Name _____Period _____Lab # _____ Date _____

Human Karyotype & Pedigree Analysis

Introduction:

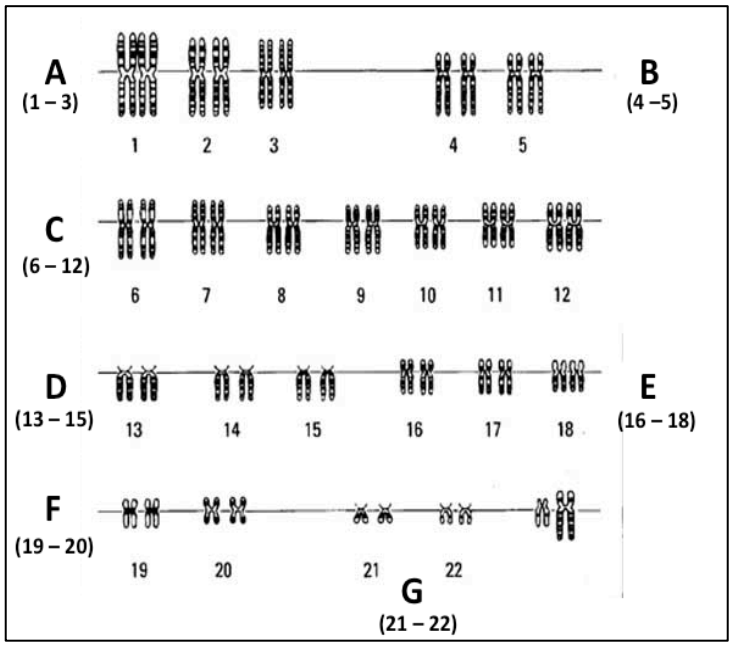
Each species has a characteristic number of chromosomes; for example, corn cells have 20 chromosomes, mouse cells have 40 chromosomes, and human cells have 46 chromosomes. In order to view the chromosomes so that they may be counted, a cell will be allowed to reproduce and colchicine is added to stop the cell division during metaphase. The resulting cells are placed in a hypotonic solution that causes the cell membranes to rupture. The chromosomes are stained and photographed. The chromosomes may then be cut out of the photograph and arranged by homologous pairs. The resulting display is called a karyotype.

Part 1: Typical Karyotype

The normal human karyotype is composed of **seven** groups of chromosomes (**A – G**) plus the **sex chromosomes** (**X and Y**). The chromosomes are grouped according to size, position of the centromere and the characteristic banding pattern. The first seven groups are called the autosomes while the larger X and smaller Y chromosomes are called the sex chromosomes.

Observe the normal human karyotype chart found in FIGURE I.

Figure I – Human Karyotype Chart



1. What is the *total number of chromosomes* found in this cell?

2. How many *autosomal chromosome pairs* are visible in the karyotype?

3. What is the *sex of the individual*?

4. Which pair is *not given a number*? Why?

5. In many karyotype charts, the **X chromosome** is placed in the **C group** and the **Y chromosome** at the end of **row 4**. Suggest a reason *why* this is done.

6. Could 2 individuals have the *same karyotype*? Explain why or why not.

Part 2: Abnormal Karyotype

Karyotypes can be used to identify a number of **chromosomal mutations**. Translocation defects, inversion mutations, addition and deletion mutations are all chromosome structure mutations. nondisjunction and polyploidy are chromosome number mutations.

Nondisjunction is the failure of chromosomes to separate during anaphase of cell division. This results in monosomy and trisomy conditions.

7. What is the difference between monosomy and trisomy?

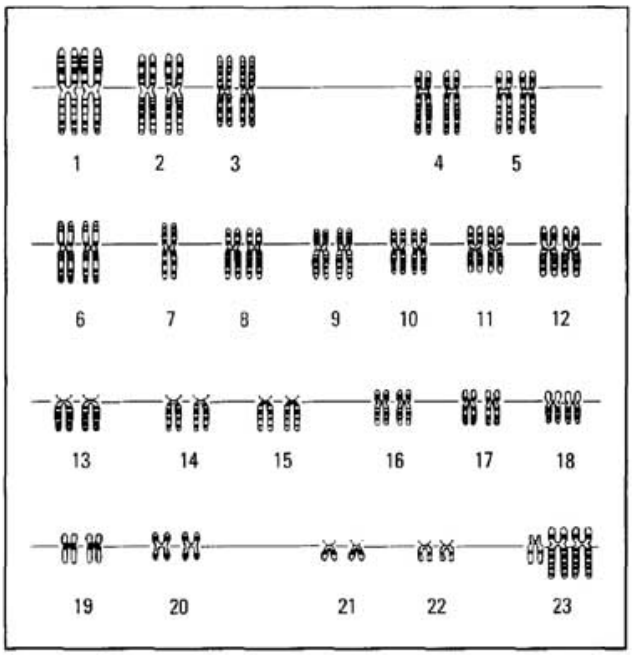
8. How could nondisjunction result in an individual with Down's Syndrome?

