

## POLYGENES AND QUANTITATIVE INHERITANCE

In the previous chapters the variation consisted of sharply contrasting allelic traits. Most traits, however, are brought about by many genes producing phenotypes which do not always fall into easily identifiable categories. This chapter will examine these characters, called quantitative traits. To do this, it will be necessary to describe the statistical methods required for analysis and then, using these methods, determine the relative proportion of the population variability attributable to the environment and heredity.

Many traits of economic importance in domesticated plants and animals as well as characters in laboratory animals and human beings belong to the quantitative category. Comprehension of the methods used to study quantitative inheritance is essential to an understanding of later chapters on selection, inbreeding, and heterosis.

The genetic variation within a population depends upon genes and their action. In a population we observe the phenotypes and then make inferences concerning the genotypes. In the preceding chapters the traits we considered fell into easily identifiable classes. Cattle had horns or they didn't; Drosophila had red or white eyes; men had bald heads or a full head of hair; there was the production of an enzyme or there wasn't. In all such cases the variation was discontinuous with scientists calling such traits mendelian, because Gregor Mendel worked with such traits. Also, they are known as qualitative traits because they refer to the qualities of individuals.

### QUANTITATIVE TRAITS

Many traits do not fit into sharply contrasting classes; they differ by small degrees on a continuous scale. Weight measurements are expressed in grams or pounds, height or length in centimeters or inches, and volume in liters or bushels. Examples of these quantitative traits are height and weight in humans, yields in crops, amount of milk produced in dairy cattle, weight gains in beef cattle, scores on mental tests, longevity, basal metabolism, and other physiological traits such as blood pressure. Thus, there are a large number of characteristics which do not manifest themselves as sharply defined phenotypes.

Quantitative traits are influenced by more than one gene, called polygenes, each with a small effect. The environment may modify the trait considerably because the genetic effect does not divide the trait into greatly differing categories.

## Genetic basis of quantitative inheritance

A simple example of quantitative inheritance is kernel color in wheat. Nilsson-Ehle in 1909 described a series of crosses in which he had scored the color of the wheat kernels from red to white. In one cross he mated a dark redkerneled variety with a white kerneled strain. The F<sub>1</sub> was intermediate in color between the two parents while the F<sub>2</sub> gave a range of phenotypes from red to white. When he scored the colors they fell into five distinct classes with a 1:4:6:4:1 ratio. The extremes, dark red and white, similar to the parental varieties, were each 1/16 of the F<sub>2</sub>.

If we assume that there were two genes affecting kernel color, with A and B each adding some red pigment and a and b no **color**, the cross would be as shown in Figure 17-1.

The F<sub>2</sub> can be analyzed as follows:

| Genotype         | "Red"<br>Alleles | "White"<br>Alleles | Phenotype        | F <sub>2</sub><br>Proportion |
|------------------|------------------|--------------------|------------------|------------------------------|
| AABB             | 4                | 0                  | dark red         | 1/16                         |
| AABb AaBB        | 3                | 1                  | medium red       | 4/16                         |
| AaBb, AAbb, aaBB | 2                | 2                  | intermediate red | 6/16                         |
| Aabb, aaBb       | 1                | 3                  | light red        | 4/16                         |
| aabb             | 0                | 4                  | white            | 1/16                         |

