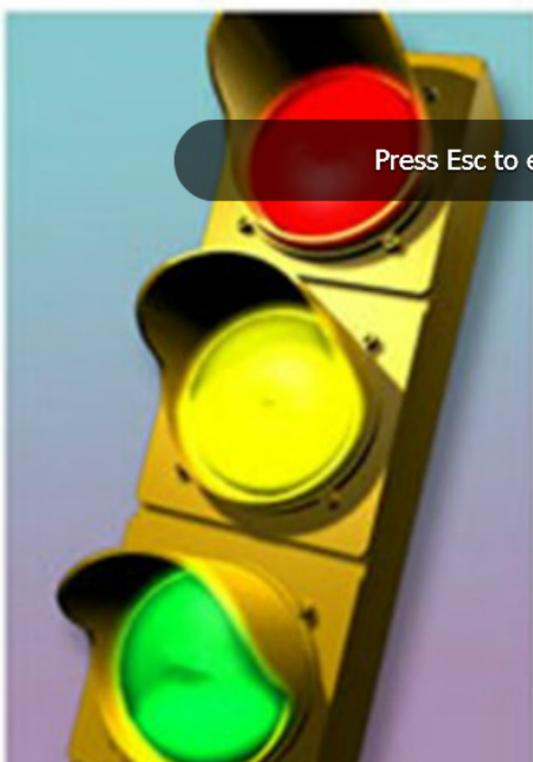
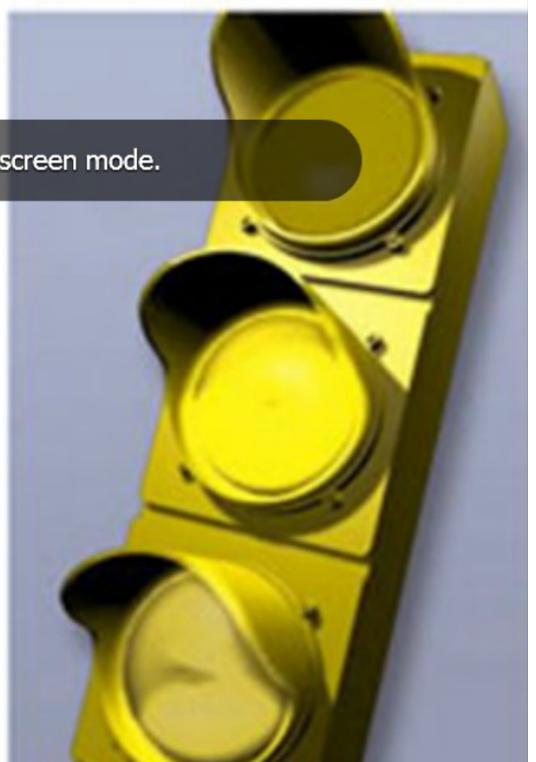


Sex-Linked Traits and Pedigrees

Original Image



Deutanope Simulation

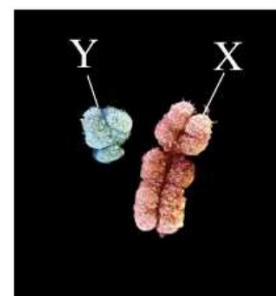
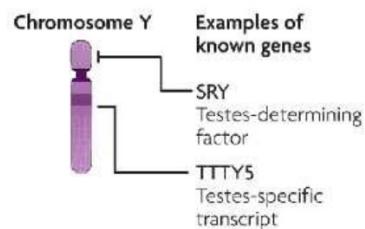
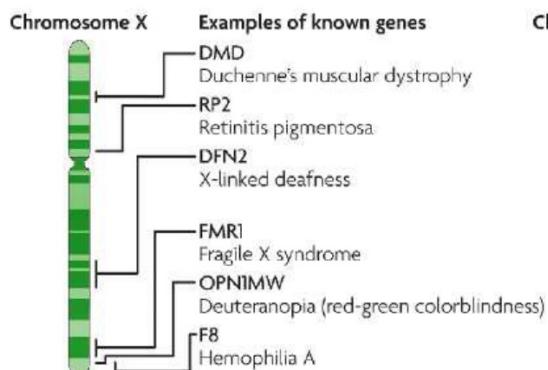


Press Esc to exit full screen mode.

7.4 Human Genetics and Pedigrees

• Females can carry sex-linked genetic disorders.

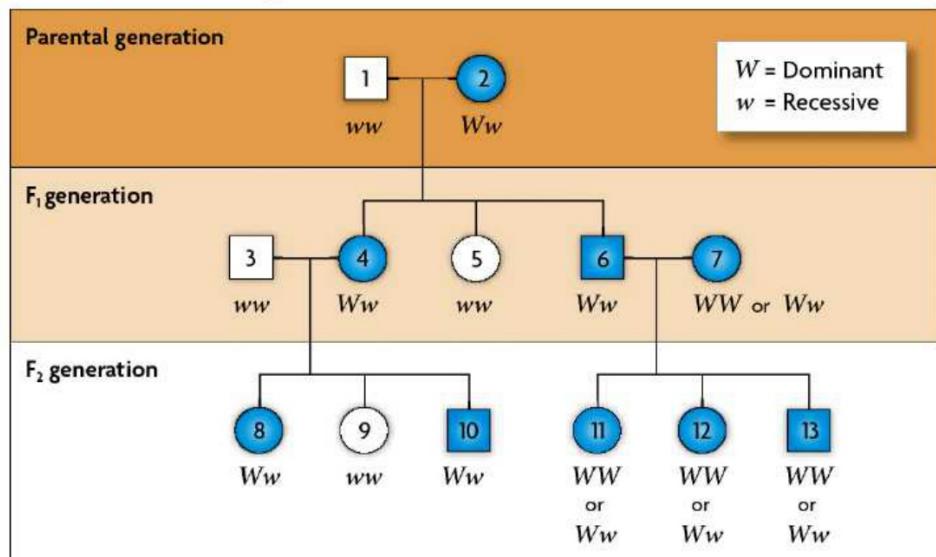
- Males (XY) express all of their sex linked genes.
- Expression of the disorder depends on which parent carries the allele and the sex of the child.



