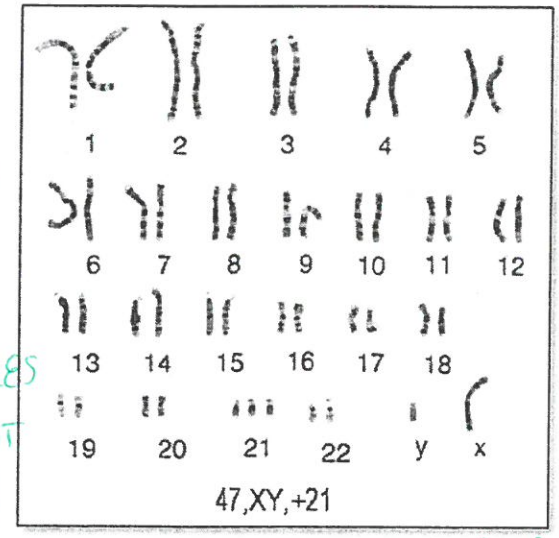
**Chapter 14 Review Guide Biology** Name: _____

The Human Genome

1. Understand how to analyze a karyotype →




a. What sex is this person? Male

b. What chromosome abnormality is present? trisomy 21

c. How many chromosomes are present? 47

d. Compare and Contrast sex chromosomes and autosomes.
(You cannot say they are both chromosomes for a similarity.)

SEX
- determine gender
- 23rd pair of chromosomes

BOTH
- found in pairs
- found in karyotypes

AUTOSOMES
- ~~dete~~ NOT determine gender
- 1st 22 chromosome pairs

2. What is a Chromosomal Abnormality?

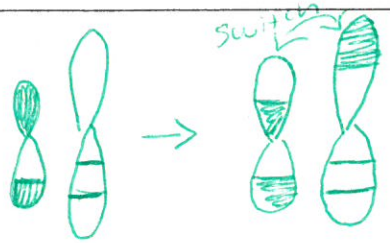
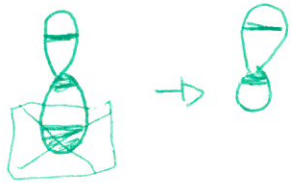
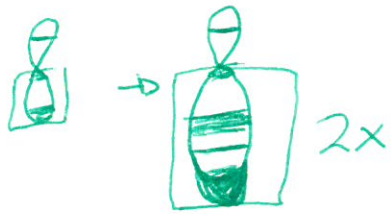
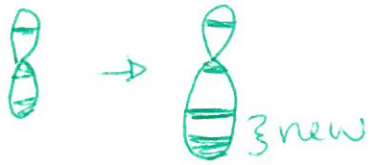
Something that makes a chromosome abnormal

3. What are the two types of chromosomal abnormalities?
   1. Chromosome Structure
   2. Chromosome Number

4. Fill in the table below regarding chromosome number errors.

| Term | Definition | How it occurs |
|---|---|---|
| Monosomy | only 1 of a chromosome | Errors in Meiosis (Nondisjunction) |
| Trisomy | 3 of a chromosome | Errors in meiosis (Nondisjunction) |
| Polyploidy | Multiple sets of each chromosome | Plants Meiosis Errors |

5. Use information from your notes, vocabulary, and activities in class to fill in the table below:

| Type of Chromosome Structure Mutation | Explanation of what changes | Picture illustrating the change. |
|---|---|---|
| Inversion | A section of the chromosome that is flipped/rotated. |   |
| Translocation | when chromosomes switch segments of DNA/genes with other chromosomes. |   |
| Deletion | when a segment of a chromosome is removed. |  |
| Duplication | when a segment of a chromosome appears more than once. |   |
| Insertion | when new DNA/genes are placed into a chromosomes |   |

6. What is a carrier?

A. What gender(s) can carry recessive Sex-Linked traits? female

B. What gender(s) can carry dominant Sex-Linked traits? no-one: dominant = no carriers

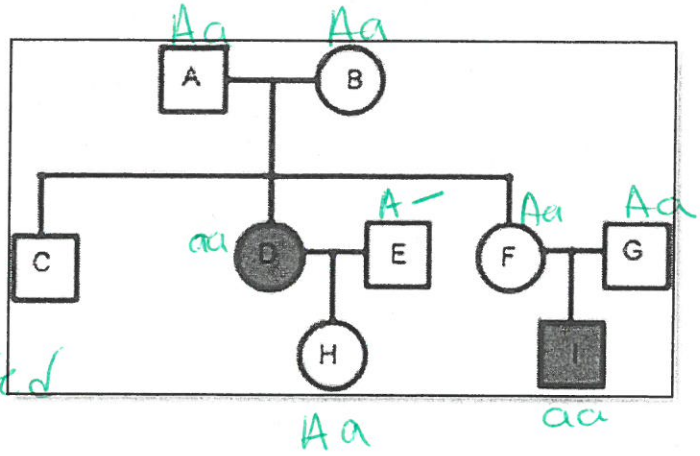
C. What gender(s) can carry recessive autosomal traits? male + female