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FIGURE 17-1

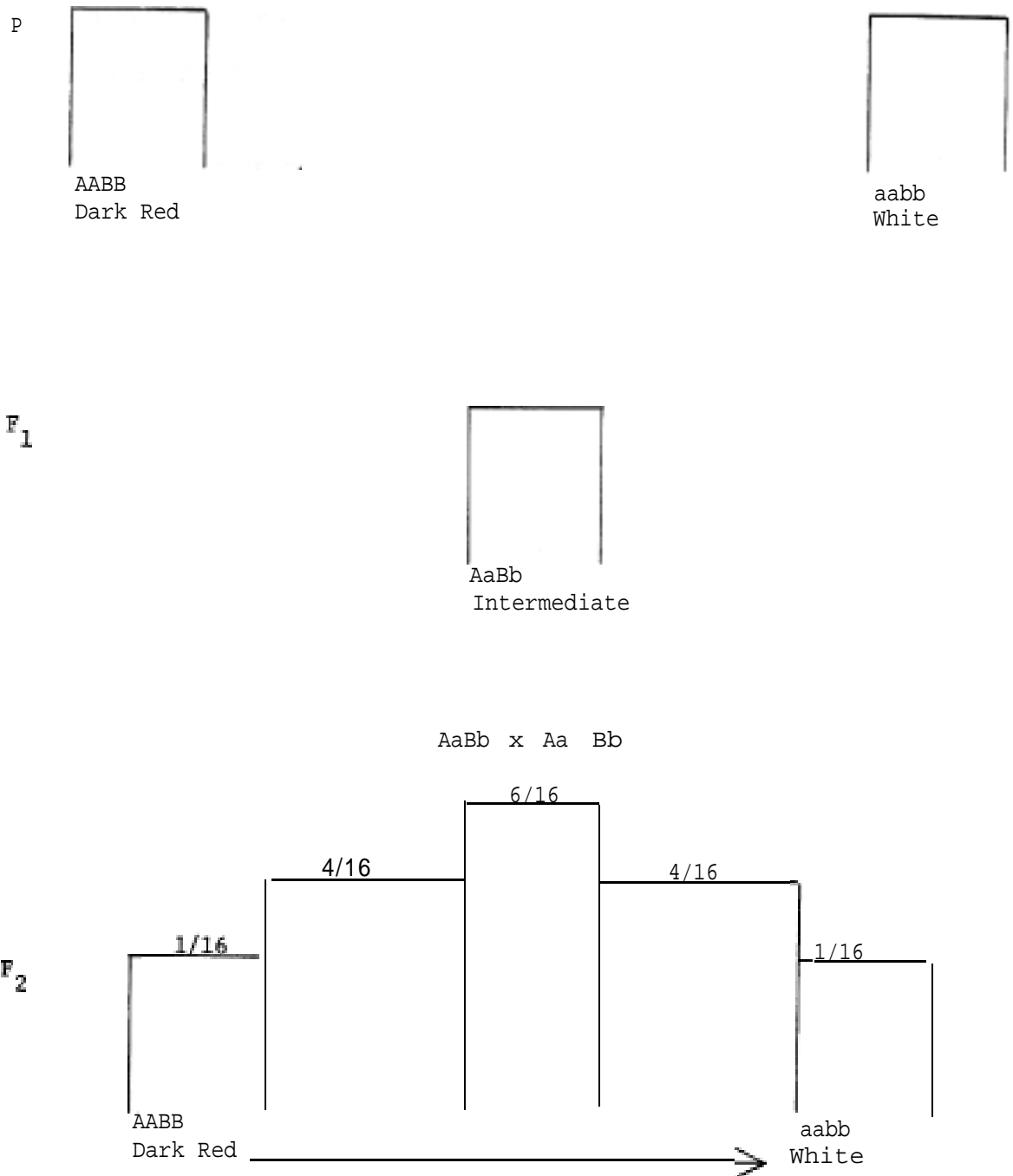


Figure 17-1. Results of a cross between two wheat varieties differing in two pairs of alleles for kernel color,

The proportions of each phenotype in the  $F_2$  may be determined by the methods used in Chapter 9 on two or more pairs of alleles.

The expected proportion of AABB is  $1/4 \times 1/4 = 1/16$ ; that of AABb is  $1/4 \times 2/4 = 2/16$  as is that of AaBB, adding the two together gives  $4/16$ . The same procedures provide the expected proportion of the other phenotypic classes.

The alleles in these crosses are said to be additive or cumulative because they may be added together to determine the phenotype.

The average degree of color of all the individuals in the  $F_2$  may be obtained by multiplying the degree of "redness" (4 being dark red, 3 medium, 2 intermediate, 1 light, and 0 white) by the proportion of individuals in that category and adding the products together,

$$[(4 \times 1) + (3 \times 4) + (2 \times 6) + (1 \times 4) + (0 \times 1)]/16 = 32/16 = 2$$

or intermediate red.

The average redness of the  $F_1$  is also 2 on a scale of 0 to 4 even though each individual is genotypically the same.

Nilsson-Ehle crossed a different red variety and obtained  $1/64$  of the  $F_2$  resembling either parent. He managed to score the kernels into seven classes with a 1:6:15:20:15:6:1 ratio. This proportion of extreme classes as well as the ratio suggests three pairs of alleles affecting kernel color in wheat. The extreme classes AABBCC and aabbcc each have a relative proportion of  $(1/4)^3 = 1/64$  because the probability of obtaining a homozygous dominant or recessive at each locus is  $1/4$ . The general term for the proportion of these extreme classes is  $(1/4)^n$  n being the number of loci involved. As the number of loci increase the proportion of individuals in the extreme classes decrease.

As more pairs of alleles become involved in a quantitative trait, the number of phenotypic classes becomes greater in the  $F_2$ , making it more difficult to classify the progeny into definite categories. The steps between class divisions become smaller and environmental influences cause overlap at the class divisions.

#### MEANS AND VARIANCES

Quantitative traits measured on a continuous scale require different methods of analysis than ratios or percentages. Means and variances are the principal statistics used for analysis of quantitative traits.

The mean is a measure of the central tendency or magnitude of a population. The arithmetic mean, also known as the average, is obtained by adding the measurements taken on a sample from a population and dividing by the number of measurements, n. Symbolically, this is written as

$$\bar{x} = \frac{X_1 + X_2 + \dots + X_n}{n} \text{ or } \bar{x} = \frac{\sum X}{n}$$

where  $\bar{x}$  is the mean,  $\Sigma$  is the symbol for sum of, X is the measurement, and n is the number of measurements.