7.4 Human Genetics and Pedigrees

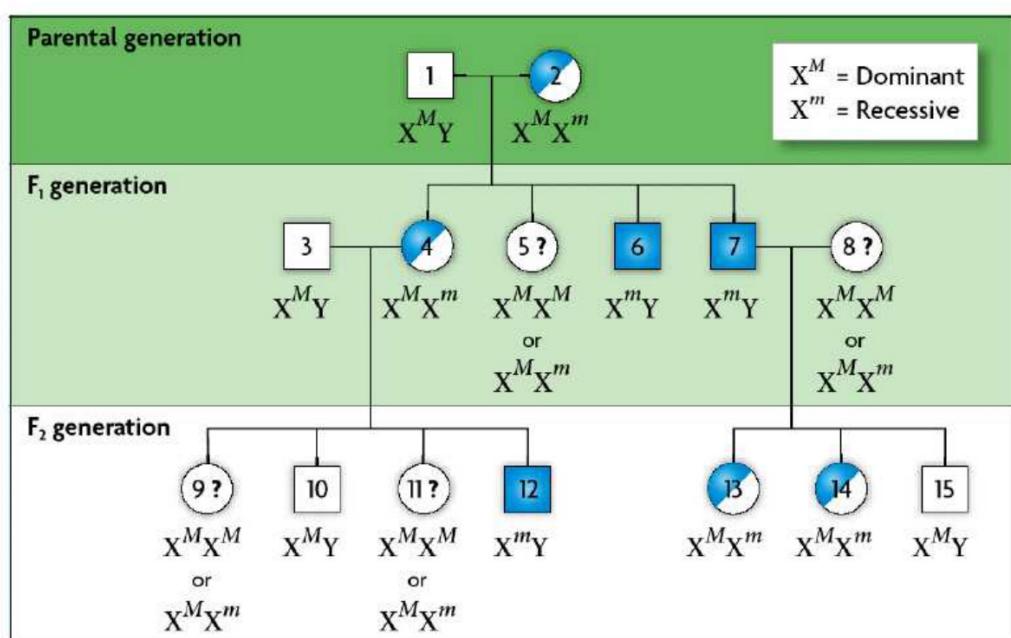
- A pedigree is a chart for tracing genes in a family.

- Phenotypes are used to infer genotypes on a pedigree.
- Autosomal genes show different patterns on a pedigree than sex-linked genes.



7.4 Human Genetics and Pedigrees

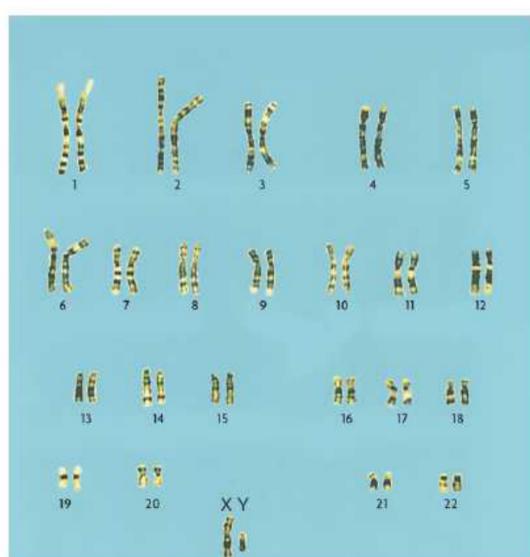
- If the phenotype is more common in males, the gene is likely sex-linked.



7.4 Human Genetics and Pedigrees

► Several methods help map human chromosomes.

- A karyotype is a picture of all chromosomes in a cell.



7.4 Human Genetics and Pedigrees

- Karyotypes can show changes in chromosomes.
 - deletion of part of a chromosome or loss of a chromosome
 - large changes in chromosomes
 - extra chromosomes or duplication of part of a chromosome



The Location of Genes

Remember that genes for specific traits are coded for in our DNA

- our (humans) DNA is divided into 23 pairs of chromosomes
 - all chromosome pairs are homologous (coding for the same traits, EXCEPT for the 23rd pair)
- The 23rd pair are known as sex chromosomes
- they determine whether an individual is male or female
 - #23 chromosomes come in two varieties, X, and Y

X and Y Chromosomes

If a human individual has two X chromosomes (XX), they are female, and both chromosomes are homologous

If a human individual has one X and one Y chromosome, they are male, and the 23rd pair of chromosomes is NOT homologous

-- in other organisms, XX individuals are male and XY individuals are female

X and Y chromosomes differ in many ways

The X Chromosome

The X chromosome contains genes for over 2300 traits

-- most of these traits are not involved in determining sex

Genes on the X chromosome are expressed in both males and females

The X chromosome has a centromere in the middle and is fairly large in size

How Sex is Determined

We can treat the sex chromosomes just like alleles (remember alleles are on each chromosome in a homologous pair)

The father has an X and a Y chromosome to give

The mother has two X chromosomes.

Therefore the cross looks like this:

	X	Y
X	XX	XY
X	XX	XY

As you can see, the chance of each sex being born is 50%

Traits on X and Y

What about traits located on the X chromosome?

Traits on the X chromosome behave in the normal dominant/recessive relationship for individuals with two X chromosomes (female), however, what about individuals who only have one X chromosome and one Y?

-- remember Y chromosomes contain almost no genes

For XY individuals (males), whatever trait is expressed on the X chromosome will be shown in the male individual

-- we call traits on the X-chromosome **sex-linked**

The Bigger Problem

Many unwanted traits are recessive traits located on the X chromosome.

-- this is not a big deal for someone who has two X chromosomes (female), because the other chromosome could have the dominant allele

-- if a male gets the recessive trait on his X chromosome, however, he gets the unwanted phenotype

Traits that are passed this way:

- red/green colorblindness
- hemophilia
- baldness

Sex-Linked Crosses

When we want to see how sex-linked traits are passed, we do a sex-linked crossed.

We write alleles so that the chromosome X is shown with an upper or lower-case superscript for the trait (dominant or recessive), and Y has no superscript

We also try to figure out if it is a boy or girl, what the percentage of each phenotype will be

Females who are heterozygous for a recessive trait are called **carriers**

Practice Problem

Hemophilia, a disease where blood does not clot properly, is caused by a recessive sex-linked allele. A female, who is a carrier for hemophilia and a normal male have a child. What is the probability that the child will be born with hemophilia?

What is the probability that a female child will be born with hemophilia? What about a male child?

