Genetics Lab 5

Human Monogenetic Traits
(genetic individuality, probability of inheritance, pedigrees)

Development in humans is programmed by thousands of genes on our 23 pairs of chromosomes. Many physical traits such as overall size and shape are influence by multiple genes as well as well as by non-genetic, environmental factors. This is in contrast to the pea plant traits observed by Mendel, all of which were controlled by a single gene having 2 alternate alleles.

Some physical human traits appear to be monogenetic---that is, they are determined by variations of a gene at a single locus, and are not influenced significantly by environmental factors. Some of these traits (phenotypes) are considered *dominant* (expressed when the individual carries at least one dominant allele), while others are considered *recessive* (expressed only when no dominant allele is present). These traits are passed from parent to offspring in a Mendelian fashion, and can be followed as one might follow the transmission of a characteristic in pea plants (although we must use a pedigree approach).

Today's exercise (and some of next week's activities) involves observing human monogenetic traits in ourselves and in our lab partner. Once we have observed a number of phenotypes we will be able to use our data in a number of ways.

1. Predict/determine your genotype for each trait.

2. Note genetic individuality in a population (classmates).

3. Analyze the class data to predict probabilities of phenotypic combinations and to predict the frequency of genotypes in a population.

4. Create a limited pedigree/family tree for showing inheritance of one or more of the traits. (or of another trait which you know is controlled by a single gene).

**In lab 5 we collect data regarding our phenotypes and discuss how it can be used. In lab 6 we used combined class data in several applications of population genetics.

Procedure

1. Follow the instructions (on handout and verbal) for determining your blood type. Record your blood type in the handout. Blood type is controlled by a gene having 3 alleles, and gives us an example of co-dominance. We will discuss this briefly today.

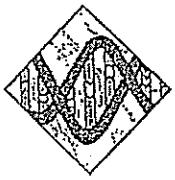
2. Determine your phenotype and that of your lab partner for as many of the genetic traits listed in your handouts as possible. Using the information given and your phenotype to try to predict your genotype. Pay attention to variation between yourself and other's at your lab table.

3. Record your phenotypes in a class data table for further analysis. We will collect class data to perform further analysis as a class next week.

4. Jot down ideas for completing a pedigree. Ask questions, and make a plan.

CHAPTER 11

HUMAN GENETICS



❖ INTRODUCTION

Human genetic traits can be used to illustrate a number of genetic examples. Such examples include complete dominance, incomplete dominance, codominance, and sex-linkage. McKusick's catalog lists many traits in humans that are genetic. The listing indicates the mode of inheritance of a particular characteristic, any complexities of inheritance, and the phenotype. In this laboratory exercise, you will use human genetic traits to further your understanding of heredity.

❖ PROCEDURE

1. Some Monogenetic Human Characteristics

Human heredity is complicated by the fact that many characteristics result from the action of polygenes and many are influenced by environmental factors. In this exercise, however, you will restrict yourself to some easily observed characteristics that appear to be monogenetic, determined by variations of a gene at a single locus, and are not subject to much environmental modification. A number of such characteristics are listed (see Figure 11.1). You should determine your phenotype for each of these and your genotype so far as is possible. When you have a dominant phenotype, you may have no way to know if you carry the recessive allele. In such cases you can use a dash (–) to represent the unknown second gene of your genotype. For instance, you probably do not have cystic fibrosis, a recessive characteristic. Hence, you know that you carry at least one of the dominant alleles (C), but you do not know whether the second allele of the pair is (C) or (c). You, therefore, represent your genotype as (C–).

Attached earlobes. In most people the earlobes hang free, but when a person is homozygous for a certain recessive gene (e), the earlobes are attached directly onto the side of the head so that there is no lobe hanging free. By means of a mirror or with the help of your classmates, determine your phenotype for this characteristic and tabulate it on the worksheets. You will find that there is considerable variation in the size and appearance of the lobes of those who have them. This is due to other genes, and you should concentrate only on the presence or absence of lobes in this study.

Widow's peak. In some people the hairline drops downward and forms a distinct point in the center of the forehead. This is known as widow's peak. It results from the action of a certain dominant gene (W). Determine your phenotype by examining your front hairline for a widow's peak or a continuous hairline. (You will have to skip this tabulation if a gene for baldness has had its effect at the front part of the head.)