Let us run through an example to illustrate the process. Male Coturnix quail were weighed at six weeks of age and the following individual weights in grams obtained: 76, 85, 98, 100, 103, 110, 111, 117, 120, and 130. What is the mean?

$$\bar{x} = \frac{\sum X}{n} = \frac{76 + 85 + 98 + 100 + 103 + 110 + 111 + 117 + 120 + 130}{10} = \frac{1050}{10} = 105$$

Means are useful for comparing various populations. The mean of a parental generation is compared with the mean of its progeny generation to determine shifts because of environmental or genetic changes.

Means tell us little more than the central value of the particular set of numbers. We do not know, from the mean, how much the numbers vary. Are they far apart or are they clustered close together?

We may ascribe different properties to the variation of individuals of a specific population. The variation may be genetic because of the different genotypes of the individuals or it may be environmental, or it may be a combination of both genetic and environmental effects.

The dispersion of values around the mean is measured by the variance. The variances of different populations may be **compared to one another and inferences** made concerning the genetic make up of the population. The symbol for variance is  $s^2$  and is defined as the sum of the squared differences between the individual measurements and the mean, divided by the number of observations minus one.

$$s^2 = \frac{(X_1 - \bar{x})^2 + (X_2 - \bar{x})^2 + \dots + (X_n - \bar{x})^2}{n-1} = \frac{\sum (X - \bar{x})^2}{n-1}$$

The example of Coturnix quail body weights used to illustrate the calculation of a mean may be utilized to show the steps. The mean is  $\bar{x} = 105$  grams.

| Observation<br>x   | Deviation from Mean<br>$X - \bar{x}$ | Deviation Squared<br>$(X - \bar{x})^2$ |
|--------------------|--------------------------------------|--|
| 76                 | - 29                                 | 841                                    |
| a5                 | - 20                                 | 400                                    |
| 98                 | - 7                                  | 49                                     |
| 100                | - 5                                  | 25                                     |
| 103                | - 2                                  | 4                                      |
| 110                | 5                                    | 25                                     |
| 111                | 6                                    | 36                                     |
| 117                | 12                                   | 144                                    |
| 120                | 15                                   | 225                                    |
| 130                | 25                                   | 625                                    |
| $\Sigma X = 1,050$ |                                      | $\Sigma (X - \bar{x})^2 = 2,374$       |

$$s^2 = \frac{2374}{10-1} = 263.8$$

If you have access to a calculator, the formula for obtaining the variance would be:

$$s^2 = \frac{\Sigma X^2 - (\Sigma X)^2}{n-1}$$

$\Sigma X^2$  is the sum of each squared observation ( $76^2 + 85^2 + \dots + 130^2$ ) = 112,624 and  $(\Sigma X)^2$  is the square of the sum of all observations  $(1,050)^2 = 1,102,500$ .

$$s^2 = \frac{112,624 - \frac{1,102,500}{10}}{10-1} = \frac{2374}{9} = 263.8$$

Calculate the mean and variance of six-week body weight of mice in grams: 10, 12, 15, 16, 17, 17, 19, 23, 25, 26 29, 31. (Answer 1)

The environment may drastically affect each individual in a population. If we measured the weight of a group of people we would find that some people were heavier because they overate, and some were light because they dieted or were ill. In a population, each individual is affected by his own environment. The pine trees in the forest grow short or tall depending upon their genotype and whether they are exposed to winds, are shaded by other trees, become diseased, are at a high or low elevation on the mountain side, or are situated in soil with optimum fertility.

The effect of the environment is to make it more difficult to determine genetic effects with the result that the frequency distribution looks like the curves in slide 17-2. The two curves each form a normal distribution. Populations A and B have the same mean with B having the greater variance. Most quantitative traits show a normal curve when enough individuals are measured in the population.

Refer ahead to slide 17-4 for two actual distributions of ear length in corn. Here the mean of the  $F_1$  and  $F_2$  are about the same but the variance of the  $F_2$  is greater than that of the  $F_1$ .

While the variance designates the variation of a population the standard deviation (square root of the variance, symbolized by  $s$ ) is useful because it describes the normal curve in terms of probabilities and because it is in the same units as the mean. Note in slide 17-3 that the areas in the curve are divided into sections with reference to the mean. The area bounded by  $\pm s$  (plus or minus  $s$ ) encloses about two-thirds of all the observations. The area within the mean  $\pm 1.98s$  has 95% of the total observations.

Knowledge of the population's standard deviation may determine whether or not an observation is relatively frequent or rare. An individual measurement falling outside of three standard deviations from the mean would be infrequent. However, if an individual's measurement is within one-half of a standard deviation from the mean, it would be within a class of high frequency.

Refer to the previous example using quail body weights. The standard deviation is  $s = \sqrt{263.8} = 16.2$ . The area of  $105 \pm 16.2$  extends to 88.8- 121.2 and has about two-thirds of the observations enclosed within these limits. There were 10 observations in the sample and 3 (76, 85, and 130) fall outside, so about two-thirds of the values were within the limits and one-third were outside.

#### FURTHER ASPECTS OF QUANTITATIVE INHERITANCE

Other studies have brought out further aspects of quantitative inheritance. Emerson and East (1913) measured ear length in corn (*Zea mays*). The trait does not fall readily into discernible categories, thereby demonstrating continuous variation.

