





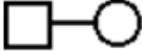

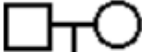




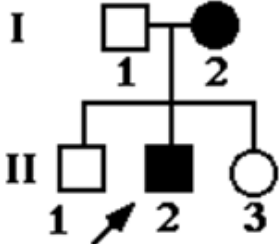







Name _____

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.

	_____		_____
	_____		_____
	_____		_____
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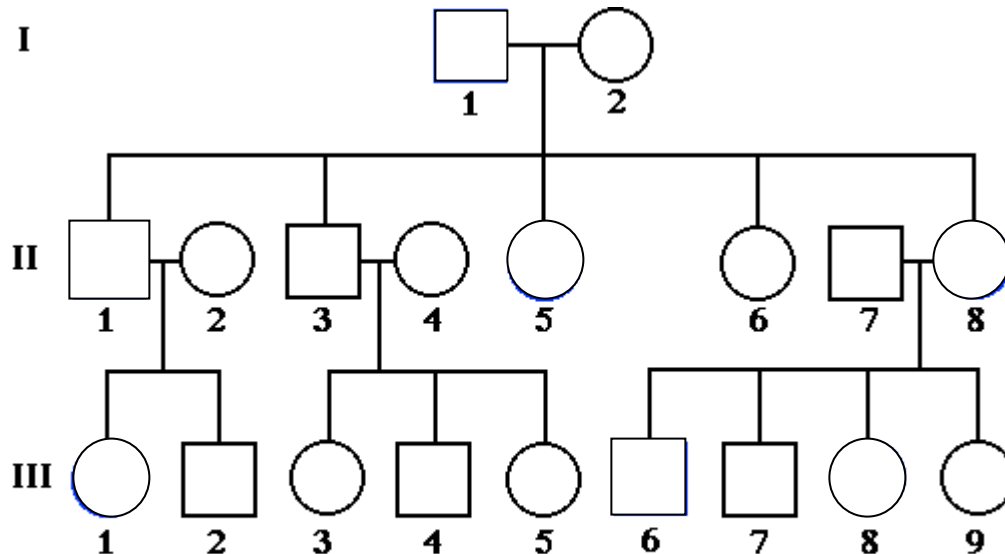
Remember:

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.

- Children of a mating are connected to a _____ line, called the _____, by short vertical lines.
- The children of a sibship are always listed in _____, the oldest being on the _____.
- 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.
- Normal individuals are represented by an _____ or _____, depending upon the gender, and affected individuals by a solid square or circle.
- Each generation is numbered to the _____ of the **sibship** line with _____.
- 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.

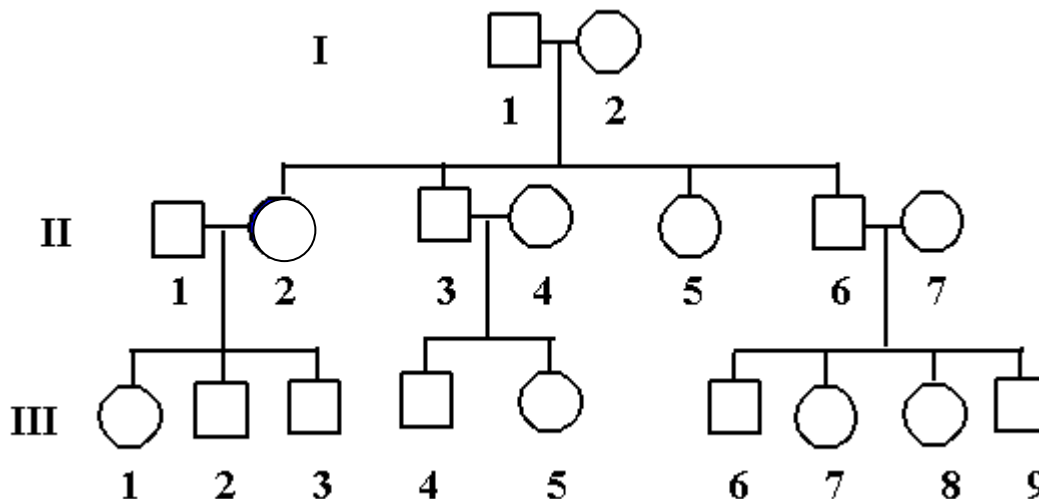
GO TO → *AUTOSOMAL RECESSIVE INHERITANCE*

(<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. Males and females are _____ likely to be affected.
2. On average, the recurrence risk to the unborn sibling of an affected individual is _____.
3. The trait is characteristically found in _____, not parents of affected or the offspring of affected.
4. Parents of affected children may be _____. The rarer the trait in the general population, the more likely a _____ mating is involved.
5. The trait may appear as an _____ (sporadic) event in small sibships.