Tracing Inherited Traits

Many times, individuals have questions about a specific trait or traits that run in families

-- this could be a question as to whether or not future generations could get a gene for an undesirable trait, like a genetic disease

-- this could also be a question as to whether a gene that codes for a specific trait is dominant, recessive, **autosomal** (on chromosomes 1 – 22), or sex-linked

In order to trace the development of a trait through a population, and figure out the answer, we construct a chart known as a **pedigree**

Reading a Pedigree

Pedigrees are read from top to bottom

Each generation of a family is placed in its own row, with the older generations on top of younger generations

Squares represent **males**

Circles represent **females**

If a male and female are connected by a square, they have mated and had offspring

-- offspring are hung “bracket-style” from the line connecting the two parents

Individuals who are shaded-in are afflicted by the trait

Individuals who are half-shaded are carriers (heterozygous) for the trait

Symbols in a Pedigree

	Male		Heterozygotes for autosomal recessive
	Female		Carrier of sex-linked recessive
	Mating		Death
	Parents & Children 1 boy 1 girl (in order of birth)		Abortion of stillbirth sex unspecified
	Dizygotic twins		Proband
	Monozygotic twins	 	Method of identifying persons in a pedigree Here the proband is Child 2 in Generation II
	Sex unspecified		Consanguineous marriage
	Number of children of sex indicated		
	Affected individuals		

Reading Pedigrees

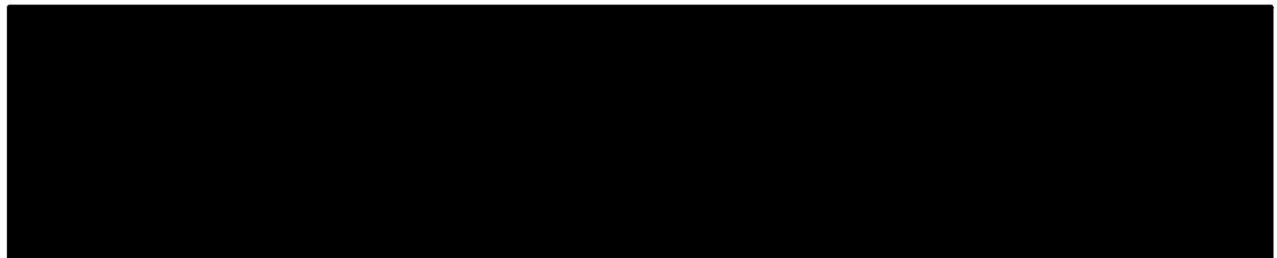
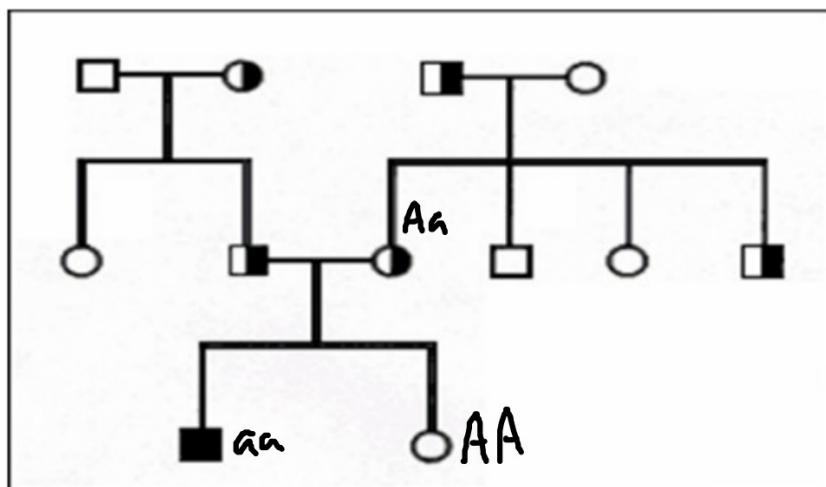
The hardest part of pedigrees is reading the pedigree and then trying to assess whether the trait was either:

- 4 modes of inheritance*
- A -- autosomal dominant AA, Aa
 - B -- autosomal recessive aa , $X^A X^A$, $X^A X^a$, $X^a X^a$, $X^A Y$, $X^a Y$
 - C -- sex-linked dominant
 - D -- sex-linked recessive

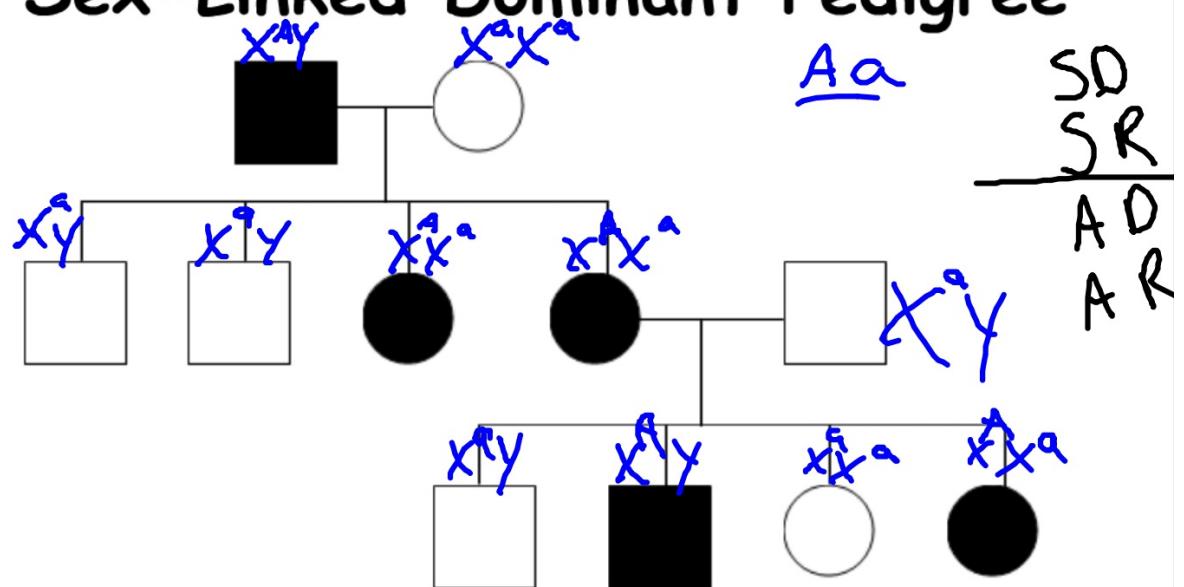
The best method to figure this out is to assign genotypes to affected individuals and then look at what the offspring of individuals should be if the cross follows each of the four patterns . . .