Tom Thumb popcorn (short length) was crossed with Black Mexican sweet corn (long length). The alleles affecting ear length might be assumed to be homozygous within each parent (slide 17-4). The  $F_1$  has a range of 9 to 15 cms while the  $F_2$  runs from 7 to 19. There is no reason to expect greater environmental influences on the  $F_1$  individuals than on the  $F_2$  progeny so the greater  $F_2$  variation can be attributed to chromosomal segregation and recombination.

When populations with different means are compared, the numerical magnitude of the variances (or, more exactly, of the standard deviations) are meaningful relative to the magnitude of their means. The variation may be expressed as the coefficient of variation [C.V.% =  $(\frac{s}{\bar{x}}) 100$ ] which is the standard deviation in terms of the units of the mean. A 1 kilogram variation among elephants hardly means as much as 1 kilogram variation among mice.

In the following table the means, variances, standard deviations, and coefficients of variation were calculated from the data.

| Designation   | Mean   | Variance | Std. Dev. | c. v. (%) |
|---------------|--------|----------|-----------|-----------|
| Tom Thumb     | 6.632  | 0.67     | .8185     | 12.34     |
| Black Mexican | 16.802 | 3.56     | 1.887     | 11.23     |
| F1            | 12.116 | 2.31     | 1.520     | 12.54     |
| F2            | 12.678 | 3.98     | 1.995     | 15.74     |

Note the  $F_2$  coefficient of variation was greater than the F1 coefficient.

Skin color in humans is a characteristic which appears to follow the pattern of quantitative inheritance. If we observe white Caucasians and dark colored blacks, there is a wide variation between individuals within each of the races, although it is usually easy to assign individuals to each race.

Harrison and Owen studied the inheritance of black and white human skin color by using a reflectance spectrophotometer, a machine measuring the light reflectance of the skin. The skin color on the upper right arm was measured through several filters and the log transformation of the 425  $\mu$  filter data was found to be best for genetic analysis. It should be noted that the darker the skin the lower the reflectance.

Most of the subjects were residents of Liverpool, England. The Europeans had a large Irish component while the blacks came from West Africa and consisted of three groups: 1) settled residents, 2) seamen on the West African "run", and 3) university students. Because of the diverse origins of the West African population a greater variation was expected than for the European population.

Marriages between whites and blacks were investigated and the F1's were intermediate in color to that of the parents. Marriages of F1 x F1 results in an array of colors in the  $F_2$  offspring while the cross of F1's back to the parental groups resulted in color intermediate between the F1 and the parental group (Table 17-1). The number of F2 individuals was small (13) so not much reliance can be placed on that specific variance.

TABLE 17-1

REFLECTANCE OF SKIN COLOR ON UPPER ARM USING A 425  $\mu$  FILTER FOR WEST AFRICANS, EUROPEANS, AND VARIOUS CROSSES IN LIVERPOOL, ENGLAND. DATA GIVEN IN LOGARITHMS FROM HARRISON AND OWEN (1964)

| Groups             | Number | Average | Variance x 10 <sup>-5</sup> |
|--------------------|--------|---------|-----------------------------|
| Europeans          | 104    | 1.556   | 317                         |
| West Africans      | 40     | 1.082   | 1,236                       |
| F1 hybrid          | 94     | 1.308   | 885                         |
| African Backcross  | 21     | 1.245   | 1,024                       |
| European Backcross | 30     | 1.420   | 1,310                       |
| $F_2$ hybrid       | 13     | 1.336   | 1,047                       |

It appears that there is a minimum of three and possibly as many as six additive loci involved with skin color. All investigations indicate that dominance has little or no effect on alleles affecting color. If we assume that there are four pairs of alleles involved and that A, B, C and D each add more pigment than a, b, c, or d, then the white person would be aa bb cc dd and the black individual AA BB CC DD.

Marriages of a white person, aa bb cc dd, to a slightly darker person, Aa Bb cc dd, would not be expected to produce a child darker than the darkest parent because Aa Bb cc dd is the darkest progeny possible. There would, therefore, be no sound scientific basis for the tales of white marriages producing "black babies." However, a marriage between two people with intermediate color and genotypes Aa Bb Cc Dd could produce some individuals which were darker or lighter than either parent.

Skin color is susceptible to tanning by the sun; individuals react differently--some are deeply tanned, others only redden. Environment is a factor in the variation of skin color obscuring the differences attributable to the genotypes.

In the foregoing examples where the loci affecting the quantitative traits are additive in nature, the following findings are typical:

1. The  $F_1$  is approximately intermediate between two extreme homozygous parents.
2. The average of the  $F_2$  is similar to that of the  $F_1$ .
3. The variation of the  $F_2$  is greater than the  $F_1$  because of the segregation and recombination of the alleles.
4. The traits may be modified by the environment.

#### Dominance in quantitative inheritance

It is unrealistic to assume that all alleles influencing quantitative traits are additive. Let us examine the effect that dominant alleles would have on quantitative inheritance.

