

**Heredity**  
**Unit 4 Test Review**

Name: Answer Key Date: \_\_\_\_\_ Hour: \_\_\_\_\_ Test #: \_\_\_\_\_

1. Define the following terms:

a. Sex chromosomes: chromosomes involved in sex determination, X and Y in humans

i. Male: XY

ii. Female: XX

b. X-linked trait: trait related to sex, allele for which is found on the X chromosome but not the Y chromosome

i. What are the patterns of x-linked recessive inheritance?

1. Hemizygous males (with trait) pass the allele to all daughters, who then are carriers or have the trait
2. Carriers pass the allele to 50% of their sons.
3. Fathers cannot pass the trait to their sons, only to the sons of their daughters, their grandsons.
4. Phenotype will be present at a much higher rate in males than females.

c. Pedigree: family record that shows how a trait is inherited over several generations

d. Nondisjunction: when chromosomes fail to properly separate at anaphase

e. Trisomy: possessing three homologous chromosomes instead of a homologous pair

i. What is the most common trisomy genetic disorder? Trisomy 21 - Down Syndrome

f. Aneuploidy: possessing a chromosome number that is not an exact multiple of the haploid

g. Carrier: a person who is heterozygous for a trait, usually used when referring to a genetic disease

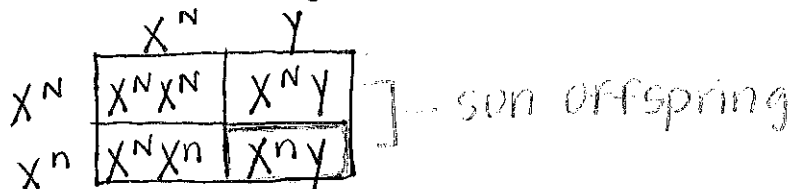
h. Homogametic: produce gametes that contain only one type of sex chromosome (XX)

ti. Hemizygous: produce gametes that contain different kinds of sex chromosomes

j. Heterogametic: when an individual (usually male) has only one allele, controlling a trait, single dominant allele causes dominant trait, single recessive allele causes recessive trait

2. How can humans assess the risks of passing on a genetic disorder to their offspring? Analysis of a pedigree, genetic counselor, prenatal testing

3. Can two normal parents have a son with an x-linked disorder? Yes, if mother is a carrier she passes the recessive gene to half her sons.



4. Name and describe the three X-linked Recessive diseases discussed in your notes. Include genotype, phenotypic characteristics, and rate of occurrence.

- a. Hemophilia: Genotype  $X^hY$ ,  $X^hX^h$  / carrier -  $X^H$ ,  $X^h$  / occurrence males 1:10,000 females 1:100,000 / disease in which blood lacks one of the clotting factors / phenotypic - blood clots very slowly, may bleed to death from cut, rare in females bc affected
- b. Duchenne's Muscular Dystrophy: Genotype  $x^mY$ ,  $x^mx^m$  / carrier  $x^Mx^m$  / occurrence males 3:10,000 / X-linked form of the disease that produces muscular weakness / Gower sign, onset 1-6 years, confined to wheelchair by 12, death by 20, passed on by carriers
- c. colorblindness: Genotype  $x^nY$ ,  $x^nx^n$  / carrier  $x^Nx^n$  / occurrence 8:100 / X-linked disorder in which an individual cannot distinguish between certain colors, usually red and green, colors appear as shades of gray

males do not survive to reproductive age, may be treated with injection

5. Which parent determines the sex of the offspring? Why? Father, can contribute X or Y

6. What is the actual ratio in live births of males to females? 1.05 male : 1 female

a. What is the sex ratio at age 25 of males to females? 1 male : 1 female

b. Why do more males die at a young age? X-linked disease and accidents

7. Which form of monosomy is always lethal?  $\emptyset Y$

8. Name and describe the diseases caused by XY aneuploidy discussed in your notes. Include genotype, phenotypic characteristics, and rate of occurrence.

a. Turner Syndrome - Monosomy X (45,X) Genotype:  $X\emptyset$  phenotypic characteristics: short, wide chested w/ under-developed breasts, lack of ovarian development, narrowing of aorta / rate of occurrence: 1% of all conceptions, 1 in 10,000 live births

b. Klinefelter Syndrome - 47, XXY Genotype: XXY phenotypic characteristics: no problems until puberty, poor sexual development, lowered fertility, breast development in 50% of cases, 1 in 1000 male births language deficits and academic difficulties

c. XYY Syndrome - 47, XYY Genotype - XYY phenotypic characteristics: above avg. height, personality disorders, learning disabilities, hyperactivity, impulsive behavior, lack of emotional control rate of occurrence: 1 in 1000 male births

d. XXX Syndrome: 47, XXX Genotype - XXX phenotypic characteristics: most perfectly normal, slight increase in sterility and learning disabilities rate of occurrence: 1 in 1000 female births

