent left out is not from the family being studied and is genotypically _____ for the trait being studied.
9. Normal individuals are represented by an _____ or _____, depending upon the gender, and affected individuals by a solid square or circle.
10. Each generation is numbered to the _____ of the **sibship** line with _____.
11. Individuals in each generation are numbered sequentially, beginning on the left, with _____. For example the third individual in the second generation would be identified as individual_____.

SCROLL DOWN TO "**AUTOSOMAL DOMINANT INHERITANCE**"
Read the short passage, shade the appropriate boxes and fill in the blanks below




**Pedigree 1. An idealized pedigree of a family with hypercholesterolemia, an autosomal dominant disease where the heterozygote has a reduced number of functional low density lipoprotein receptors.**

The family represented by Pedigree 1 is a good example of how **autosomal dominant** diseases appear in a pedigree. Each of the four hallmarks of autosomal dominant inheritance are fulfilled.

- Each affected individual has an _____; there is no skipping of generations.
- _____ and _____are equally likely to be affected.
- About 1/2 of the offspring of an affected individual are affected (_____).
- _____ (II-3) of affected individuals have all _____. Low density lipoprotein receptors are structural proteins or polypeptides, not enzymes.

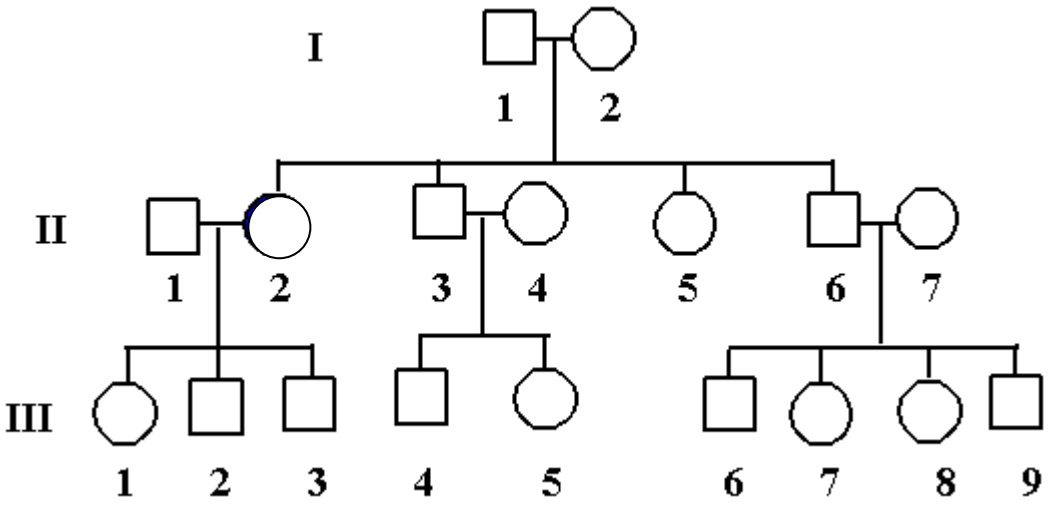
- If _____, an affected female, were to produce a child that child would have a 1/2 chance of being normal and a 1/2 chance of being affected. If her normal brother, _____, were to produce a child that child would have a nearly 0 chance of being affected.

GO TO → **AUTOSOMAL RECESSIVE INHERITANCE**
(http://www.uic.edu/classes/bms/bms655/lesson5.html)

The first, and most important, thing to remember about autosomal recessive inheritance is that most, if not all, affected individuals have parents with normal phenotypes.