Using the model shown in Figure 17-5 the values for the homozygotes BB and bb are placed at each end of the model with a zero point at the midpoint between the two homozygotes. The additive effect of BB is +a while the similar effect of bb = -a. The heterozygote may fall on either side of the origin or midpoint or exactly on the midpoint, depending upon d. In this particular case, the mean of the heterozygotes falls between the midpoint and BB with a dominance deviation of +d. If there were complete dominance of allele B then Bb would equal BB with the result that d = a. If the alleles show no dominance (d = 0) they are said to be "additive alleles" or to act additively. It is also possible to have a negative d when the heterozygote value falls between and the midpoint.

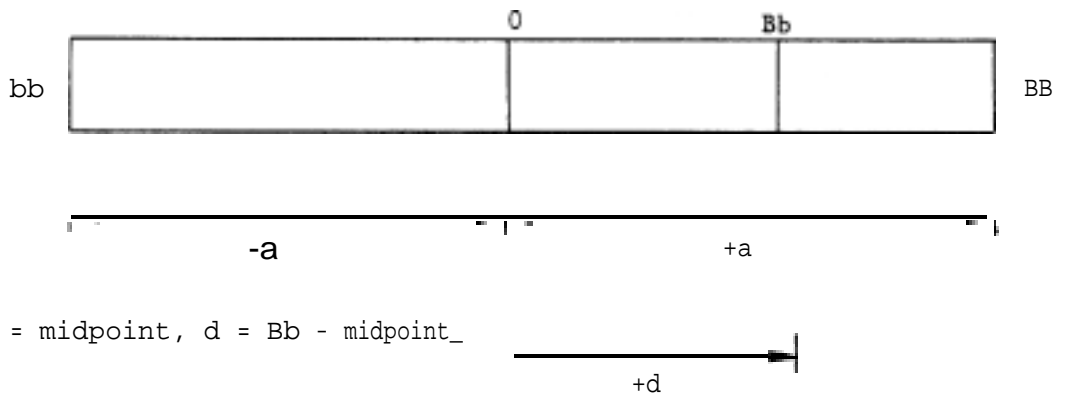
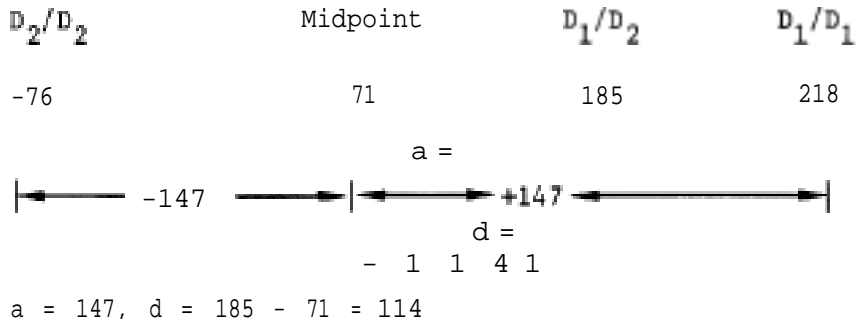


Figure 17-5. Hypothetical model for effect of one locus on a quantitative trait.

An example in dairy cows may be used to illustrate the model. In a study of the five breeds of dairy cattle, Jamieson and Robertson reported on the relationship of transferrins (specific B-globulin factors in the blood) and milk production. The genotypes  $D_1/D_1$ ,  $D_1/D_2$  and  $D_2/D_2$  are given below with the first lactation milk yield expressed as deviations from the breed mean in pounds of milk.



The additive effect,  $a$ , is 147 and the dominance effect,  $d$ , is 114.

In a study of the adaptive value of blood group genes in chickens, Shultz and Briles (1953) reported that hens with genotypes  $A^7/A^7$  laid eggs weighing 56.8 grams,  $A^7/A^9$  58.0 grams, and  $A^9/A^9$  58.4 grams. Calculate the  $a$  and  $d$  values for this locus. (Answer 2)

Quantitative traits are influenced by several genes. Suppose the genotypic value of  $AA$  is  $a_A$ ,  $BB$  is  $a_B$ , and  $CC$  is  $a_C$ . Then adding these values together for  $AA BB CC$  gives a genotypic value of  $a_A + a_B + a_C$ . With this additive combination each locus contributes to the joint effects upon the quantitative trait. The same considerations apply to the other genotypes at the various loci with each contribution being summed to provide the final effect on the trait.

## Estimation of genetic variation

If two parental lines are both homozygous for the alleles affecting the observed trait, the F<sub>1</sub> individuals would be uniformly heterozygous. Each of these populations, the two parents and the F<sub>1</sub>, would have no genetic segregation and any variances present must reflect environmental modifications. Their variances estimate the environmental variance, V<sub>E</sub>. An average of the two parental and the F<sub>1</sub> variances may be used as an estimate of the environmental variance.

$$(V_{P1} + V_{P2} + V_{F1})/3 = V_E$$

The F<sub>2</sub> variance is larger than the F<sub>1</sub> because of the genetic segregation and recombination. The F<sub>2</sub> variance estimates the genotypic plus the environmental variance. This is also known as the phenotypic variance, V<sub>P</sub>.

$$V_{F2} = V_P = V_G + V_E$$

An estimate of the genotypic variance may be obtained by subtracting the F<sub>1</sub> from the F<sub>2</sub> variance.

$$V_{F2} - V_{F1} = (V_G + V_E) - V_E = V_G$$

If we substitute a single allele in a gene pair and it increases or decreases the phenotypic value, the effect is additive. This effect is equivalent to a in Figure 17-5. It determines the additive genetic variance, V<sub>A</sub>. The deviation of the heterozygote from the midpoint is d and depends upon the interaction of the two different alleles at the same locus. This dominant effect contributes to the dominance variance, V<sub>D</sub>.

