**KARYOTYPING LAB**

Instructors will:

1. Answer any questions concerning last week's lab.

2. Define, explain what a karyotype is and what it is useful for.

3. Go over the terms and information on pages 1 and 2 of the student handout

Point the students to the stared text on page 2 of their karyotyping

handout which gives them clues to the hair and eye color used in this

exercise-otherwise they may get confused.

CORRECTIONS to the key to the traits: The sex-linked **recessive** traits

show up as XC (color blindness), XD (Duchene muscular dystrophy), and XH (hemophilia), and should have been denoted by lower case letters.

4. Explain the procedure for part I of the lab and assign envelopes (gametes).

Inform the students to leave out the display of karyotypes at their table.

Have students go around the lab to at least 3 other groups’ karyotypes and read the results. Have them record the genetics of each baby by the number on the BAG. This will give them practice in reading the traits for the lab practical on karyotypes.

5. Point out the resource materials in lab for part II (poster of traits/disorders of

human chromosomes, use index of textbooks to look up the nature of the genetic traits/disorders). Pay particular attention to autosomal or sex-linked nature of the disorder. You **may** allow 5 bonus points for Part II due the week following this initial lab. Use the questions in Part II as a guide for the report.

6. Monitor student progress, explain the process again if needed.

7. The last 15 minutes of lab have each group report on their "baby".

8. MAKE SURE STUDENTS RETURN ORIGINAL SETS OF CHROMOSOMES

TO THEIR CORRECT ENVELOPES BEFORE LEAVING CLASS. This will ensure that the next class has the proper number and kinds of chromosomes when they start the lab.

9. Make assignment for next lab on **forensic science.**

Give out the handout.

Students will:

1. Learn the terms and information on pages 1 and 2 of the handout.

2. Understand the process of karyotyping and its importance/social impact.

3. Complete the lab report parts I and II.

4. Report to the class their results.

5. Replace chromosome sets in correct envelopes and return to the instructor.

Items needed for this lab:

envelopes each containing 1 set of male chromosomes (Numbers match key)

envelopes each containing 1 set of female chromosomes (Numbers match key)

plastic coated keys for student use in lab

Answers to the Karyotyping Worksheet:

1a. 23

1b. 23

1c. 46

1d. Most students should answer 46, couples #6 and #13 should answer 47; Couple #5 should answer 45.

2a. meiosis

2b. meiosis

3a. XY= boy, XX = girl.

3b. For most couples, the sex of the baby will be obvious. Baby #5 will only have one X chromosome, resulting in a female with Turner’s Syndrome.. Baby #13 will have 2 X chromosomes and one Y, resulting in a male with Klinefelter’s Syndrome.

4. Chromosome number 1 is the longest. Successive chromosomes generally are numbered in successive order by descending size, although some are the same size. Hence the banding patterns are also used to arrange the chromosomes.

5 – 9. Answers will vary. Refer to instructor’s key to the babies.

**Karyotyping With a Plain**

**English Map of Human Chromosomes:**

Student Guide

**GLOSSARY**

**Allele** - one of two forms of a gene that exist at a specific gene site. One of these forms can be

dominant, such as green eyes (G) and the other form can be recessive, such as blue eyes (g).

**Chromosome** - a cellular structure composed of a DNA coiled around proteins. Chromosomes contain

inherited information.

**Diploid cells** - cells with two chromosomes from each homologous pair in humans, body cells are

diploid. Gamete - sex cells (egg or sperm).

**Gene** - a unit of heredity. Many genes consist of a sequence of DNA nucleotides that determine the

amino acid sequence of a protein.

**Haploid cells** - cells with one chromosome from each homologous pair. In humans, gametes are

haploid.

**Heterozygous** - instance where two genes that control a specific trait are different - for

example, a human with a gene for green eyes (G) and a gene for blue eyes (g).

**Homologous** - two chromosomes that share the same alleles.

**Homozygous** - instance where both genes for a specific trait are the same - for example, a human with two genes for green eyes (GG).

**Human Genome Project** - a multi-national, 15-year, $3 billion project to detect the specific locations of the genes that control human traits. There are between 50,000 and 100,000 genes in a haploid human genome (23 chromosomes). The goal of the Human Genome Project is to identify and map all the human genes and eventually sequence the nucleotides in their DNA.

**Locus** - a specific location on a chromosome where a gene can be found. Loci is the plural of locus.

**Mendel's Law of Unit Characteristics** - law which states that all traits are controlled by two genes, one from the egg and one from the sperm.

**Mendel's Law of Dominance** - law which states that some genes are dominant over other genes

(known as recessive genes), thereby preventing the expression of the recessive genes.

**Mendel's Law of Segregation** - law which states that during meiosis only one gene from each gene pair is passed on to future offspring through the sperm of egg.

**Mendel's Law of Independent Assortment** - law which states that during meiosis all gene pairs are assorted independently, resulting in a variety of gene combinations, and subsequently, genetic

traits.

**Multiple alleles** - several forms of a gene. The ABO blood group is an example where humans can

possess one of three allele forms.