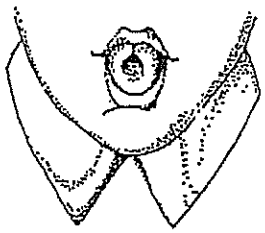
Tongue rolling. A dominant gene (R) gives some people the ability to roll the tongue into a distinct U-shape when the tongue is extended from the mouth. Others, who do not possess this gene, can do no more than produce a slight downward curve of the tongue when it is extended from the mouth. It has been suggested that this ability is not genetic. Try rolling your tongue and tabulate your phenotype for this characteristic.

Bent little finger. A dominant gene (B) causes the last joint of the little finger to bend inward toward the fourth finger. Lay both hands flat on the table, relax the muscles, and note whether you have a bent or a straight little finger.

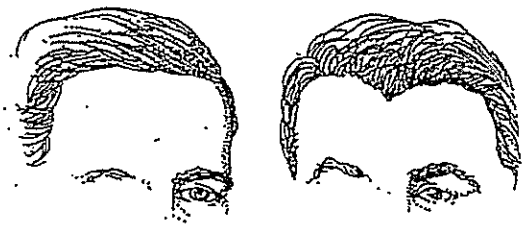
Hitchhiker's thumb. This characteristic, known more exactly as "distal hyperextensibility of the thumb," can be determined by bending the distal joint of the thumb back as far as possible. While there tends to be some degree of continuous variation, it will be found that certain persons can bend it back until there is almost (but not quite) a 90 degree angle between the two joints (see photograph). Evidence indicates that this characteristic is due to a recessive gene (h). There is some variation in expressivity, for occasionally it will be found that the ability is to be found in one thumb only. Also, it should be mentioned that there seems to be a five percent reduction in penetrance—

Figure I: Single Gene Traits

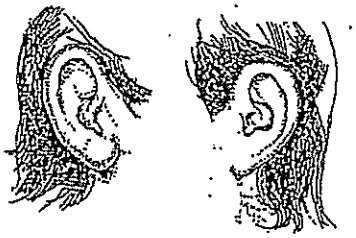
Tongue rolling



Widow's peak

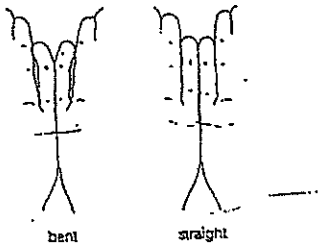


Ear lobe attachment

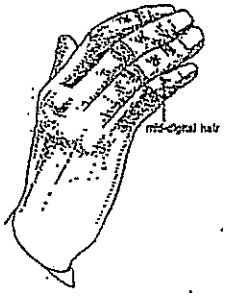


Hitchhiker's thumb

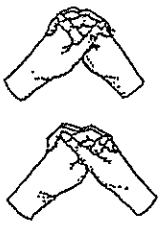
Bent little finger



Mid-digital hair



Hand clasping



JURE 11.1 Some inherited human characteristics. Upper left: widow's peak. Upper right: ability to roll tongue. Lower left: attached earlobe. Lower right: hitchhiker's thumb—note the angle of the distal joint.

that is, about one person in twenty who carries the gene will not express the characteristic. With these facts in mind, study your own thumbs and tabulate your findings.

Long palmar muscle. When a person is homozygous for a certain recessive gene (l), he or she has a long palmar muscle, which can be detected by examination of the tendons that run over the inside of the wrists. Clench your fist tightly and flex your hand. Now feel the tendons. If there are three, you have the long palmar muscle. If there are only two (the large middle one will be missing), you do not have this muscle. Examine both wrists, as it sometimes is present in one and not the other because of variations in the expression of the genes. If you find it in one or both wrists, you have the two recessive genes. If it is not present in either wrist, you have the dominant gene (L).

Pigmented iris of the eyes. When a person is homozygous for a certain recessive gene (p), there is no pigment in the front part of the eyes and a blue layer at the back of the iris shows through. This gives blue eyes. A dominant allele of this gene (P) causes pigment to be deposited in the front layer of the iris and masks the blue to a varying degree. Other genes determine the exact nature and density of this pigment and we have brown, hazel, violet, green, and other eye colors. We will concern ourselves here, however, only with the presence or absence of such pigment. Determine your phenotype for pigmented or unpigmented iris.