The additive variance is important in artificial selection systems such as selection for growth in beef cattle, sheep, and swine while the dominance variance of a population plays a major role in exploitation of hybrid vigor involving heterozygotes.

Using these combined values of a and d each individual in the population can be assigned an additive value and a dominance deviation. Variances for the population are calculated providing a genotypic variance, V<sub>G</sub>, additive genetic variance, V<sub>A</sub>, and a dominance variance, V<sub>D</sub>. If d = 0, then there would be no dominance variance and all the genotypic variance would be additive.

These variances depend upon the numbers of the involved genotypes, e.g., if the whole population consisted of AA BB CC genotypes there would be no genotypic variance at all. Thus, the genetic variances depend upon the magnitude of the a and d values as well as the differences between genotypes.

The additive and dominance variances are independent so they may be added together to give the genotypic variance

$$V_G = V_A + V_D$$

The variance of an F2 will then consist of

$$V_{F_2} = V_P = V_A + V_D + V_E$$

A backcross of the F<sub>1</sub> to one of the parental groups will estimate one-half of the additive genetic variance and all of the dominance and environmental variance.

$$V_B = 1/2 V_A + V_D + V_E$$

summing the two backcrosses (one to each of the parental types) provides

$$V_{B_1} + V_{B_2} = V_A + 2V_D + 2V_E$$

Estimates of the different variances may be obtained from quantitative data of different crosses.

$$V_E = (V_{F_1} + V_{P_1} + V_{P_2})/3$$

$$V_D = (V_{B_1} + V_{B_2}) - V_{F_2} - V_E$$

$$\begin{array}{r} V_{B_1} + V_{B_2} - V_{F_2} - V_E \\ \text{subtract } V_{F_2} \quad \frac{V_A + V_D + V_E}{V_D + V_E} \\ \text{subtract } V_E \quad \frac{V_A}{V_D} \end{array}$$

$$2V_{F_2} = 2V_A + 2V_D + 2V_E$$

$$\begin{array}{r} \text{subtract } (V_{B_1} + V_{B_2}) \\ \quad \quad \quad \frac{V_A + 2V_D + 2V_E}{V_A} \end{array}$$

$$V_A = 2V_{F_2} - (V_{B_1} + V_{B_2})$$

The data from an experiment by Chai (1956) where large mice were crossed to small mice illustrates the estimation of the different variances. The means and variances of the crosses are presented in Table

TABLE 17-2

DATA ON MOUSE BODY WEIGHT OF PARENTAL LINES AND CROSSES.  
VALUES BASED ON LOGARITHMIC TRANSFORMATION FROM CHAI (1956).

|   | No.<br>Individuals | Mean  | Variance x 10 <sup>-5</sup> |
|---|--------------------|-------|-----------------------------|
| P <sub>1</sub> Small                              | 41                 | 1.130 | 44                          |
| P <sub>2</sub> Large                              | 65                 | 1.541 | 49                          |
| F <sub>1</sub>                                    | 161                | 1.397 | 36                          |
| F <sub>2</sub>                                    | 216                | 1.395 | 191                         |
| B <sub>1</sub> (F <sub>1</sub> x P <sub>1</sub> ) | 156                | 1.294 | 184                         |
| B <sub>2</sub> (F <sub>1</sub> x P <sub>2</sub> ) | 164                | 1.471 | 125                         |

The logarithms of the actual values were used to compensate for the multiplicative nature of body growth (instead of adding, growth doubles). The variances estimated by the formulas are:

$$V_E = (44 + 49 + 36)/3 = 43$$

$$V_D = (184 + 125) - 191 - 43 = 75$$

$$V_A = 2(191) - (184 + 125) = 73$$

The  $V_{F_2}$  is then

$$V_{F_2} = V_A + V_D + V_E = 73 + 75 + 43 = 191$$

The variances of economic traits in domesticated plants and animals are of importance because a high additive variance indicates that selection will readily change the trait. A large dominance variance provides evidence that a line crossing mating system utilizing hybrid vigor would be successful.