**Non-disjunction** - failure of a chromosome pair to separate during meiosis, resulting in extra or missing chromosomes.

**Trisomy** - a genetic condition caused by non-disjunction of chromosomes during meiosis. The result is

three homologous chromosomes with the same alleles instead of the normal pair.

**BACKGROUND INFORMATION**

For the last 25 years karyotyping has been used to detect chromosome abnormalities in humans. The

chromosomes are usually obtained from blood samples. Since the white blood cells contain large nuclei, these cells usually contain the best source of human chromosomes. Through a series of staining procedures, the cells with the largest visible chromosomes are isolated, photographically enlarged, and then cut out and arranged into chromosome pairs. Highly trained scientists and doctors analyze the results to determine if there are any variations in the shape, length, or number of chromosomes. Over the years, this technique has been used to detect, and in some cases correct or treat, an increasing number of genetic diseases.

A related technique called amniocentesis now permits the detection of abnormal chromosome numbers in a fetus as early as the sixteenth week of pregnancy. In this procedure, the tip of a syringe is inserted through the mother's abdominal wall and uterus into the fluid-filled amniotic sac surrounding the fetus. Dead skin cells and cells from the respiratory passage that enter the amniotic fluid are sucked into the syringe. These cells can then be karyotyped and analyzed for chromosomal abnormalities.

Specific groups of chromosomal abnormalities are accounted for by the occurrence of non-disjunction. Nondisjunction is the failure of chromosomes to appropriately separate during meiosis at Anaphase I or Anaphase II. Non-disjunction during the first meiotic division results in the abnormality of all gametes formed. Half will contain the paternal and maternal chromosomal haploids, giving rise to trisomic zygotes; and half will contain no chromosomes at all, producing monosomic zygotes. Non-disjunction in the second meiotic division can produce half of the gametes normally. In the other half, however there will be some gametes lacking chromosomes, again creating monsomic zygotes, as well as gametes which contain two copies of a chromosome, either paternal or maternal, producing trisomic zygotes. The consequences of non-disjunction are serious, resulting in Down's Syndrome, Turner's Syndrome, and Klinefelter's Syndrome, among others.

Karyotyping can also be useful in evaluating the personal traits of babies. Characteristics such as blood type, hair color and eye color can be determined by the presence of a gene on a specific chromosome. Many times the paternal gene can contribute a different quality trait than the maternal gene. For example, the paternal gene may indicate brown eyes while the maternal gene indicates blue eyes. One of these gene traits must take precedence over the other and be expressed in the baby. The gene trait which takes precedence is called the dominant trait; the other is recessive. The dominant gene is denoted by a capital letter, and the recessive gene is denoted by a lower case letter. Although a baby may outwardly display the dominant trait, It can still genotypically contain the recessive gene as well. However, for the recessive gene to be expressed, it is necessary to have both parents give recessive genes.

Refer to your Map of the Human Chromosomes. A key to the symbols included on this map is listed on the following page. (These same symbols will apply to the chromosomes you will receive as part of this lab, in which you and your partner will join to produce a "baby.") On your map, you will note that the gene for brown eyes (B) is not listed. While it is known that brown eyes are dominant over blue and green eyes, the locus of the gene for brown eyes has not been found. Due to this fact, in the following lab, when an indication of eye color is absent in the chromosomes, it can be assumed that the dominant brown eye gene is present. Additionally, if the indication of eye color is absent in one parent, but present in the other, the dominant trait of brown eyes will take precedence. The gene for brown hair (B) is also dominant over the gene for red (R) hair. Red or blond hair is only expressed when brown hair is not coded on Chromosome #19.

As for many of the other traits noted on your map, keep in mind that all people have genes for all traits.

However for the sake of simplicity, only the abnormal genes are shown. In fact, each chromosome contains between 900 and 4,000 genes. This map shows only a few of the genes on each chromosome.

In this lab, you and your partner will join chromosomes, to produce a "baby." For each baby "produced" during the lab you will need to determine eye color, hair color, and blood type. Each baby will also have and carry a genetically-linked disease, which you will need to determine. Keep in mind that this is done for study purposes only. In reality, very few people actually have a genetic disease. This is because even through most people carry at least one recessive gene that, if expressed, would cause a serious genetic disease, the recessive gene is masked by a normal dominant gene. Also, remember that genes contain information for "putting together a person," It is only when a gene malfunctions that you get a genetic disease.

**KEY TO GENE SYMBOLS**

A Blood Type\* IA Hemophilia XH

Albinism a Huntington's Disease H

B Blood Type\* IB Marfan Syndrome M

Blond Hair r and b Rh type (negative) Rh-

Blue Eyes g or b O Blood Type\* Io

Brown Eyes\*\* B Phenylketonuria (PKU) p

Brown Hair B Rh type (positive) Rh+

Color Blindness XC Red Hair R

Cystic Fibrosis c Retinoblastoma\*\*\* R

Duchenne Muscular Dystrophy XD Sickle Cell Anemia s

Dwarfism D Tay-Sach's Disease t

Green Eyes G

\* Blood type of which A, B, and O are three different alleles; thus the notations IA, IB, and Io.