Some examples. . .

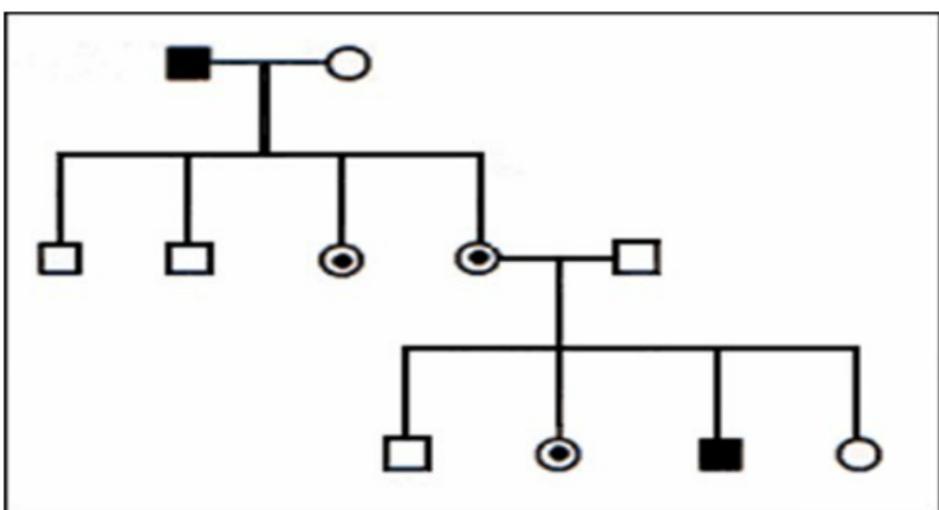
Autosomal Recessive Pedigree



Sex-Linked Dominant Pedigree



Sex-Linked Recessive Pedigree



Notice how there are no male carriers. Additionally, the female carrier in generation 2 gave the trait to 50% of her male offspring and the affected male in generation 1 had 100% of his daughters as carriers. This is definitely sex-linked and recessive

Is it a dominant pedigree or a recessive pedigree?

1. If two affected people have an unaffected child, it must be dominant pedigree: D is the dominant mutant allele and d is the recessive wild type allele. Both parents are Dd and the normal child is dd.
2. If two unaffected people have an affected child, it is a recessive pedigree: R is the dominant wild type allele and r is the recessive mutant allele. Both parents are Rr and the affected child is rr.
3. If every affected person has an affected parent it is a dominant pedigree.

Muscular Dystrophy Pedigree Chart

Background Information

Pedigree charts are very important to many different fields of science. One reason they are important is because, they help scientists understand the genetic patterns of diseases. It is important to be able to interpret pedigree charts in order to learn the pattern of a disease or condition. Specifically, using a pedigree chart, you can tell if the disease or condition is autosomal or X-linked, and dominant or recessive. Before you start this activity it is important to review several symbols:

- | | |
|---|---|
| <input type="checkbox"/> -Unaffected male | <input checked="" type="checkbox"/> - Affected male |
| <input type="circle"/> -Unaffected female | <input checked="" type="circle"/> -Affected female |

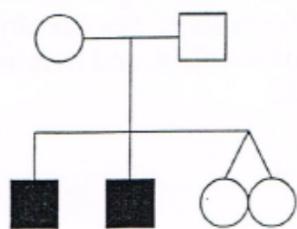
Procedure:

- A. First you need to become comfortable in making a pedigree chart. Complete the following examples.
 1. How can you tell if a couple is married on a pedigree? Write a one sentence description and draw example below.

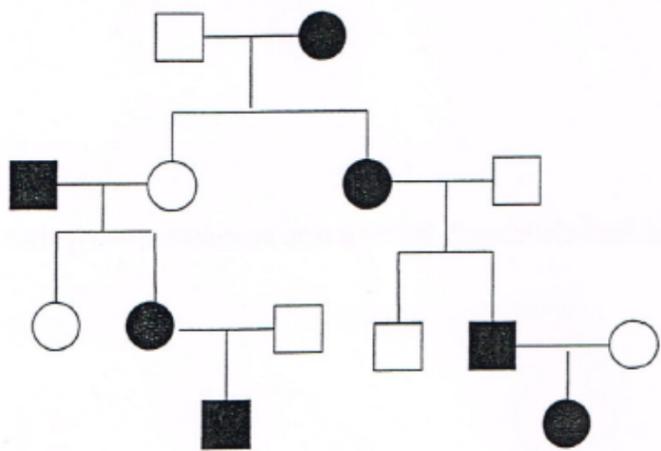
2. How can you tell if the couple who is married had children? Write a one sentence description and draw an example below.
3. Draw a pedigree that represents Mary married to Greg and with 2 sons (Scott and Tyler) and 1 daughter (Karen). Label the pedigree with the names of the people.
4. Draw a pedigree that represents Mary married to Greg, with 2 sons and 1 daughter. Their son, Scott, married April and had Sutton (a boy) and Kendall (a girl). Their daughter, Karen, married Harry and had Eli (a son) and Tanner (a son). Please label the pedigree with the names of the people.

B. Identify the following pedigree charts as autosomal or X-linked, and then as either recessive or dominant.

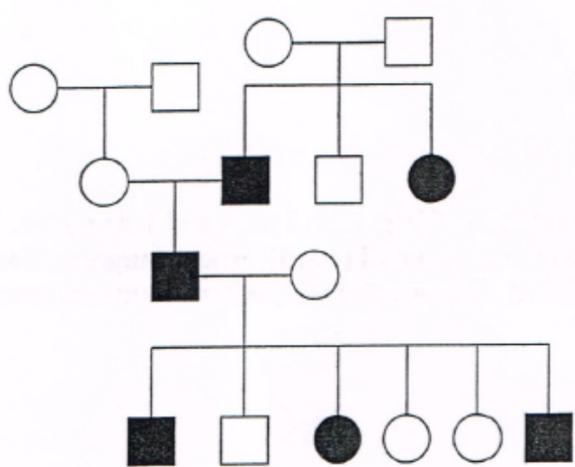
1. Is the following autosomal or X-linked? Is it dominant or recessive? Please explain your answer.