In a cross of the homozygous wheat varieties Ramona and Baart 46, Allard measured the dates of heading.' The means and variances of the parental and hybrid generations are given in Table 17-3. Estimate the environmental, additive, dominance and phenotypic variances. (Answer 3)

TABLE 17-3

DATA ON DAYS TO HEADING OF WHEAT  
SOURCE OF DATA: ALLARD (1960)

| Generation  | No.<br>Individuals | Mean         | Variance |
|---|--------------------|--------------|----------|
| P <sub>1</sub> Ramona                             | 159                | 12.99        | 11.036   |
| P <sub>2</sub> Baart 46                           | 148                | 27.61        | 10.320   |
| F <sub>1</sub>                                    | 171                | 1a.45        | 5.237    |
| F <sub>2</sub>                                    | 552                | 21.20        | 40.350   |
| B <sub>1</sub> (F <sub>1</sub> x P <sub>1</sub> ) | 326                | 15.63        | 17.352   |
| B <sub>2</sub> (F <sub>1</sub> x P <sub>2</sub> ) | 314                | <b>23.38</b> | 34.288   |

#### HERITABILITY

In the foregoing examples, it was possible to estimate the genetic and environmental variances in certain populations.

When the genetic variance is large and the environmental variance small, then the offspring will resemble their parents to a high degree.

Consider the case of the polled and horned alleles in cattle affecting the appearance of horns with very little environmental interference. The appearance in the progeny can be usually predicted by observing the parental phenotypes. However, there is still some uncertainty because polled is dominant in expression to horned so that a mating of polled cattle could produce horned offspring.

This uncertainty is reduced when the traits show no dominance. Human skin color is such a trait where marriages between Caucasians have white skinned children, between Blacks dark skinned children, and between Caucasians and Blacks brown children. Knowing the parental genotypes enables us to predict the color of the offspring to a high degree.

Therefore, the additive portion of the genotype is more important in predicting the resemblance of relatives to one another than the dominance portion. The relative importance of heredity and environment is expressed by the ratio

of the additive genetic variance to the total or phenotypic variance. This expression is called the heritability of a trait with the symbol H.

$$H = V_A/V_P = V_A/(V_A + VD + VE)$$

Heritability is the relative influence of the genotype on the phenotype of a quantitative trait. When it is large the trait is genetically influenced to a greater degree by the genotype than when the heritability is small.

Knowledge of heritability is useful to the plant and animal breeder because it enables them to predict how the selection of parents will influence the performance of their progeny. Also knowing the heritability of a trait provides some indication of the familial inheritance. Human height has a high heritability; so one might predict that tall children will be borne by tall parents and short children by short parents. Therefore, there will be clusters occurring by families when a trait has a high heritability while a low heritability trait will be evenly distributed throughout the population.

The heritability of mouse body weight estimated from the data given in Table 17-2 is

$$H = 73/191 = 0.38$$

Estimate the heritability of days to heading in wheat (Table 17-3).  
(Answer 4)

The variances considered so far do not take into account the interactions between genes or differential responses of genotypes to various environments, genotype x environment interactions. These factors further complicate estimates of heritability.

The situation for quantitative inheritance is far more complicated than our simple models would indicate. Sometimes the genotype appears to act in a multiplicative way on a quantitative trait such as growth where growth measurements are multiplied by a constant at intervals; in others there is interaction among genes, with resultant difficulties of analysis, and, overall, individuals with certain genotypes may seek out certain environments, creating a genotype x environment correlation.

Other methods of estimating heritability using the relationships among relatives will be described in Minicourse 18.

#### THRESHOLD TRAITS

During development the differentiation of cells in an individual to form various organs and structures can take one of several alternative routes. In the human embryo if the lateral parts of the face fail to fuse, a harelip and cleft palate forms in the jaw (see slide 17-6). In other situations teeth may

grow but fail to erupt. Often, such traits are determined by a single pair of alleles; however, for many such traits, polygenes determine the developmental pathway.

Traits which are all-or-none (they either appear or they don't) and are determined by polygenic inheritance are called threshold traits.

Sewall Wright crossed two strains of guinea pigs, one with three toes while the other with four toes was polydactylous. The  $F_1$  were three-toed while the  $F_2$  segregated into 3 three-toed to 1 four-toed individuals. Further tests of the segregants, however, showed that 4 pairs of alleles were probably involved, divided into three-toe and four-toe alleles (slide 17-7). A total of eight alleles were affecting the trait with a minimum of five four-toe alleles required for a four-toed individual. The five four-toe alleles provided the threshold beyond which four-toed guinea pigs occurred.

Threshold traits are fairly common in humans. Table 17-4 gives the incidence of a few characters which have been studied by Carter (1965). They constitute a notable source of disease incidence in our society. These traits depend upon a threshold determined by both genotype and environment. For example, an individual with a genotype susceptible to a specific disease such as tuberculosis will contract the disease if exposed to the tuberculosis bacilli. In a population of genetically susceptible individuals in a poor environment (lack of food, exposure to wet and cold) with constant exposure to the disease organism, the threshold would be low and the disease incidence high. Improve any of the elements, genotypes, physical environment or disease exposure, and the threshold will rise and the incidence fall.

In Slide 17-6 the identical twins show the effect of the environment on the expression of harelip. Both have identical genotypes; the one on the left expresses the trait, the twin on the right does not.