PTC tasting. Some persons detect a distinct bitter taste from a chemical, phenylthiocarbamide (PTC), while others do not taste it in weak concentrations. The closely related compound, phenylthiourea (PTU) may be substituted if PTC is not available. To prepare the test paper, dissolve about 0.5 percent of the chemical in acetone. Dip absorbent paper, such as paper toweling, into this solution until the paper is thoroughly wet with it. Then allow the paper to dry and it is ready for use. Cut the paper up into small strips, about 1/2 inch by 1 inch. A dominant gene (T) seems to confer the ability

to taste these chemicals at this concentration, while those homozygous for the recessive allele (t) lack this ability. Incidentally, this does not mean that nontasters of these chemicals have less acute taste sensations for other substances, so do not feel that you are missing something in life if you are a nontaster.

To test your tasting ability, place a paper strip impregnated with the chemical on your moist tongue and allow it to remain there for about ten seconds. If you are a taster, you will know it; if you have to wonder if you taste it or not, you are a nontaster.

Mid-digital hair. Some people have hair on the second (middle) joint of one or more of the fingers, while others do not. The complete absence of hair from all fingers is due to a recessive gene (m), and its presence is due to a dominant allele (M). There seem to be a number of these alleles that determine whether the hair shall grow on one, two, three, or four fingers. This hair may be very fine, and you should use a hand lens and look very carefully on all fingers before deciding whether this hair is absent from any one of your fingers.

Blood groups. You have probably already done the study of blood groups in the exercise on multiple alleles. You can tabulate your phenotype and possible genotype here as a part of this study. Use the symbol (A) for the A antigen, (A^B) for the B antigen, and (a) for the absence of either of these antigens. Or, you can use the symbols (I^A), (I^B), and (I^O), which are preferred by some authorities.

Second finger shorter than the fourth. This is a characteristic that appears to be sex-influenced and has already been studied in the exercise on heredity influenced by sex. The results of that study should be tabulated here, using the symbol (S^S) for the short second finger and the symbol (S^L) for the longer second finger. These should be tabulated according to sex since the frequency should vary among the sexes.

Interlocking fingers. When the fingers are interlocked, some people will almost invariably place the left thumb on top of the right and others will place the right over the left. Studies of family pedigrees suggest that the placing of the left over the right is due to a dominant gene (F), while the right thumb on top is due to a recessive (f); however, nongenetic factors may also be involved.

2. Genetic Individuality (Next week)

With so many people on the earth, it might seem that at times a person would be born with a gene constitution just like that of another person. This does not happen, however, except in the case of identical twins who start life as a single cell and are duplicated by an asexual method. There is no one on earth just like you from the standpoint of heredity, unless you have an identical twin. In this exercise, you will try to use the results of the previous study to show that, even when considering only a few traits, genetic individuality can be demonstrated.

Your instructor will ask the entire class to stand, and then choose one to call his or her characteristics one by one. The other members of the class will sit down as soon as they find themselves different from the one who is calling out the characteristics. As an example, suppose it is your turn and there are 30 people in the class, all standing. Let us say that you have free earlobes, and as you call out this trait, 6 persons sit down because they have attached earlobes. Then you call out widow's peak and 14 others sit down, leaving only 10 standing, including you. You are a tongue roller, and 5 more sit down when you call out this trait. You have a straight little finger and 3 sit down. You have no long palmar muscle and the last person sits down, leaving you standing alone—a genetically unique individual in your class after considering only five characteristics. When you consider the many thousands of genes and the numerous possible variations of each gene, you can understand why no two persons will get the same combination of genes, not even brothers and sisters, unless they happen to be identical twins. If your class is fortunate enough to include a pair of twins, it should be very interesting to note their similarities in these genetic traits, and it should be a good indication as to whether they are identical or fraternal. Time may not permit each member of the class to call out his or her characteristics, but several others may be chosen to illustrate human genetic individuality.

3. Analysis of Inherited Characteristics

Through a law of probability, you can predict the chances of two independent events happening simultaneously. This can be done by multiplying the chance of either happening separately. You can apply this law to some of the characteristics you have already studied. For instance, suppose you wanted to know what the chances were that a person would have both a widow's peak and attached earlobes. Assume that your results show that .30 (30%) of the members of the class have a widow's peak and .20 (20%) have attached earlobes. By multiplying these two fractions, you obtain .06 (6%), which is the chance that a person will show both of these characteristics. By actual count, then, you can determine if this is somewhere near the amount that you actually have in the class with these two characteristics. Of course, you must realize that the sample of students in a class is much too small to get any reliable ratios for the general population and considerable deviation from the expected may be found. Combining results from several classes will be more satisfactory from a statistical standpoint.

