Complex Traits Activity

INSTRUCTION MANUAL

ANT 2110 Introduction to Physical Anthropology
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Introduction

Human variation is complex. The simplest form of variation in a population comes from **polymorphisms**. A gene is said to be polymorphic if more than one **allele** (version of the gene) occupies that gene's locus within a population. For instance, the ABO blood system is polymorphic, having three different alleles – A, B, and O - in our population.

The genetic material that codes for the ABO blood system is entirely present at a single **locus**, or position on a chromosome. It is only the alleles an individual carries at this spot on chromosome 9 that determines their blood type.

However, most phenotypic traits are what we call **polygenic**, meaning that many different genes are involved when constructing the phenotype of an individual. Therefore, a phenotype can be the result of the polymorphisms present at many different loci across chromosomes.

Sometimes, a single gene can have multiple effects, known as **pleiotropy.** Humans have only about one-third of the genes expected for such a diverse species, so it is likely the case that many genes are pleiotropic in ways we are yet to understand.

Another reason we are so complex is that humans are **sexual reproducers**. This type of reproduction, as opposed to something like cloning, requires the input of genetic material from two parents. Because of **independent assortment** and **segregation**, it is completely random which allele at a locus is contained in a parent's gamete. Therefore, each offspring will represent a different mixture of the two parent genomes, guaranteeing that variation will always be present in the next generation.

There are also variations in how genes are expressed. Sometimes genes are Mendelian, meaning that they have a dominant/recessive determinant to their expression. For some genes, you always express what either your mother or father contributed, while for other genes it is completely random which parent's allele gets expressed.

Lastly, don't forget the role of the **environment**. We do not perfectly produce phenotypes based on our genotypes; the environment can have an affect on how these genes function. For instance, your genes give a framework for how tall you will be, but it is nutrition and other environmental factors during development that will affect whether you reach your full potential, or whether your growth gets stunted at any given period.

For this activity, we will be calculating two traits.

Below is a list of 6 loci involved in these traits. The second column lists how many polymorphisms (or alleles) exist at that locus in the population's gene pool. The third column lists which trait the gene at that locus affects.

Locus number	Alleles	Phenotypic affects
1	3	Blood type
2	6	Melanin production
3	6	Melanin production
4	6	Melanin production
5	20	Melanin production

In order to remind you that locus 1 affects a different trait than the rest of the loci, it is shaded gray on all of the tables.

To start, using **TABLE 1** of the data notebook, you will determine the genotype of a random female in the population.

The locus number as well as the number of polymorphisms (alleles) at that site are already there in the first two columns.

To establish this female's genotype, you will use your dice.

If the listed number of alleles is "6," then use the six-sided die known as a d6. The associated values for each result are the value on the die, therefore 1=1, 2=2, etc.

If the listed number of alleles is "3", use the d6 and record a value from 1-3 based on the following roll result: 1-2 = 1; 3-4 = 2; and 5-6=3 (this is ONLY for blood type – locus 1, the shaded row).

If the listed number of alleles is "20," then use the 20-sided die known as a d20. This particular locus can have a greater affect on phenotype than the other polygenic loci.

Remember, an individual's genome **contains a copy of each gene from their mother (their maternal genome) and a copy of each gene from their father (paternal genome).** Therefore, in order to create a hypothetical genome for this individual, we need to determine TWO values for each locus.

TABLE 1a of the data notebook: Identify whether you will use the d6 or d20 for each locus. You will then roll this *twice* for each locus to determine both the maternal and paternal contribution.

Example: At locus 1, I roll the d6 and get a 3. I write down "3" as the result for the maternal genome and then because of the rule written above (1-2 = 1; 3-4 = 2; and 5-6=3) I write "2" for the value. I then roll the d6 AGAIN and get 5. I write down "5" in that same row but different column for the result for the paternal genome and then a "3" for the value. It looks like this:

		Maternal Genome		**Paternal** Genome	
Locus #	Alleles	Result	Value	Result	Value
1	3	**3**	**2**	**5**	**3**

For the other loci that have 6 alleles, the result and value will be the same number.

2	6	**1**	**1**	**4**	**4**
3	6	**6**	**6**	**2**	**2**

Next, you will determine what your female's genes say she should look like. This is part of her **phenotype.**

The question is, for each locus, which of the parent's genes are going to be expressed? There are different ways in which gene expression is determined.

We discussed **Mendelian** dominance in class. If a gene is listed as "**Co-Dom**" then every dominant allele will be expressed because of the principle of co-dominance. Therefore a copy of that allele in the genotype will mean that it is expressed, no matter the value of the other allele. Our trait is the ABO blood system where alleles A and B are co-dominant, while O is recessive.

Sometimes it is completely **random** whether you express what you received from your mother or father, and this is the rule we will use for the loci that affect melanin production.

In **TABLE 1b** of the data notebook, **use your results from TABLE 1a.**

First, in the row for locus 1, each of the three possible results is paired with an A, B, or O. Circle the two results you obtained in **Table 1a** along with its corresponding letter. Write down dominant alleles in the last column (A &/or B).