2. Is the following autosomal or X-linked? Is it dominant or recessive? Please explain your answer.

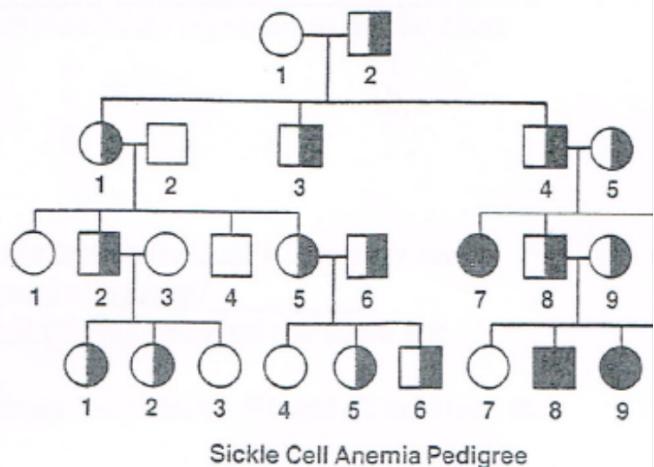
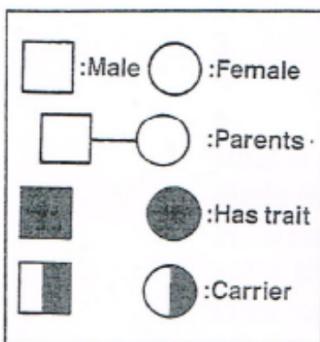


3. Is the following autosomal or X-linked? Is it dominant or recessive? Please explain your answer.



Procedure

1. Study the following key for the symbols used on a human pedigree.
2. Study the pedigree shown here. This pedigree traces the pattern of inheritance of sickle cell anemia in several generations of a single family.



Observations

1. How many generations are shown on the pedigree? _____
2. Which parent in the first generation had sickle cell anemia? _____
3. How many children were born in the second generation? _____
4. How many of these children are carriers of sickle cell anemia? _____
5. How many children in the third generation have sickle cell anemia? How many are carriers? _____

Analysis and Conclusions

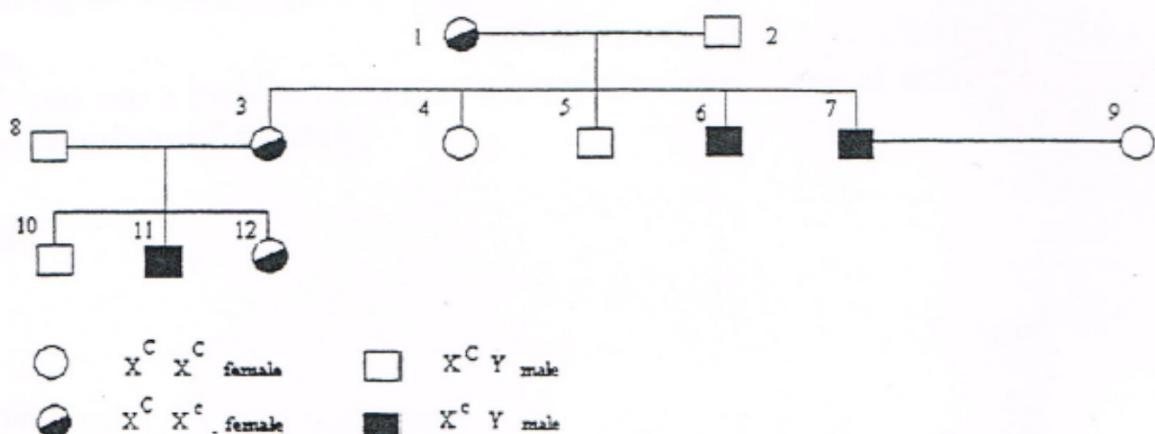
1. Is sickle cell anemia a sex-linked trait? How can you tell? _____

2. Is the gene for sickle cell anemia more likely to be dominant or recessive? Explain your answer. _____

3. **On Your Own** How could a genetic counselor use a pedigree to advise parents who are worried about passing on an inherited disorder to their children? _____

Procedure: Complete the questions and activities below the chart.

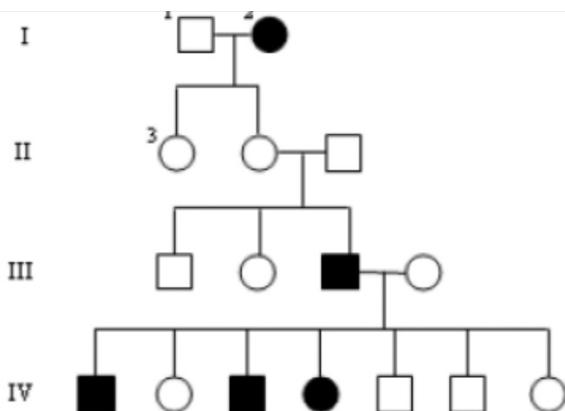
Data: The pedigree chart below shows the inheritance of the recessive sex-linked trait of red-green color blindness. The P (parent) generation shows a female carrier ($X^C X^c$) and a normal male ($X^C Y$). Small case "c" represents the allele for color blindness.



Analysis:

1. Write the genotype for each individual represented on the chart
1. 6. 11.
2. 7. 12.
3. 8.
4. 9.
5. 10.
 2. What is the chance for the mother, individual #1, to pass on the allele for color blindness to an offspring? _____
 3. How many children (#3 through #7) received the allele for color blindness? _____
 4. How many male children from individuals #1 and #2 received the allele? _____
 5. What are their numbers? _____
 6. How many children (#3-#7) have normal vision? _____
 7. What are their numbers? _____
 8. Individuals 8 and 3 marry. Which of their three offspring are color blind? _____
 9. Individuals 7 and 9 marry and have two male children. How many will be colorblind? _____ Draw the appropriate symbols for these children on the chart.
 10. Why aren't there any male carriers?