Choose two of the characteristics out of the group you have studied and determine the chance of each happening separately and then the chance of both happening together. Now count the number of people who show both of these characteristics and compare

(We will do this in lab Le)

this with the mathematical expectation. Use the standard error or chi-square to determine if the variation is within the bounds of chance deviation.

You can now use the Hardy-Weinberg law to determine the approximate number of students in the class who carry certain recessive genes in the heterozygous state. Such genes do not show, but the proportion of those who carry them can be determined by use of the figure for the number of people who are homozygous for the gene and who, therefore, express the characteristic. Consider attached earlobes as an illustration. Suppose there are 5 persons out of a class of 30 with attached earlobes. You know that these are homozygous for the recessive gene (ee). This is about 16 percent of the total. Now, how many of the remaining 25 are carriers of the gene (heterozygous), and how many are not carriers (homozygous)? If you allow p to represent the dominant gene and q to represent the recessive gene, then you can represent the three groups as follows:

(% homo. dom.)		(% hetero. dom.)		(% homo. rec.)
p^2	+	$2pq$	+	q^2

Now find the values of p and q, which will indicate the percentages of each gene in the class. You know that those with two recessive genes number 16 percent. Since q^2 is 16 percent, then the value of q would be the square root of .16. This gives you .40 or 40%. This means that about 40 percent of the genes in the class are recessive (e) and the balance are dominant (E). To get the value of p, subtract 40 percent from 100 percent since p plus q equals 100 percent. This gives 60% as the value of p. Since the number of persons in the class who are homozygous for the free earlobes is p^2, you square 60% and obtain 36%. If there are 30 people in the class, this is about 11 people. That leaves 14 remaining with free earlobes and these must be heterozygous. You can check on this by multiplying $2 \times .6 \times .4$ and you get 48%, which is about 14 people.

This may appear to be a complex procedure, but you can reduce it to several simple calculations. First, take the square root of the percentage of persons showing the recessive characteristic. Second, subtract this figure from 100 percent and square the answer to get the number of homozygous dominants. Those left in the class who show the dominant trait are heterozygous.

Choose one of the recessive characteristics already studied and determine the distribution of homozygous and heterozygous persons who express the dominant allele.

4. Making a Family Pedigree

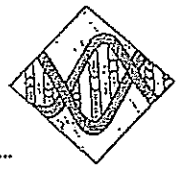
Collecting information about your pedigree may take several weeks and involve writing distant relatives you may not have contacted for some time, so this exercise should be planned for several weeks in advance. Choose some clearly defined characteristic that you possess and that is found in several other members of your family and relatives. You may choose one of the traits used for studies in this chapter, or better still, choose some other trait. You may get ideas of possibilities from genetics books. Get information from your immediate family and as many others in the direct line as possible. This would include aunts, uncles, grandparents, great grandparents, great uncles and aunts, and so on. Cousins may be omitted unless they have some particular significance. Should you have children, or any of your brothers and sisters have children, these should be included provided they are old enough to show the trait you have selected.

There will probably be some in the class who cannot get sufficient information to make a pedigree study feasible. The instructor may permit them to obtain a pedigree from some other source.

After the information has been assembled, make a pedigree chart. Males are represented as squares and females as circles. Marriage is indicated by a horizontal line, and children are shown on a horizontal line below the parents. Refer to your textbook or other books for examples, including the method of showing twins, second marriages, and so forth. Draw an arrow to the square or circle that indicates you; you are the propositus. Devise a system to indicate the members of the pedigree that show the trait you are studying. If it is a discontinuous trait, the simplest way to do this is to shade all squares or circles that show the trait and leave the others blank. Should it be an apparent intermediate trait, you can include a lighter degree of shading for those showing the trait to a lesser degree, or you can shade only one-half of the square or circle. For quantitative traits with continuous variation, you can devise various degrees of shading to indicate degree of expression of the trait. Be sure to include a key to indicate what the different shades represent. In some cases you may find out that you have a relative who cannot be contacted and you do not know whether he or she shows the trait or not. In such cases, put a question mark in the square or circle.

After the chart has been completed, analyze it and try to determine the probable means of inheritance—whether it is dominant, recessive, intermediate, codominant, sex-linked, sex-influenced, autosomal, polygenic, or if multiple alleles are involved. Also, you might consider the possibilities of incomplete penetrance and variable expressivity. Once you have decided on a plausible explanation, devise gene symbols for the trait and put the possible genotype beneath each member of the pedigree. Base your conclusions solely on the pedigree you have prepared, not on any information about the trait you may have obtained from books or other sources. Sometimes different genes affect the same trait and you may be dealing with one that is different from one that may have been a part of another study. A single pedigree is not sufficient to draw definite conclusions, but you can get some indication of the method of inheritance of the trait you have chosen.