Sex Chromosome Disorders

Abnormal Chromosome Combination	*Medical Name*	*Characteristics of Individual*
XXY	Klinefelter syndrome	Male genitals, but with female secondary sex characteristics. Penis, scrotum, and testes are small; enlarges breasts. Sometimes timid and withdrawn; possible learning disabilities; sterile.
XO (no Y present; only one X)	Turner syndrome	Female external genitals; ovaries lacking; lack of menstruation, pubic hair and breast development. Stunted growth, with several body abnormalities. Sense of direction and spatial relationships may be abnormal; may be mentally retarded.
XO/XY	Mixed gonadal dysgenesis	May have female or male genitals, or a combination of the two. Usually no other bodily abnormalities, except may not mature sexually without treatment, and tend toward short body stature.
XYY	Supernumerary Y syndrome	Appearance of normal male. Tend to be tall in stature. May show some lack of control over impulsive behaviors. Usually average intelligence levels.
XXX	Triple-X syndrome	Appearance of normal female. Sometimes infertile. Occasional impairment of intelligence.
XX/XY	May be a true hermaphrodite	Variable. Have some combination of both ovarian and testicular tissues. Usually have uterus. External genitals may be distinctly masculine or feminine, or may be an ambiguous combination of both. At puberty, most experience breast enlargement, and the majority menstruate.

Observe the abnormal human karyotype charts found in FIGURE II & FIGURE III.

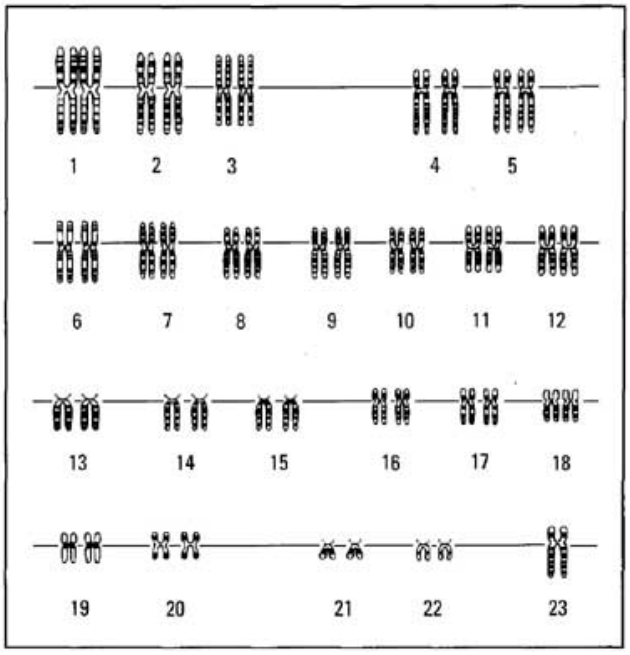
Figure II – Abnormal Human Karyotype



9. What abnormalities do you notice about the karyotype in Figure II?

10. What is the name of this disorder? What are the symptoms of it?

Figure III – Abnormal Human Karyotype



11. What abnormalities do you notice about the karyotype in Figure III?

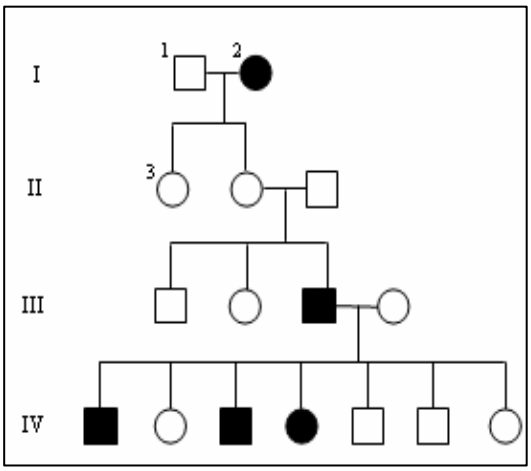
12. What is the name of this disorder? What are the symptoms of it?

Part 3: Human Pedigree

You can look at a pedigree and determine whether traits are dominant or recessive, and you can also tell if it is sex-linked or autosomal. A sex-linked recessive characteristic is determined by an allele that is carried only on the X-chromosomes. Since there is only one X in his genotype, XY, a male who carries a particular recessive allele on the X-chromosome will have the sex-linked condition. A female who carries a recessive allele in one X-chromosome will not show the condition if there is a dominant allele on her other X-chromosome. She will express the recessive condition only if she inherits two recessive alleles – one from each parent. Her chances of inheriting the condition are thus greatly reduced.

One sex-linked trait is hemophilia, a condition in which the blood does not clot properly. Most people who have hemophilia are men.

View the pedigree below and interpret the diagram to answer the questions.



13. In a pedigree, a square represents a **male**. *Darkened squares* represent males with hemophilia.
    a. How many males are there? _____
    b. How many males are affected? _____
14. A circle represents a **female**. *Darkened circles* represent females with hemophilia.
    a. How many females are there? _____
    b. How many females are affected? _____
15. A horizontal line connecting a circle and a square indicates a marriage.
    a. How many marriages are there? _____
16. 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 (*Row I*) have? _____
    b. How many children did the third couple (*Row III*) have? _____

Create Your Own Pedigree:

Albert and Betty are married. They both have normal vision. They had 2 daughters and then a son. Both daughters, Cassie & Debra, had normal vision and never had any children of their own. Their son, Elliot, was colorblind. Elliot married Fallon who also had normal vision and they had 2 of their own children, George and then Hailey. Hailey was colorblind but George was not.

Colorblindness is sex-linked recessive (X^1). Use the letters X, Y or X^1 for the genotype. Draw the pedigree below placing the genotypes for Albert, Betty, Elliott, Fallon and Hailey in their respective shapes.