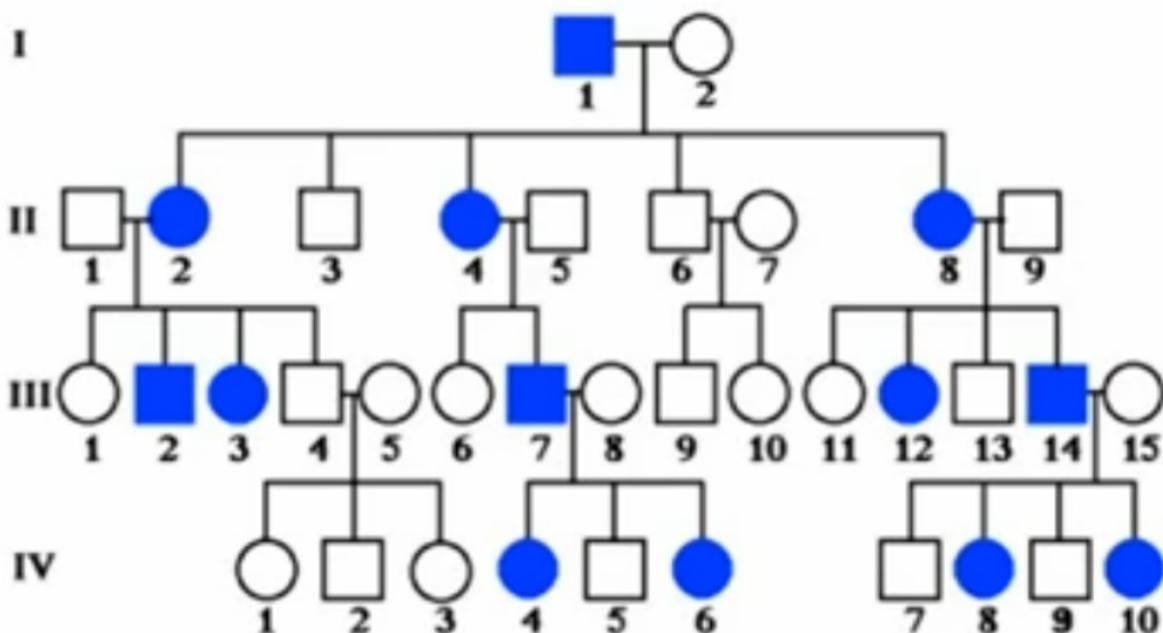
Use the pedigree below to answer 1-5

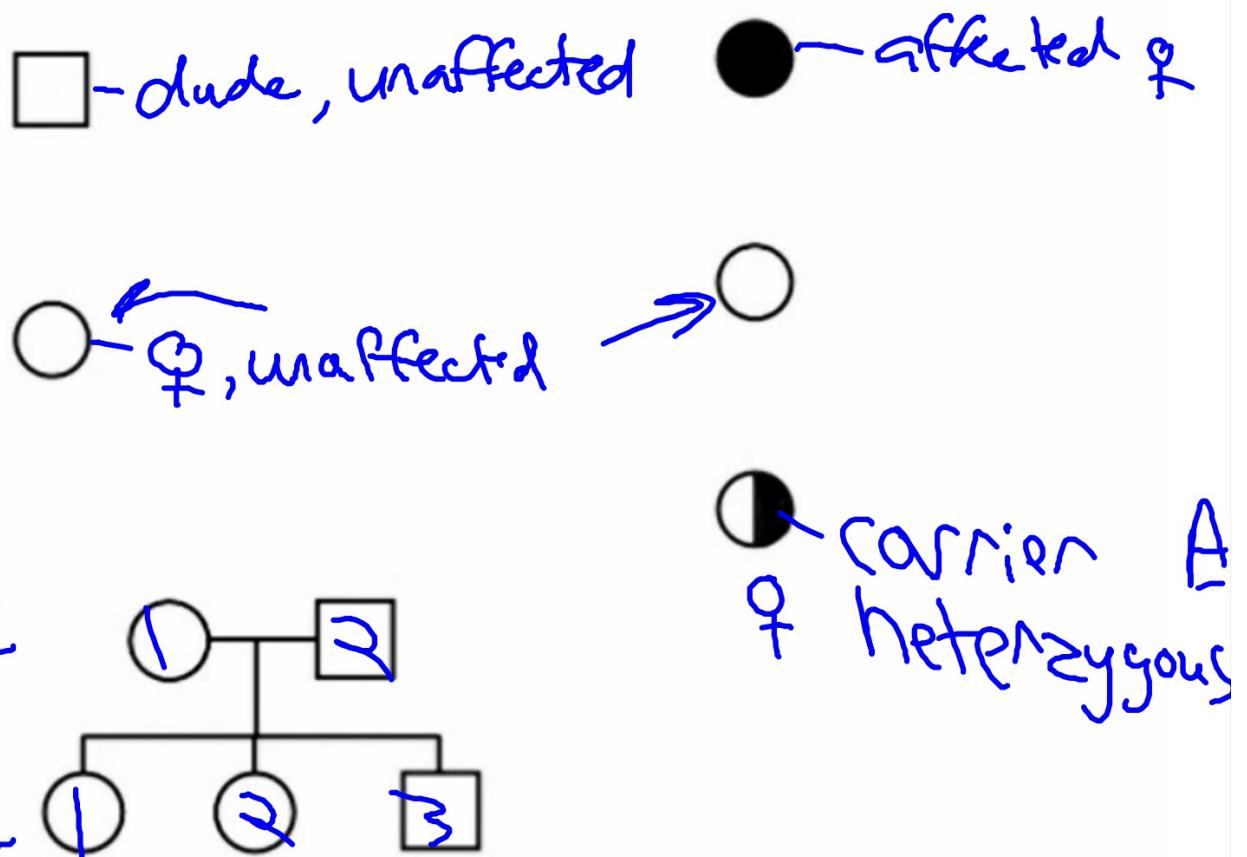


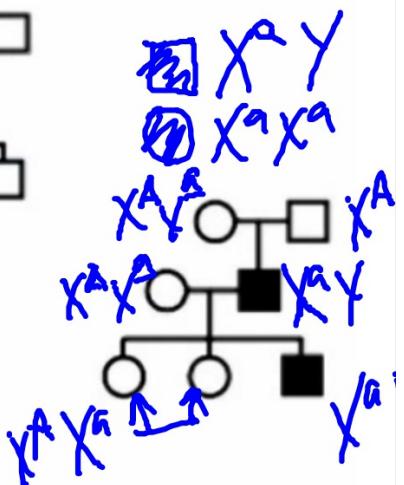
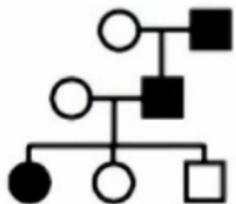
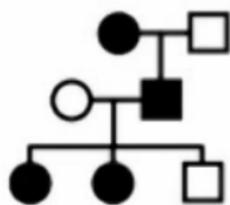
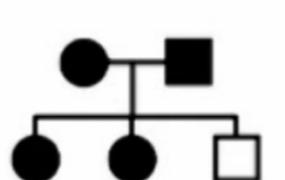
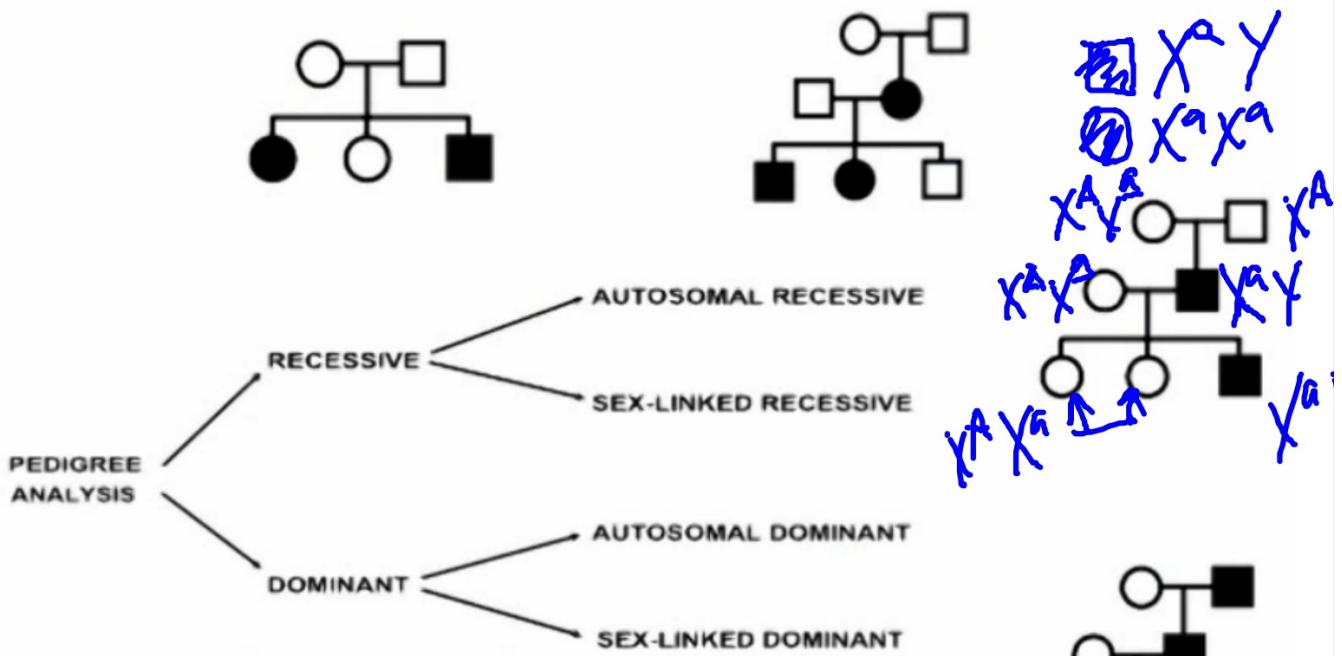
1. In a pedigree, a square represents a male. If it is darkened he has hemophilia; if clear, he had normal blood clotting.
 - a. How many males are there? _____
 - b. How many males have hemophilia? _____
2. A circle represents a female. If it is darkened, she has hemophilia; if open she is normal.
 - a. How many female are there? _____
 - b. How many females have hemophilia? _____
3. A marriage is indicated by a horizontal line connecting a circle to a square.
 - a. How many marriages are there? _____
4. A line perpendicular to a marriage line indicates the offspring. If the line ends with either a circle or a square, the couple had only one child. However, if the line is connected to another horizontal line, then several children were produced, each indicated by a short vertical line connected to the horizontal line. The first child born appears to the left and the last born to the right.
