Variance Components: Phenotypic, Environmental and Genetic

You should keep in mind that the Simplified Model for Polygenic Traits presented above is <u>very</u> simplified. In many cases, polygenic or quantitative traits are influenced by environmental factors. For example, skin color in humans is determined by several gene loci **and** by environmental factors. Prolonged exposure to the sun can darken the basic skin color. In other words, environmental effects can alter the phenotype of an individual.

Our basic model for quantitative genetics can be described by a simple equation:

$$\mathbf{P}_{\mathbf{i}} = \boldsymbol{\mu} + \mathbf{G}_{\mathbf{i}} + \mathbf{E}_{\mathbf{i}}$$

where P_i is the phenotypic value of the ith individual in a population, μ is the mean of the population, G_i is the deviation of the ith individual from the population mean due to genetic factors, and E_i is the deviation of the ith individual from the population mean due to environmental factors.

The equation above implies that the variance in phenotypic value (the phenotypic variance, V_P) can be partitioned into components representing contributions from both genetic factors and environmental factors. Consequently, the total variation in skin color (or any other quantitative trait) is a sum of variation due to genetic factors (V_G) plus variation induced by environmental factors (V_E) plus variation arising from genotype-by-environment interactions (V_{GE}):

 $\mathbf{V}_{\mathbf{P}} = \mathbf{V}_{\mathbf{G}} + \mathbf{V}_{\mathbf{E}} + \mathbf{V}_{\mathbf{GE}}$ where $\mathbf{V}_{\mathbf{P}}$ = phenotypic variance

Because V_{GE} is often not easy to measure, it is often assumed to be negligible and is sometimes ignored:

$$\mathbf{V}_{\mathbf{P}} = \mathbf{V}_{\mathbf{G}} + \mathbf{V}_{\mathbf{E}}$$

The variance components in the equation above indicate that we must use statistical techniques to study these quantitative traits. The most important statistics are the mean and variance:

Mean =
$$\overline{\mathbf{X}} = \frac{\sum_{i} X_{i}}{n}$$
 Variance = $s^{2} = \frac{\sum_{i} (X_{i} - X)}{n - 1}$

The mean (or average) is simply the sum of all the measurements divided by the sample size, n. We use n-1 instead of n as a correction for sampling error. When n is large, the correction is minor. The variance is simply the sum of the squared deviations of each measurement from the mean divided by n-1 (i.e. "the average squared deviation").

One of the most important goals of quantitative genetics is the estimation of the variance components in the equation above. In other words, how much of the variance in phenotypes is attributable to genetic factors (V_G), and how much is attributable to environmental effects (V_E)? More specifically, quantitative geneticists are interested in the **heritability** of a trait. The heritability of a trait is defined as the proportion of the phenotypic variance that is attributable to genetic factors. This is what is called the **broad-sense heritability:**

$$\mathbf{h}_{\mathrm{B}}^{2} = \frac{\mathbf{V}_{\mathrm{G}}}{\mathbf{V}_{\mathrm{P}}}$$

Broad-sense heritability is so called because it includes the effects of all genetic factors including the additive effects of alleles at all the loci influencing the trait, as well as effects due to dominance relationships between alleles and interactions among gene loci. In other words,

$$\mathbf{V}_{\mathbf{G}} = \mathbf{V}_{\mathbf{A}} + \mathbf{V}_{\mathbf{D}} + \mathbf{V}_{\mathbf{I}}$$

where V_A = additive genetic variance, V_D = variance due to dominance relationships, and V_I = variance due to gene interactions.

Most of the time geneticists are interested in the proportion of phenotypic variance that is attributable to additive genetic variance because it is the additive component that allows us to predict offspring phenotypes and it is the additive component that responds to natural selection. Thus we define **narrow-sense heritability:**

$$\mathbf{h}_{\mathrm{N}}^{2} = \frac{\mathbf{V}_{\mathrm{A}}}{\mathbf{V}_{\mathrm{P}}}$$

There are several ways to measure narrow-sense heritability. Because we often do not know which alleles an individual has, we have to use an indirect method for estimating heritability. One of the most straight forward indirect methods involves investigating the relationship between offspring phenotypes and parents' phenotypes. This is called Parent – Offspring Regression. Simply put, for two variables X (parental phenotypic value) and Y (mean value of the offspring of that parent), the regression coefficient (b_{XY}) is defined as the ratio:

$$\mathbf{b}_{XY} = \frac{\mathbf{COV}_{XY}}{\mathbf{VAR}_{X}}$$

where COV_{XY} is the "covariance" between X and Y which measures the degree to which parents and offspring vary together (i.e. "covary"). VAR_X (= s²) is the variance of the parental measure X. Covariance is calculated as the average product of the deviation from the mean of the two variables:

$$COV_{XY} = \frac{\sum (X_i - \overline{X})(Y_i - \overline{Y})}{n - 1}$$

The regression coefficient, b_{XY} , describes the slope of the regression line of offspring mean phenotype vs. parent's phenotype as in the graph below:



Parent[®] Phenotype

The regression coefficient, b_{XY} , is an estimate of the narrow-sense heritability. For the situation where we have data from one of the parents and the offspring, heritability is twice the regression coefficient:

$$\mathbf{h}_{\mathrm{N}}^2 = 2\mathbf{b}_{\mathrm{XY}}$$

For today's lab exercise, we will examine the heritability butterfly wing pattern elements.

Instructions for Butterfly Wing Lab:

Choosing characters

1. First, familiarize yourself with the wing patterns of *Asterocampa celtis* (the Hackberry Butterfly) using the sample wings provided. **Please be careful with these preserved wings.** Learn to distinguish between the forewing and hindwing and between the dorsal and ventral surfaces.

2. Choose the character for which you will estimate heritability. The character must be measurable with a ruler. Possible characters include the width of the wings (e.g. along one of the veins), width of the various eyespots, the distance between spots, the distance from a spot to the edge of the wing etc.. Talk to your lab instructor about your choice.

3. In order to calculate heritability, a character must be variable. Therefore, try to ensure that your character is variable by measuring a few of the sample individuals. Data collection

- 1. The butterfly wings are organized by family. There are 5 mother butterflies. Each mother has some female offspring (from 3 to 12 daughters). Measure the phenotypes (your character) of all the mothers and all of their offspring. Record your measurements on the attached table.
- 2. Calculate the heritability by calculating the regression coefficient of offspring mean phenotype on parent phenotype.

Data Table: Butterfly Wing Patterns

Name	
i anno.	

1. Enter data in the table below and calculate the offspring mean phenotype for each family:

Mother's	1	2	3	4	5
Number					
Mother's					
Measurement:					

Offspring					
Number:	Offspring Measurements:				
1					
2					
3					
4					
5					
6					
7					
8					
9					
10					
11					
12					
Offspring					
Means:					

2. Calculate the mean of the parents (mothers):	$\overline{\mathbf{X}}$ =			
3. Calculate the variance in the parents' phenotype: $VAR_X =$				
4. Calculate the mean of the offspring means:	<u><u></u><u><u></u><u><u></u><u></u><u></u><u></u><u></u><u></u><u></u><u></u><u></u><u></u><u></u><u></u><u></u><u></u><u></u><u></u></u></u></u>			
5. Calculate the covariance between parent and offspring means: COV_{XY} =				
6. Calculate the regression coefficient:	b _{XY} =			
7. Calculate the heritability of your character:	$\mathbf{h}_{\mathrm{N}}^2 \equiv$			

8. Provide a brief interpretation of your estimate of the narrow-sense heritability (i.e. what does it mean?):