HUMAN GENETICS

Introduction:

Multiple genes and environmental influences often complicate human inheritance patterns. Comparatively few inherited characteristics are simple and easily observed. However, some human genes produce variations in a characteristic that gives two distinct phenotypic expressions. The first activity of this laboratory is restricted to those genes that appear to be due to variations in a single gene locus.

Genes located on the X chromosome determine some inherited traits. These traits are referred to as sex-linked or X-linked traits. In the second activity of this lab we will observe a sex-linked or X-linked trait.

Exercise #1 – Common Observable Traits

In this activity, you will be the experimental subject, along with your classmates. You will attempt to determine your own phenotype and genotype for various traits and then compare genetic patterns.

Selected common traits are listed below and shown in the illustration. Determine your phenotype for each of these and attempt your genotype. Remember, when you express a dominant trait, it is not possible to individually determine if you are homozygous or heterozygous. Thus, use a dash (—) to represent the unknown second allele.

1. Determine your phenotype and possible genotype for each of the 8 described observable traits.

2. Record your results in the provided table. Circle your phenotype and fill in the possible genotype.

3. Place your phenotype on the CLASS RESULTS data table on the chalkboard.

4. Complete the CLASS RESULTS data table and answer the questions.

OBSERVABLE TRAITS

Widow's Peak — The hairline over the forehead dips downward in the mid-line, forming a definite point. The action of this gene dominant (W). Skip this trait if the gene for baldness has already been expressed.

Tongue Rolling — The ability to roll up the edges of the tongue to form a trough-like "U" is regarded as a dominant gene (R).

Ear Lobes — Human earlobes may attach directly onto the side of the head or may hang free. Assume the gene for the free condition is dominant (E) over that for the attached condition.

Hitch-Hiker's Thumb — Some individuals can bend the distal joint of the thumb back to sometimes a 45 degree angle between the two joints of the thumb. This condition is known as "distal hyper-extensibility" and is a condition (h).

Bent Little Finger — Evidence suggests that a dominant gene (B) influences the last joint of the little finger to bend inward toward the fourth finger. It is necessary to relax the hand a table to accurately observe this.

Long Palmar Muscle — This trait can be determined by feeling the tendons of the wrist when clenching the hand tightly. The long palmar muscle is present if three tendons are noticed (a recessive condition). When two (middle tendon lacking) are present, or when no muscle is present, a dominant (L) condition exists.

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Mid-Digital Hair — Some individuals have hair on the second (middle joint) of one or more fingers. Individuals with complete absence of mid-digital hair are recessive (m). Examine all fingers.

PTC Tasting — The ability to taste PTC paper (phenylthiocarbamide) appears to be influenced by a dominant gene (T). Tasting (T) is dominant to non-tasting (t). PTC is an antithyroid that prevents the thyroid gland from incorporating iodine into the thyroid hormone.

Name_____

Common Observable Traits Report Sheet

1. Personal Traits

Trait	Phenotype	Genotype
Widow's Peak	present – absent	_____
Tongue Rolling	roller – non-roller	_____
Ear Lobes	free – attached	_____
Hitch-Hiker's Thumb	present – absent	_____
Bent Little Finger	present – absent	_____
Long Palmer Muscle	present – absent	_____
Mid-Digital Finger	present – absent	_____
PTC Tasting	taster – non-taster	_____

2. Class Results

Traits	Phenotype	Class Total	Percent
Widow's Peak	____ Present	_____	_____
	____ Absent	_____	_____
Tongue Rolling	____ Roller	_____	_____
	___ Non-roller	_____	_____
Ear Lobes	____ Free	_____	_____
	____ Attached	_____	_____
Hitch-Hiker's Thumb	____ Present	_____	_____
	____ Absent	_____	_____
Bent Little Finger	____ Present	_____	_____
	____ Absent	_____	_____
Long Palmer Muscle	____ Present	_____	_____
	____ Absent	_____	_____
Mid-Digital Hair	____ Present	_____	_____
	____ Absent	_____	_____
PTC Tasting	____ Taster	_____	_____
	___ Non-taster	_____	_____

TOTAL STUDENTS _____

3. For EACH of the observed traits, which is most frequent in our class population, the dominant phenotype or the recessive phenotype?