**Practice Problems**

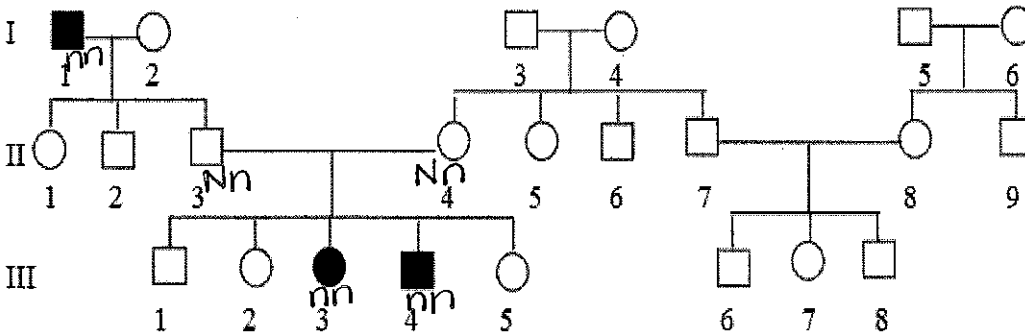
9. In a family, a female child has wiggly ears, whereas her brother does not. Both parents are normal. Can the wiggly ear trait be an autosomal recessive trait? Yes Can it be an x-linked recessive trait? No - no daughter offspring are  $X^w X^w$
- Autosomal Recessive  
 Mom -  $Ww$  Dad -  $Ww$
- |     |      |      |
|-----|------|------|
|     | $W$  | $w$  |
| $W$ | $WW$ | $Ww$ |
| $w$ | $Ww$ | $ww$ |
- x-linked Recessive  
 Mom -  $X^W X^w$  Dad -  $X^W Y$
- |       |           |           |         |
|-------|-----------|-----------|---------|
|       | $X^W$     | $X^w$     | $Y$     |
| $X^W$ | $X^W X^W$ | $X^W X^w$ | $X^W Y$ |
| $X^w$ | $X^W X^w$ | $X^w X^w$ | $X^w Y$ |
10. In humans, red-green color blindness is an X-linked recessive trait. If a man with normal vision and a color-blind woman have a son, what is the chance that the son will be color-blind? What is the chance that a daughter will be color-blind? \_\_\_\_\_

**Pedigree Practice Problems**

A pedigree is a chart of a person's ancestors that is used to analyze genetic inheritance of certain traits – especially diseases. The symbols used for a pedigree are:

- female, unaffected
- male, unaffected
- female, affected
- male, affected

- Siblings are placed in birth order from left to right and are labeled with numbers.
- Each generation is labeled with a Roman numeral.
  - Example: we would name an individual II-3 if he/she was in the second generation and the 3<sup>rd</sup> child born



Try to identify the genotypes of the following individuals using the pedigree above. (homozygous dominant, homozygous recessive, heterozygous)

- III-3: nn - homozygous recessive
- I-1: nn - homozygous recessive
- II-1: Nn - heterozygous
- II-4: Nn - heterozygous

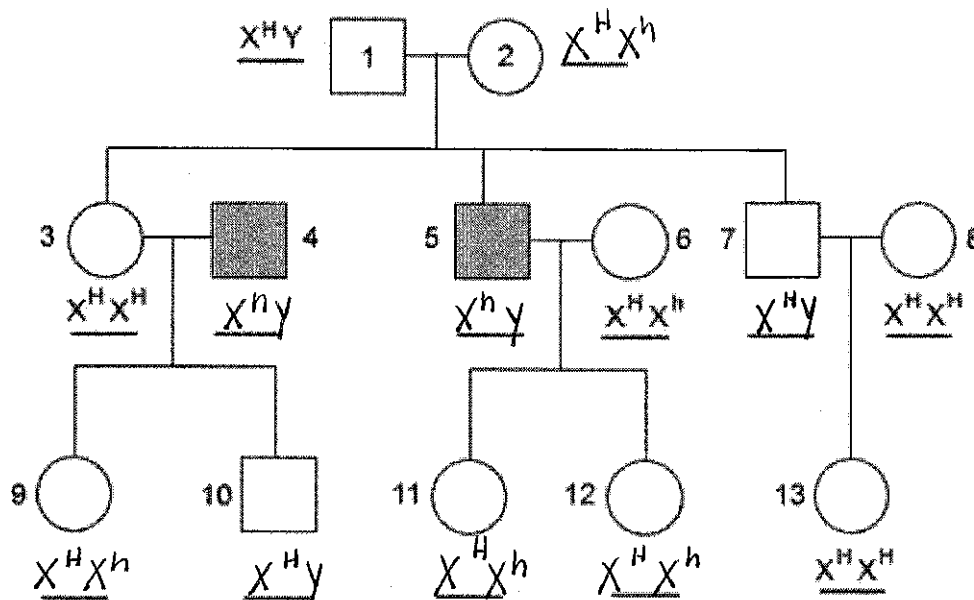
1. Is this trait dominant or recessive? Explain your answer.