**There are five hallmarks of autosomal recessive inheritance:**

1. _____
2. _____
   _____
3. _____
   _____
4. _____
   _____
5. _____
   _____



- The above pedigree illustrates four of the five hallmarks of autosomal recessive inheritance. _____ and _____are unrelated, yet they produced an affected offspring (_____**)**.
- By chance, they both must have been carriers. Even though II-2 is affected, she produced no affected offspring (_____**)**.
- By far the most probable genotype for an individual from _____ (II-1) is _____. III-1, III-2 and III-3 are all _____ (heterozygotes), since they are not affected but could only have inherited the recessive gene from II-2 II-3, II-5, and II-6 each have a 2/3 chance of being a carrier and a 1/3 chance of being homozygous normal. They are not affected, but they come from _____.
- II-4 and II-7 have a high probability of being _____ since they are from outside the family. III-4, III-5, III-6, III-7, III-8, and III-9 all have a 1/3 chance of being _____ and a 2/3 chance of being _____.
- One parent of each is probably _____, the other has a 2/3 chance of being a carrier and a 1 in 2 chance of passing on the recessive allele if they were a carrier.

GO TO → **X-LINKED INHERITANCE**
(http://www.uic.edu/classes/bms/bms655/lesson6.html)

When the locus for a gene for a particular trait or disease lies on the X chromosome, the disease is said to be _____. The inheritance pattern for X-linked inheritance differs from autosomal inheritance only because the X chromosome has _____ in the male, the male has an X and a Y chromosome. Very few genes have been discovered on the Y chromosome.

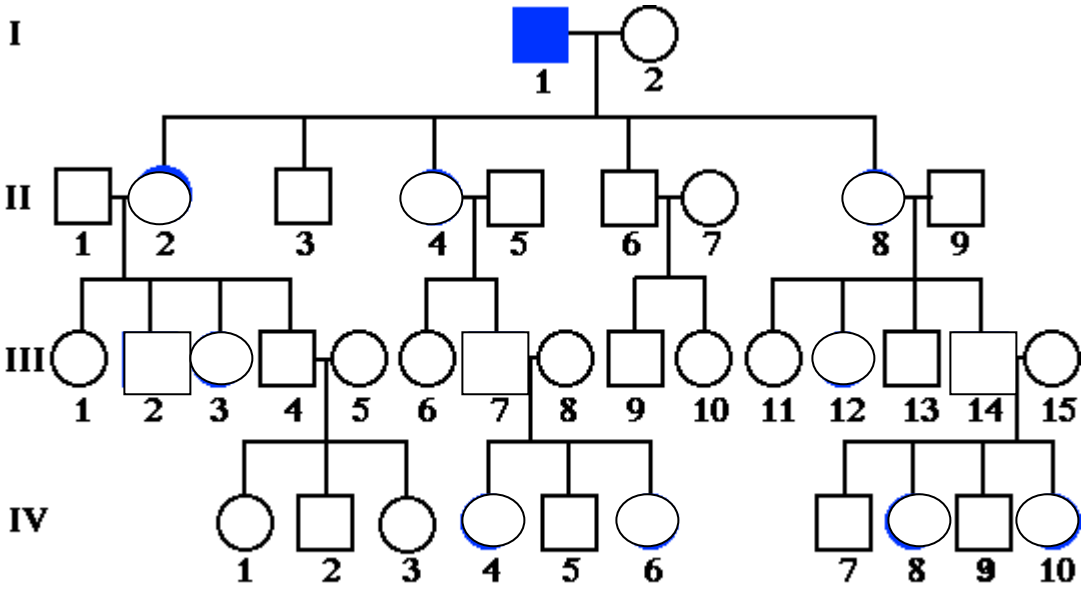
The inheritance pattern follows the pattern of segregation of the X and Y chromosomes in _____ and _____. A male child always gets his X from one of his _____ and his Y chromosome from his _____. **X-linked genes are never passed from _____.** A _____ child always gets the father's X chromosome and one of the two X's of the mother. **An affected female must have _____.** Males are always hemizygous for X linked traits, that is, they can never be heterozygoses or homozygotes. **They are never _____. A single dose of a mutant allele will produce a mutant phenotype in the male, whether the mutation is dominant or recessive.** On the other hand, females must be either homozygous for the normal allele, heterozygous, or homozygous for the mutant allele, just as they are for autosomal loci.