D. What gender(s) can carry dominant autosomal traits?

no-one; dominant disorders = no carriers

7. Examine and analyze a pedigree chart. The pedigree shows the incidence of blue skin color in a family.

A. What are the genotypes of each of the family members?




B. Is the disorder dominant or recessive? _____

**How do you know? two unaffected parents have an affected child.

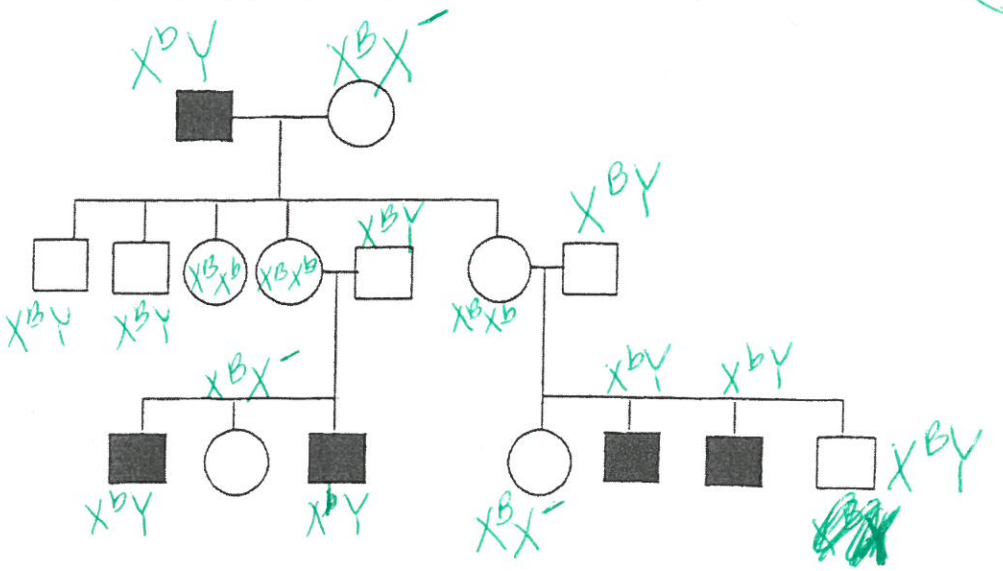
C. How many children did A & B have? 3

D. How many grandchildren do they have? 2

8. Determine the *pattern of inheritance* in the following pedigrees.

a. Write correct genotypes for **all** of the individuals.

b. Patterns of Inheritance: Autosomal Dominant, Autosomal Recessive, Sex-linked recessive



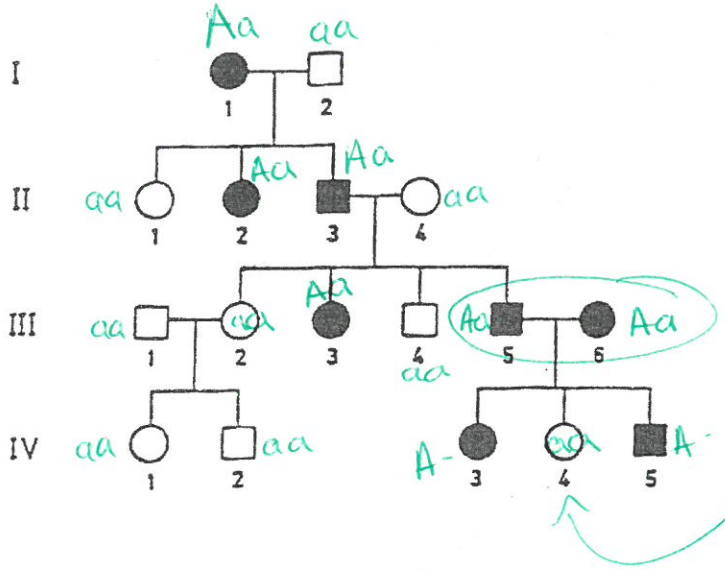

c. Why/where the other patterns of inheritance were eliminated?

only males are affected with the disorder

5 Determine the *pattern of inheritance* in the following pedigrees.

a. Write correct genotypes for **all** of the individuals.

b. Patterns of Inheritance: Autosomal Dominant, Autosomal Recessive, Sex-linked recessive