4. Choose ONE trait observed in class and explain why one form of the trait is more frequent than the other form of the trait (i.e. why the dominant form is more frequent than the recessive form or vis-à-vis).

5. How do your phenotypes compare with the most frequent phenotypes in the class population?

Exercise #2 — Inheritance Related to Sex

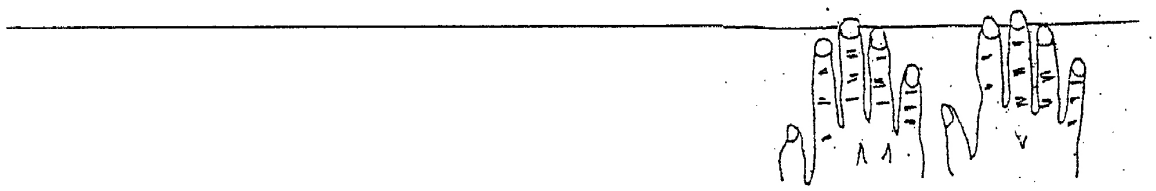
Gregor Mendel suggested that sex determination might be inherited in a similar manner as other traits he had studied. Previously (and even today) sex determination was attributed to such factors as the direction of the wind or the phases of the moon.

In the early part of the 20th century, several investigators showed that the X and Y chromosomes determined sex. Following these studies, a number of different sex chromosome associations were found in various organisms. In some cases, the female is found to be XO while the male is XX. In some organisms, such as fruit flies, the Y chromosome does not influence maleness or femaleness. A single X results in a male and the female is XX.

In humans the Y chromosome is necessary for maleness. The X chromosome is essential for "life" or viability. Although sex chromosomes influence maleness and femaleness, phenotypes may vary depending on interactions of the autosomal genes, embryonic development and environmental conditions.

The length of the second (index) finger in relation to the fourth (ring) finger is often assumed to be an inherited trait in humans (see the figure below). The index finger length trait appears to be X-linked or sex-linked recessive. In this activity you will attempt to determine if this X-linked recessive hypothesis holds true for our class population.

1. Place one of your hands on the line. The fourth (ring) finger should just touch the line.



2. Make a pencil mark at the uppermost tip of the second (index) finger. A short index finger would show the mark below the line. A mark on or above the line would show an index finger which is the same or longer.

3. In the table below, tabulate class results for males and females. According to the theory concerning this trait, the shorter index finger results from an X-linked recessive gene.

Name_____

Finger Length Results Report Sheet

Table 1 — FINGER LENGTH RESULTS

	Males		Females	
	Number	Percent	Number	Percent
Short Index Finger				
Long Index Finger				

1. Do the class results reveal the pattern of X-linked recessive inheritance?

2. Explain why a gene, which causes the index finger to be short, is dominant in males and recessive in females.

Name_____

Exercise #3 — Barr Bodies Report Sheet

In 40% to 80% of the somatic cells of normal females (XX) one of the X chromosomes is inactivated or turned off. An inactivated X chromosome can be visualized when a cell is stained. A stained cell shows a disk or rod shaped structure in the nucleus that represents the inactivated X chromosome. A stained inactivated X chromosome is called a Barr body, named after Murray Barr who first discovered them in 1949.

There was often some question in the past as to the sex identity of certain Olympic athletes. It has been demonstrated that Barr bodies can be accurately determined in female and male hair roots. This method has been adopted for precise testing of Olympic athletes.

A more simple technique of determining the sex chromosome complement of an individual is to examine squamous epithelial cells from scrapings of the cheek lining.

The Barr body of a somatic cell is a very small structure and usually disk or rod shaped. Normally it is pressed to the inner surface of the nuclear membrane (see figure below). In somatic cells of females, the number of Barr bodies is **one less** than the number of X chromosomes.

Observe the slide on the demonstration microscope. Identify a Barr body.

1. How many Barr bodies would be in the following individuals?

   XY _____ XX _____ XO _____ XXY _____ XXXX _____

2. Are there Barr bodies in the cells of a male transvestite?

3. Will an adult female who undergoes a sexual phenotype change by surgery (i.e. female to male) lose her sex identity or Barr bodies?

4. No individuals, either live births or spontaneously aborted fetuses, completely lacking the X chromosome have been found. Why do you think this is so?