 - a. How many children did the first couple (couple in row I) have? _____
 - b. How many children did the third couple (couple in row III) have? _____
5. Level I represent the first generation, level II represents the second generation.
 - a. How many generations are there? _____
 - b. How many members are there in the fourth generation? _____

Kickoff: If the mode of inheritance for a particular trait is sex-linked dominant, what are the possible genotypes could exhibit the phenotype?

Determine the mode of inheritance and give the possible genotypes for individual III - 7.

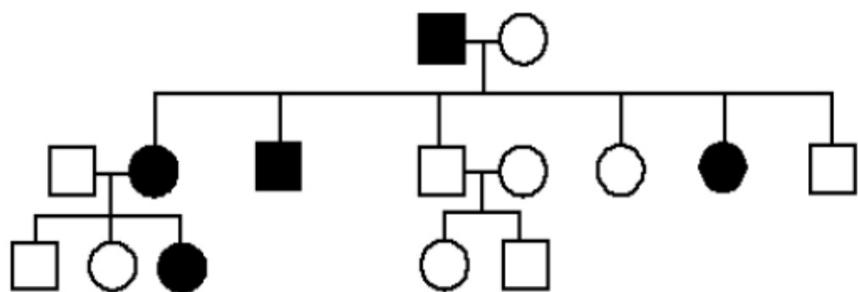






Use the pedigree below to answer 6-12

Shaded individuals have Huntington's Disease



6. Write the generation on the pedigree numbers (roman numerals).

7. Which members of the family above are afflicted with Huntington's Disease? _____

8. There are no carriers for Huntington's Disease- you either have it or you don't.

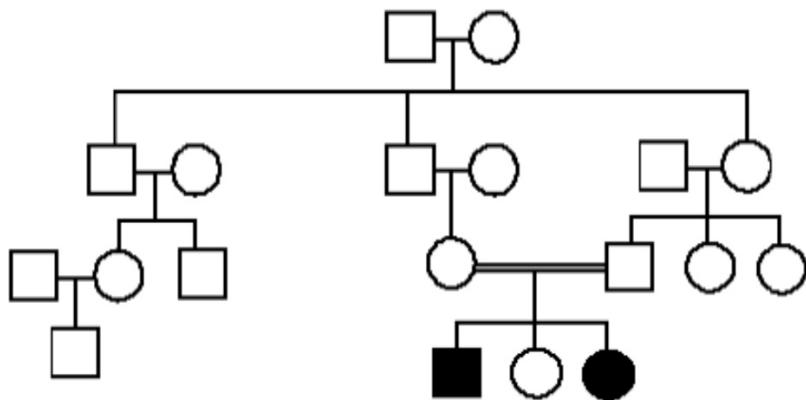
With this in mind, is Huntington's disease caused by a dominant or recessive trait? _____