CHAPTER 11



HUMAN GENETICS

Name: _____

Lab Section: _____

Date: _____

1. Some Monogenetic Human Characteristics

In the place below, tabulate your phenotype for each of the human characteristics studied, also give your possible genotype and the total number of students in the class that show each characteristic.

	Check Your Phenotype	Give Your Genotype	Number of Each in the Class (We will use a spreadsheet)
Attached earlobes	_____	_____	_____
Free earlobes	_____		_____
Widow's peak	_____	_____	_____
No widow's peak	_____		_____
Can roll tongue	_____	_____	_____
Cannot roll tongue	_____		_____
Bent little finger	_____	_____	_____
Straight little finger	_____		_____
Hitchhiker's thumb	_____	_____	_____
No hitchhiker's thumb	_____		_____
Long palmar muscle	_____	_____	_____
No long palmar muscle	_____		_____
Pigmented iris (can see color)	_____	_____	_____
No pigmented iris (blue)	_____		_____
PTC taster	_____	_____	_____
Nontaster of PTC	_____		_____

	Phenotype	Genotype	Number
No mid-digital hair			
Mid-digital hair			
Fingers with mid-digital hair (indicate if the finger on both hands has mid-digital hair).	2nd		
	3rd		
	4th		
	5th		
When only one finger has hair, is it always the same finger?			
If so, which finger is it? (2nd, 3rd, 4th, 5th)			
If two fingers have hair, which two are usually involved?			
If three fingers have hair, which are usually involved?			
Blood groups:			
Type O			
Type A			
Type B			
Type AB			
Second finger shorter than the fourth—			
Male			
Female			
Second finger not shorter than the fourth—			
Male			
Female			
Interlocking fingers:			
Left thumb on top			
Right thumb on top			

4. Making a Family Pedigree

Use this page to diagram your family pedigree indicating those that express the trait you have chosen to analyze. Turn the page on its side if the pedigree fits best this way.

Week 2

2. Genetic Individuality

How many persons are there in the class? _____

Tabulate the number of persons who remain standing after you call out your phenotype for each characteristic. Include yourself in the number standing and continue to call out your characteristics until only you remain standing. If time does not permit you to call out your phenotypes, then record the data of one of your lab mates who does have the opportunity to do this.

	Persons Standing
1. Condition of earlobes	_____
2. Widow's peak	_____
3. Tongue-rolling ability	_____
4. Bent little finger	_____
5. Hitchhiker's thumb	_____
6. Long palmar muscle	_____
7. Pigmented iris of the eye	_____
8. PTC tasting	_____
9. Mid-digital hair	_____
10. Blood group	_____
11. Short second finger	_____
12. Interlocking fingers	_____

How many characteristics must be considered before you stand out as an individual in your class? _____

Record the number of characteristics that must be considered before the other members of the class stand out as an individual in the class. _____

What is the average number for the class? _____

What proportion of the above characteristics would you expect to find the same in identical twins? Explain. _____

_____

_____

Would you expect a class of genetics in Sweden to require about the same average number of characteristics to show genetic individuality? Explain. (Note: Sweden has remained a relatively unmixed race for many centuries while the United States and Canada have had immigration from diverse genetic groups.)

_____

_____

_____

_____

3. Analysis of Inherited Characteristics

List the two characteristics that have been chosen and determine the chance of each showing in any person picked at random.

Characteristic Number 1. _____

Number of persons in the class who show it. _____

Total number of persons in the class. _____

Percentage showing it. _____

Characteristic Number 2. _____

Number of persons in the class who show it. _____

Percentage showing it. _____

Consider the percentage showing a characteristic as the chance of any one person showing it. Now calculate the chance of any one person showing both characteristics. Show figure below.

According to the figures obtained above, how many in the class would be expected to show both of these characteristics? (Show results to one decimal place, e.g, 4.7 persons.) Show how you get your results below.

How many in the class actually show these two characteristics? _____

What is the deviation? _____

Does the deviation appear to be significant in light of the number of persons involved in your studies?

_____

Use the standard error or chi-square to determine if the variation is sufficient to be significant.

Does the result above indicate that there must be free assortment among the genes responsible for the characteristics chosen, or does there appear to be any positive or negative correlation between the two? Explain your answer and give any possible explanations for correlation if such appears to be the case.