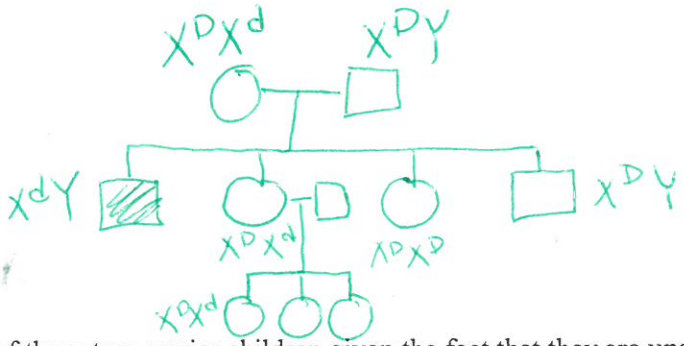

two affected parents have a normal unaffected daughter. If this was recessive, all their children would have to be affected.

c. Indicate why the other patterns of inheritance were eliminated.

6 Draw a pedigree for this family.

a. An unaffected man marries a woman who is a carrier for Duchenne Muscular Dystrophy, which is attributed to an Sex-linked gene. They have four children, one with Duchenne, one carrier daughter and a daughter and son who are unaffected. The child with Duchenne Muscular Dystrophy dies in childhood. The carrier daughter marries and has three children of her own, two of which are carriers and one of which is unaffected.

D = normal
d = Duchenne




b. What is the most likely sex of these two carrier children given the fact that they are unaffected by the X-linked gene?

females only females can carry sex-linked disorders (have 2 alleles)

9. **Draw your own pedigree.**

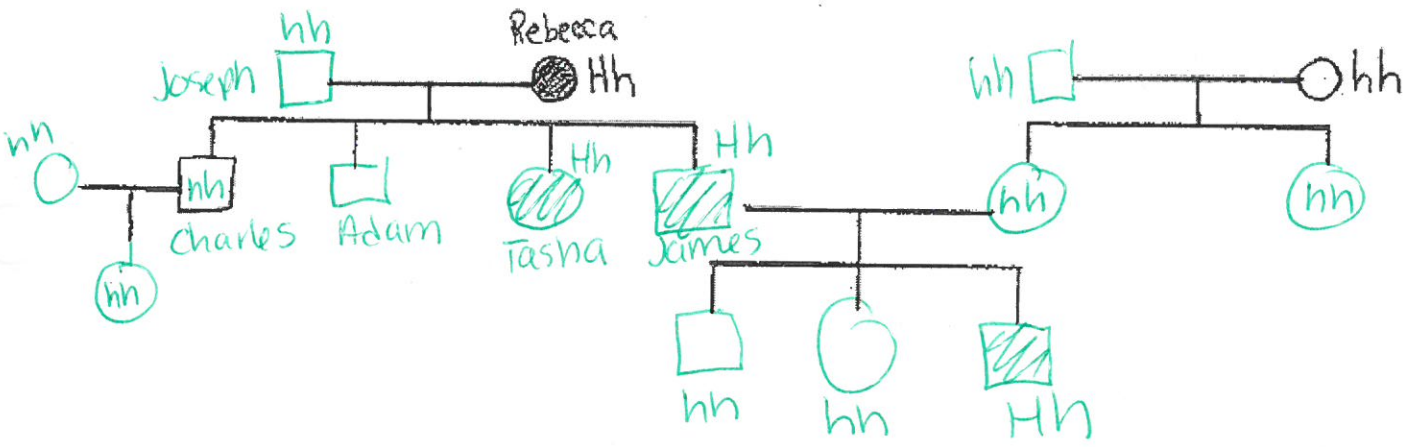
**Condition of Interest: Huntington's Disease** (also known as HD or Huntington's chorea)
*Huntington's disease is a neurodegenerative genetic disorder that affects muscle coordination and leads to cognitive decline and dementia.*

**Inheritance Pattern:** the allele for the normal "Huntingtin" protein is **h**; Huntington's disease is caused by **H** which codes for an abnormal form of the "Huntingtin" protein. Symptoms are more severe in homozygous individuals.

H = Huntington's
h = normal

Use **H** or **h** to represent the alleles.

A normal man (Joseph) marries a woman (Rebecca) who is heterozygous for HD and they have four children. Two of their sons (Adam and Charles) are born healthy without HD. Charles marries a woman without HD and they have a normal daughter. Joseph and Rebecca's daughter Tasha and their last son (James) both have HD. James marries a non-HD woman whose sister and parents also do not suffer from HD. James and his wife have three children - a normal boy, a normal girl, and a son with HD.




a. Patterns of Inheritance: Autosomal Dominant, Autosomal Recessive, Sex-linked recessive

b. Why/where the other patterns of inheritance were eliminated?

read the problem.