9. How many children did individuals I-1 and I-2 have? _____

10. How many girls did II-1 and II-2 have? _____ How many have Huntington's Disease? _____

11. How is individual III-2 and II-4 related? _____ I-2 and III-5? _____

12. Write the genotypes of each individual on the pedigree.



13. Write the generation on the pedigree numbers (roman numerals).

The pedigree to the above shows the passing on of Hitchhiker's Thumb in a family. Is this trait dominant or recessive? _____

14. How do you know? _____

15. How are individuals III-1 and III-2 related? _____

16. Name 2 individuals that have hitchhiker's thumb. _____

17. Name 2 individuals that were carriers of hitchhiker's thumb. _____

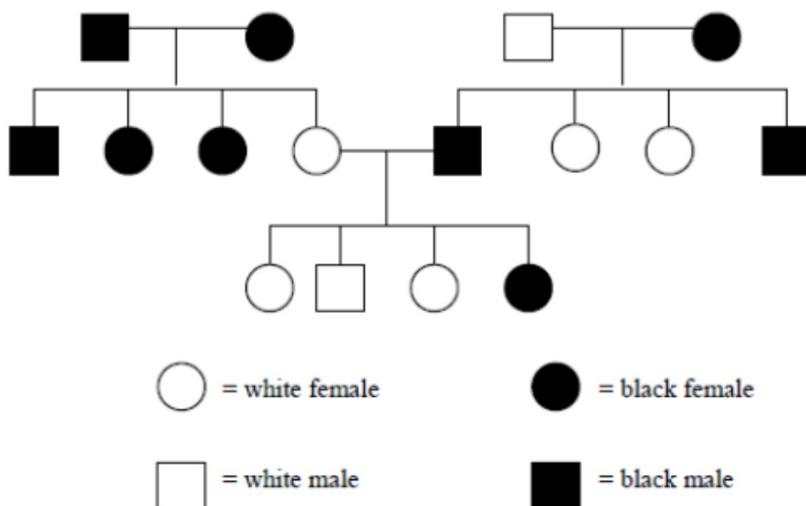
18. Write the genotypes for each individual on the pedigree.

Determining Inheritance Patterns

19. When working through a pedigree, the first thing you need to do is figure out which characteristic is dominant – the shaded one or the un-shaded one. Then you need to choose a letter (let's use A) and begin assigning genotypes. Remember that recessive individuals are **always** homozygous, so assign their genotype first. Then go back and look at all of the dominant individuals. For some, you will only be able to determine one allele of the genotype, so just write the one capital allele followed by a question mark (A?).

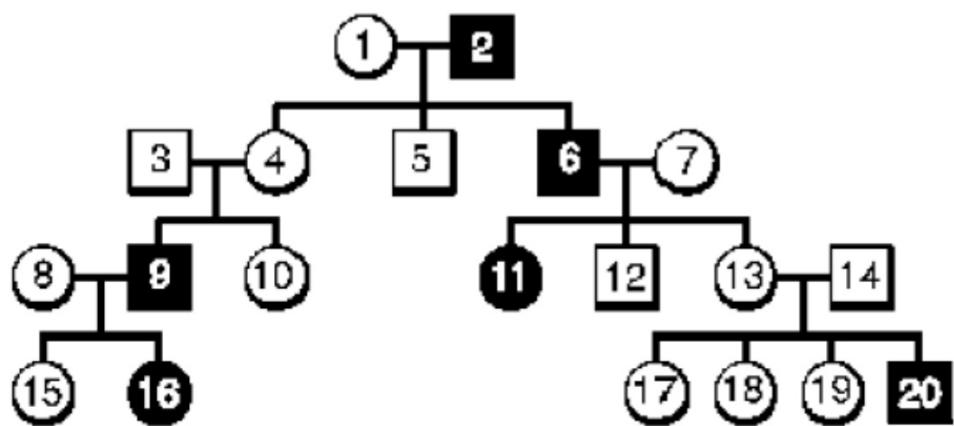
- Which characteristic is dominant? _____
- Which characteristic is recessive? _____
- Determine the genotypes of all individuals. You will have three "A?". Write your Genotypes beneath each individual.

Fur Color in Mice



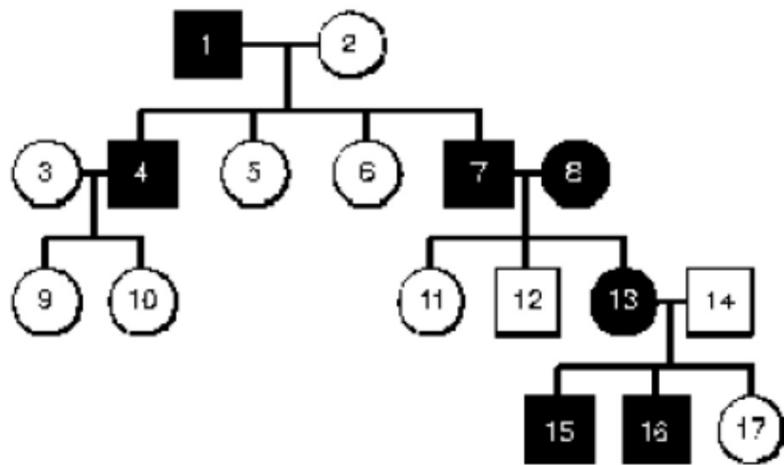
20. Is the trait dominant or recessive? _____

Write the genotype for each individual (use the letter A)



21. Is the trait dominant or recessive? _____

Write the genotype for each individual (use the letter A)



Making Conclusions

22. If a child has an autosomal dominant trait, what can you say about the parents?
23. If two parents have an autosomal dominant trait, what can you say about their children?
24. If two parents have an autosomal recessive trait, what can you say about their children?
25. If two parents do not have an autosomal recessive trait, what can you say about their children?
26. Can autosomal recessive traits skip generations?