E*xample: For locus 1, my female in the example above received allele 2 from her mom and 3 from her dad. Using the information in the table, I can circle the corresponding ABO proteins and determine her genotype by writing down only the dominant alleles*

Locus #	Rule		Result
1	Co-Dom	1=A, 2=B, 3=O	B

Then, for the other loci, you will flip the coin to randomly determine which of the two alleles will be expressed. Use this rule: **Heads=maternal, Tails=paternal.** Write down the corresponding parent's allele from your **Table 1a.**

Locus #	Rule	H/T	Result
2	Random	T	4

In **Box 1** total the values for alleles 2-5 to determine the overall contributions of the genome to melanin production

In **TABLE 2**, you will use your coin to see which of your female's alleles end up in two of her different gametes, or eggs, after meiosis. Each gamete contains half of her genetic information, and which alleles are present in each are random due to the principles of independent assortment and segregation.

Use the genotypic information from **TABLE 1a** to fill out **TABLE 2** to determine the genetic information in the gametes. For each locus, you will flip your coin. Using the rule **Heads = maternal, Tails = paternal**, fill in the allele from the corresponding genome. You will do this *twice* for each locus in order to create two gametes.

Example: For locus 1, I flip my coin and get heads, which tells me that the allele for locus 1 of egg 1 comes from the maternal genome. Looking back at my information in TABLE 1a I see the value (not the result) shows the mother contributed a "2" at this locus. I enter that value in TABLE 2. I then flip the coin again to see what allele for locus 1 goes in the second gamete. I get tails this time, which is from the paternal genome. I see it is a "3" from the father in my TABLE 1a and write down that value for egg 2.

	Egg 1			Egg 2		
Locus #	Result H/T	Maternal/ Paternal	Value	Result H/T	Maternal/ Paternal	Value
1	H	Maternal	2	T	Paternal	3

Your female has now found a male suitor, and listed below is the genetic makeup of six of his gametes, or sperm.

Locus #	Sperm 1	Sperm 2	Sperm 3	Sperm 4	Sperm 5	Sperm 6
1	1	1	2	2	1	1
2	1	1	5	1	5	5
3	2	2	3	2	2	3
4	5	5	3	3	3	3
5	4	18	4	18	4	18

In **TABLE 3a** you will generate the genotype of their baby.

First you need to determine which sperm fertilizes which egg. To choose the egg, flip a coin to see which of your female's gametes is released. **Heads = Egg 1; Tails = Egg 2.** To determine which of the father's sperm is the one to fertilize that egg, use your d6**; a roll of 1 = Sperm 1, etc**.

Copy the corresponding genetic info for the selected egg from **TABLE 2** into **TABLE 3**. And copy the corresponding genetic info for the selected sperm from the table directly above to **TABLE 3**.

Example: I flip the coin and get Heads. I write "1" down for the egg. I roll the d6 and get 3. I write "3" down for the sperm. I look at TABLE 2 and copy down the information in Egg 1 for each locus in TABLE 3a. I look at the table above and copy down the information for Sperm 3 in TABLE 3a.

Locus #	Egg # 1	Sperm # 3
1	2	2

In **TABLE 3b** you will calculate the offspring's phenotype, just as you did for their mother in **TABLE 1b.** The same rules still apply.

Next, let's determine the final phenotype of this offspring taking into consideration some environmental factors. Because the environment affects skin color more than it affects blood type, skin color is considered a less heritable trait.

TABLE 4 lists two environmental factors that can affect skin color. Roll your d20 for each environmental factor. Using the chart below, record the corresponding adjustment number.

d20 dice roll	Phenotype adjustment
1-4	-2
5-8	-1
9-12	0
13-16	+1
17-20	+2

Example: For the first environmental factor, I roll a 16. According to the chart directly above, the corresponding adjustment is +1. I record both on TABLE 4.

SKIN COLOR *Does the adult lifestyle include:*	*d20 roll*	*adjust*
Lots of time outdoors in strong sunlight?	**16**	**+1**

Total the "adjustments" for each individual; mother in Box 3, offspring in Box 4.

Skin color can also be affected by genetic disorders. One in particular, phenylketonuria (PKU) is caused by an individual carrying two copies of a mutation that causes the metabolism of the amino acid phenylalanine to be inhibited This mutation has **pleiotropic** effects (affects many traits) most notably cognitive function and melanin synthesis. The likelihood of a random individual having PKU is 1/8000 (= 0.000125). This probability is the same as rolling the same number on the d20 three times (1/20 * 1/20 * 1/20 = 0.00125). The mother and father do not have PKU, but we will "screen" the offspring for the disorder.

In **Table 5** roll the d20 three times. If you roll the same result each time, check the box for "yes" regarding the offspring having PKU. If not, check "no."

Lastly, we will calculate final phenotypes.

In **Table 6a**, total your results from Box 1 and Box 3 in order to determine the mother's final phenotype.

In **Table 6b**, total your results from Box 2 and Box 4 and take into consideration your results from the PKU screening to determine the offspring's final phenotype.

If your offspring has PKU, take your total and multiply it by 0.10 to indicate that their severe deficit in melanin production. Your result will be a small number indicating that the individual has a nearly complete loss of melanin.

Compare your results to the color scale provided in the data notebook to determine how your individuals compare in skin color to the range of variation that exists for humans. Please note that 0 is a complete lack of pigmentation while 40 represents the darkest skin recorded for any human.

****Answer the discussion questions on the last page of the data notebook****