This trait is recessive because unaffected individuals have affected children, so they must carry the allele for the disorder.

2. How can you know for sure that individuals II-3 and II-4 are heterozygous?

II-3 and II-4 are unaffected, but have 2 affected children, so they must carry the allele for the disorder.

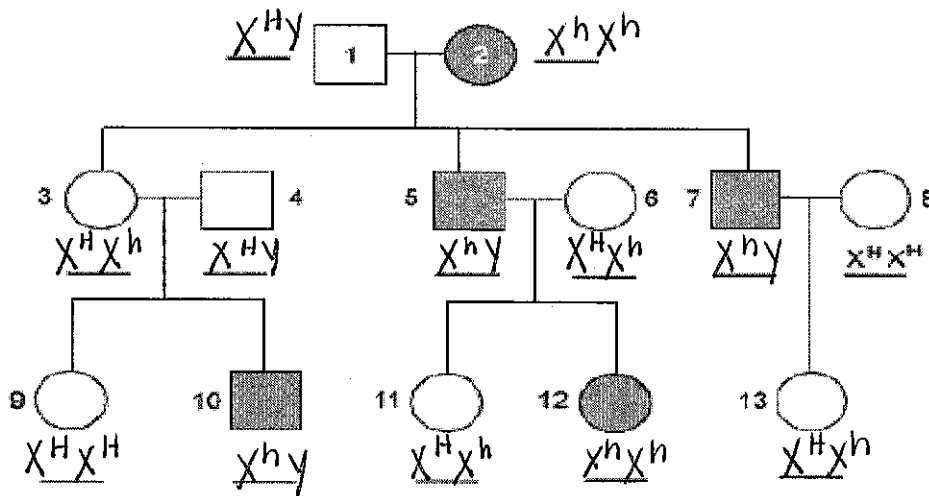
1) Hemophilia is a recessive sex-linked disorder located on the X chromosome where a person's body cannot control blood clotting or coagulation. Write in the genotypes on the line next to / below each individual.



For all phenotype questions, on the first line write: male or female and the second line write: normal, carrier, hemophilia.

- |  |               |                             |
|--|---------------|-----------------------------|
| 2) What is the phenotype of individual 2?  | <u>Female</u> | <u><math>X^H X^h</math></u> |
| 3) What is the phenotype of individual 4?  | <u>Male</u>   | <u><math>X^h Y</math></u>   |
| 4) What is the phenotype of individual 5?  | <u>Male</u>   | <u><math>X^h Y</math></u>   |
| 5) What is the phenotype of individual 7?  | <u>Male</u>   | <u><math>X^H Y</math></u>   |
| 6) What is the phenotype of individual 9?  | <u>Female</u> | <u><math>X^H X^h</math></u> |
| 7) What is the phenotype of individual 10? | <u>Male</u>   | <u><math>X^H Y</math></u>   |
| 8) What is the phenotype of individual 11? | <u>Female</u> | <u><math>X^H X^h</math></u> |
| 9) What is the phenotype of individual 12? | <u>Female</u> | <u><math>X^H X^h</math></u> |

10) Fragile-X syndrome is a recessive sex-linked disorder located on the X chromosome. Below is a pedigree tracing the passing of the fragile-X syndrome gene through 3 generations. Write in the genotypes on the line next to / below each individual.



For all phenotype questions, on the first line write: male or female and the second line write: normal, carrier, fragile-X.

- 11) What is the phenotype of individual 1? Male  $X^H Y$   
 12) What is the phenotype of individual 4? Male  $X^H Y$   
 13) What is the phenotype of individual 6? Female  $X^H X^h$

14) Below is a recessive sex-linked pedigree tracing the red-green colorblindness gene located on the X chromosome. Write in the genotypes on the line next to / below each individual.