\*\* The gene for brown eyes has not yet been located

\*\*\* Chromosome 13

NOTE: Capital letters for diseases mean that the disease will be expressed with only one allele. They

are dominant diseases.

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**PROCEDURE**

**Part 1: In-class Lab**

To better understand "What makes you unique?", you will assume the role of mother or father and

contribute one set of chromosomes to your "offspring." Your partner will contribute a second set of

chromosomes to your "offspring." In this way, you will simulate the events that contributed to the

formation of the unique individual that is you!

1. The envelope that you received contains paternal (male) or maternal (female) chromosomes. If

your chromosomes are pink, you are the mother. If your chromosomes are blue, you are the

father.

2. To begin karyotyping, spread out the contents of your envelope. Your partner should do the

same with the contents of his/her envelope. Mix the contents of the two envelopes together.

Now, match up the sets of chromosomes by size, banding, etc., until all the chromosomes have

been paired off. (NOTE: Occasionally, there may be a missing or an extra chromosome; in this

instance, you will have a chromosome that will not join with another to create a matching set.)

Next, place the chromosomes that are marked with an X or a Y together. (Again, note that in

some instances there may be an extra X or Y chromosome, or one of these chromosomes may

be missing.) Then arrange the remaining sets of chromosomes in order by size and banding.

Using your Map of the Human Chromosomes as a reference, number each set of chromosomes

in order, with the longest chromatids being #1. (Do not include the X and Y chromosomes in

your numbered sets.)

3. Answer questions 1-10 of the Student Worksheet.

**Part II: Research/Report**

After you complete the karyotyping lab you will discover that your "baby" has inherited a combination

of genes that result in a genetic disease. Your assignment is to find information on your baby's genetic

disease, and then write a report summarizing your findings. (Be sure to address the questions listed in

Part 11 of the Worksheet, and to list your sources of information.)

**Karyotyping Lab:**

**Student Worksheet**

**Results**

**Part I: In-class Lab**

Answer the following questions. If necessary, attach additional sheets of paper or continue writing

on the back side of these worksheets.

1a. Normally, how many chromosomes are found in a human sperm?

1b. Normally, how many chromosomes are found in a human egg?

1c. Normally, how many chromosomes are found in a human baby?

1d. How many chromosomes are found in the "baby" you created in this lab?

2a. What form of cell division creates sperm?

2b. What form of cell division creates the egg?

3a. Note the two chromosomes that are not numbered. (Refer to your map for chromosome

numbers.) If these chromosomes are \_\_\_\_\_\_\_\_\_\_ and \_\_\_\_\_\_\_\_\_\_, your baby is a boy. If

they are \_\_\_\_\_\_\_\_\_\_ and \_\_\_\_\_\_\_\_\_\_,your baby is a girl. What is the sex of your baby?

3b. Is the sex of the baby readily obvious? \_\_\_\_\_\_\_\_\_\_ Occasionally, complications exist which

make it difficult to determine the sex of the baby. What do you think these complications

might be, and how could they occur? Explain your answer.

4. Note the arrangement of your baby's chromosomes. Study them carefully and compare them

to the chromosomes represented on the Human Chromosome map. What criterion is used to

arrange the chromosomes 1 through 22?

5. Note the genes that are found on your baby's chromosomes. Letters are assigned to

represent each genetic trait. If your baby has a combination of dominant gene, shown by a

capital letter, and a recessive gene, shown by a lower case letter, the dominant gene

prevents expression of the recessive trait. Based on this information, try to determine all of

your baby's genetic traits. List them below. (These should include any genetic diseases your

baby has, any genetic diseases your baby is a carrier of, your baby's blood type, your baby's

hair color, and your baby's eye color.)

6. What genetic disease or diseases is/are carried by the sperm?

7. What genetic disease or diseases is/are carried by the egg?

8. Based on the "Law of Dominance (see question 6), what genetic disease has your baby inherited,

if any?

9. Based on today's laboratory, write a paragraph explaining why you are genetically unique.

**Part II: Research/Report**

After you complete the karyotyping lab you will discover that your baby has inherited a combination of

genes that result in a genetic disease. You assignment is to find information on your baby's genetic

disease and then write a report summarizing your findings. Be sure to address all of the following items

in your report, and to cite your sources, and include a bibliography.

1. What is the name of your baby's genetic disease?

2. What are the symptoms of the disease?

3. When is the disease usually detected... at birth or later in life?

4. What is the frequency of carriers of this disease in the population?

5. What is the frequency of the disease in the population?

6. What is the mode of inheritance for this disease?

7. Is the disease autosomal (on the first 22 pairs of chromosomes) or is it on the X-linked or sex

chromosomes?

8. What chromosome has been determined to carry this gene (consult your "Human Chromosome

Map" or the "Human Chromosome Map" on the bulletin board).

9. What treatment, if any, is used for this disease? How does the treatment affect the disease?

10. What is the prognosis or outlook for your baby's recovery?

11. What future techniques might be used to correct or eliminate your baby's disease?

12. Discuss in detail what difficulties, if any, a parent would encounter in raising a child with this

disease?