When an X-linked gene is said to express _____ inheritance, it means that a single dose of the mutant allele will affect the phenotype of the female. A _____ X-linked gene requires two doses of the mutant allele to affect the female phenotype. The following are the hallmarks of X-linked dominant inheritance:

- **The trait is never _____.**
- **All daughters of _____**

  **_____**
- **Matings of affected females and normal males produce _____--_____.**
- **Males are usually more _____. The trait may be lethal in _____.**
- **In the general population, _____, even if the disease is not lethal in males.**

Males are usually more severely affected than females because in each affected female there is _____ producing a normal gene product and one _____allele producing the non-functioning product, while in each affected male there is only the _____ allele with its non-functioning product and the _____, no normal gene product at all. Affected _____ are more prevalent in the general population because the female has two X chromosomes, either of which could carry the mutant allele, while the male only has one X chromosome as a target for the mutant allele. When the disease is no more deleterious in males than it is in females, _____ are about twice as likely to be affected as _____. As shown in Pedigree 5 below, X-linked dominant inheritance has a unique heritability pattern.

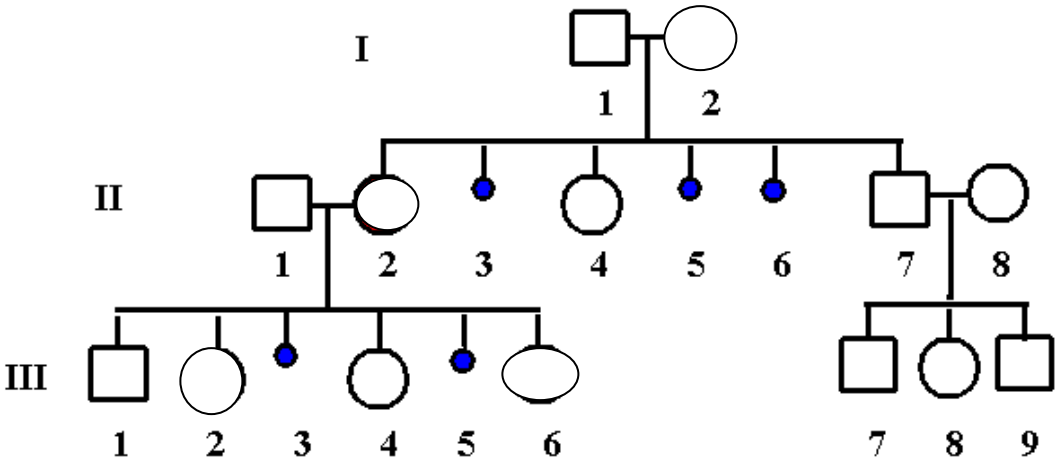



Pedigree 5. X-linked dominant inheritance.

The key for determining if a dominant trait is X-linked or autosomal is to look at the

If the affected _____has an affected son, then the disease is _____. All of his _____ must also be affected if the disease is _____. In Pedigree 5, both of these conditions are met.

What happens when _____ are so severely affected that they can't reproduce? Suppose they are so severely affected they never survive to term, then what happens? This is not uncommon in X-linked _____ diseases. There are no affected males to test for X-linked dominant inheritance to see if the produce all affected _____ and no affected _____. Pedigree 6 shows the effects of such a disease in a family. There _____, only affected females, in the population. Living _____ outnumber living males two to one when the mother is affected. The ratio in the offspring of affected females is: ____affected female: _____ normal female: _____ normal male.




**Pedigree 6.**

You will note that in Pedigree 6 there have also been several spontaneous _____ in the offspring of affected _____. Normally, in the general population of us normal couples, one in _____ recognized pregnancies results in a _____ abortion. Here the ratio is much _____. Presumably many of the spontaneous abortions shown in Pedigree 6 are _____ that would have been affected had they survived to term.