- 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>)

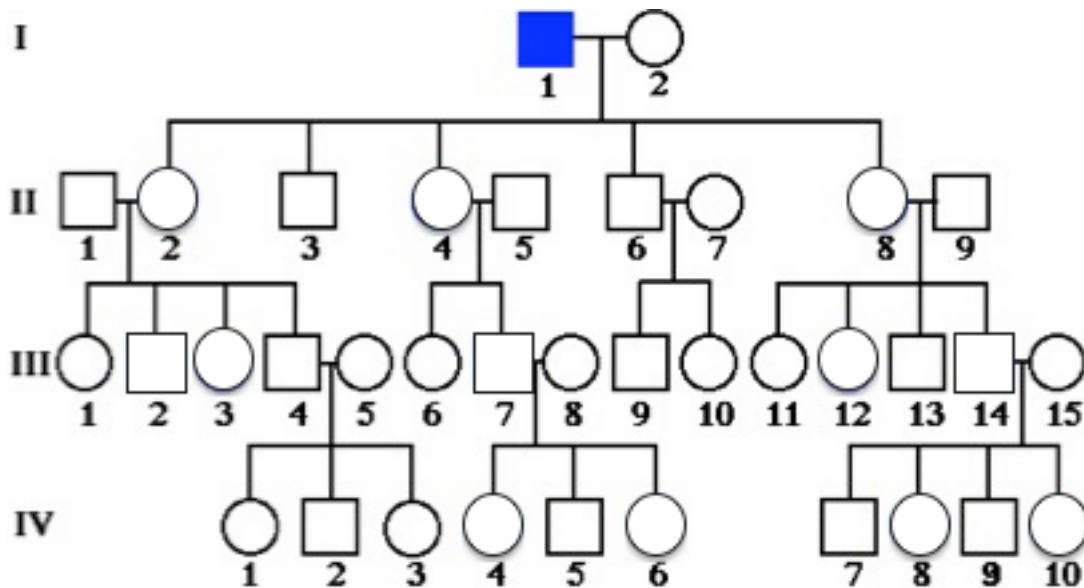
When the locus for a gene for a particular trait or disease lies on the X chromosome, the disease is said to be _____. The inheritance pattern for X-linked inheritance differs from autosomal inheritance only because the X chromosome has _____ in the male, the male has an X and a Y chromosome. Very few 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 heterozygotes 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 passed from _____ to _____.**
- **All _____ of an affected male and a normal female are affected. All sons of an affected male and a normal female are _____.**

- **Matings of affected females and normal males produce _____ the sons affected and _____ the daughters affected.**
- **Males are usually more _____ affected than females. The trait may be _____ in males.**
- **In the general population, _____ are more likely to be affected than males, 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.

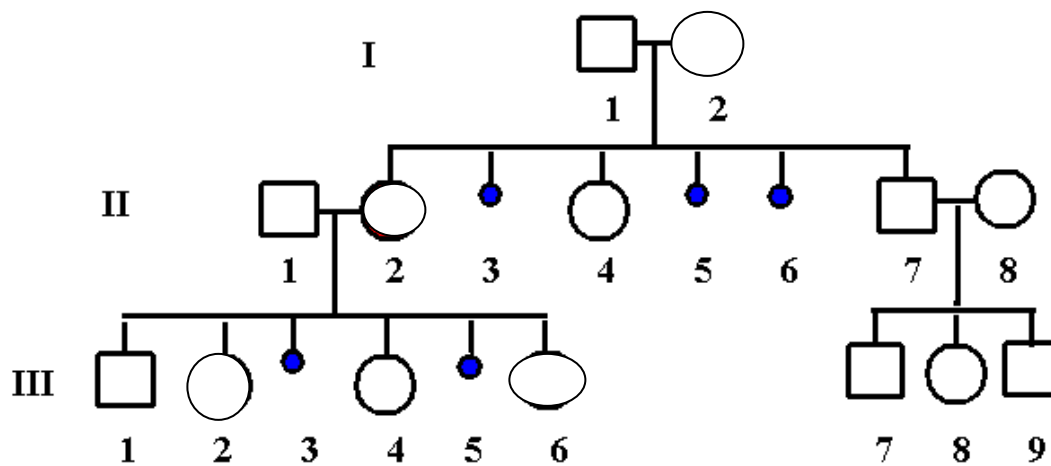
The key for determining if a dominant trait is X-linked or autosomal is to look at the

_____.

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 they 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.