TABLE 17-4

INCIDENCE OF COMMON HUMAN CONGENITAL MALFORMATIONS EXCLUDING  
CHROMOSOMAL ABNORMALITIES (CARTER, 1965)

| Malformation   | Approximate Incidence<br>per 1000 Live Births |
|--|---|
| Anencephalus (absence of parts of the brain)<br>and Spina Bifida (spinal defect) | 5   |
| Clubfoot   | 1   |
| Congenital dislocation of the hip  | 1   |
| Harelip (with or without cleft palate)   | 1   |
| Pyloric stenosis (narrowing of stomach<br>opening)                               | <u>3</u>                                      |
|  | 11 (1.1 percent)                              |

Once the trait is expressed the degree or extent of the expression is the expressivity. Diabetes mellitus is a threshold trait. When an individual has diabetes the range of expressivity may differ from a mild form of the disease easily controllable by diet intake and insulin injections to a severe manifestation leading to early death.

Thus, we see that threshold traits are affected by polygenes and the environment, falling into two classes, expressed or not expressed; when expressed, many of these type traits may be measured on a continuous scale.

## SUMMARY

In qualitative inheritance a gene with a major effect is involved and the phenotypes may be classified into separate categories. Quantitative inheritance involves traits influenced by several genes with small effects, called polygenes, so that the phenotypes cannot be placed in easily identifiable classes. Most of these traits are measured on a continuous scale. The mean and the variance, a measure of the population's variability, are required.

When the  $F_1$  between two homozygous parental lines is approximately midway between the two parental lines, the alleles affecting the measured trait are probably acting additively. The increased variability of the  $F_2$  over the  $F_1$  is due to genetic segregation and recombination. The variances of the homozygous parental lines and the uniformly heterozygous  $F_1$  represent the environmental variance or how much the individuals of these populations are modified by the environment. The difference between the  $F_2$  variance and the environmental variance represents the genotypic variance, made up of the additive and dominance variance. By considering the variances of backcrosses it is possible to estimate the additive and dominance variances of the population.

The relative influence of the genotype on the phenotype is given by the ratio of the additive to the total or phenotypic variance. This measure, called heritability, is useful to the plant and animal breeder for determining the efficiency of selection for a quantitative trait. Also, it gives an indication of the possibility of familial inheritance,

Threshold traits are an important category in quantitative inheritance. If the genotypic effect in an individual exceeds a certain developmental threshold, the trait appears in the phenotype and may be expressed in various degrees. Diabetes and harelip are two examples of threshold traits.

Analysis of traits in quantitative inheritance is more complex than characters affected by one or two single genes. However, so many traits are controlled by several genes working together that the statistical tools needed to study quantitative inheritance are an important part of genetics.

## Objectives

1. Define qualitative and quantitative inheritance and give an example of traits involved in these two types of inheritance.
2. Define mean, variance, and standard deviation. Given data, calculate these statistics.
3. When two homozygous varieties differing in a quantitative trait are crossed resulting in F<sub>1</sub>, F<sub>2</sub>, and backcross generations, be able to interpret the outcomes in terms of single genes.
4. Define phenotypic, genotypic, additive, dominance, and environmental variances of a population.
5. Given variances of homozygous parental lines, F<sub>1</sub>, F<sub>2</sub>, and backcrosses, calculate the additive, dominance, environmental, and phenotypic variances.
6. Define heritability and calculate its estimate when given data from a population.
7. Define threshold traits, providing their genetic and environmental basis.

## ANSWERS TO TEXT QUESTIONS

1. The mean of the twelve six-week mouse body weights is

$$\bar{x} = (10 + 12 + 15 + 16 + 17 + 17 + 19 + 23 + 25 + 26 + 29 + 31)/12 = 240/12 = 20 \text{ grams.}$$

The variance,  $s^2$ , is

| <u>Measurement</u> | <u>Deviation</u> | <u>Deviation Squared</u> |
|--------------------|------------------|--------------------------|
| 10                 | -10              | 100                      |
| 12                 | - 8              | 64                       |
| 15                 | - 5              | 25                       |
| 16                 | - 4              | 16                       |
| 17                 | - 3              | 9                        |
| 17                 | - 3              | 9                        |
| 19                 | 1                | 1                        |
| 23                 | 3                | 9                        |
| 25                 | 5                | 25                       |
| 26                 | 6                | 36                       |
| 29                 | 9                | 81                       |
| 31                 | 11               | 121                      |
|                    | Total            | 496                      |

$$s^2 = 496/(12-1) = 45.1$$

2. The midpoint between the two homozygotes is  $(56.8 + 58.4)/2 = 57.6$  grams. The a value is  $58.4 - 57.6 = 0.8$  grams. The d value is 0.4 grams.
3. The environmental variance is estimated by  $(V_{P_1} + V_{P_2} + V_{F_1})/3 = V_E = (11.036 + 10.320 + 5.237)/3 = 8.864$

The dominance variance is

$$V_D = (V_{B_1} + V_{B_2}) - \frac{V_P}{2} - V_E = (17.352 + 34.288) - 40.350 - 8.864 = 2.426$$

The additive genetic variance is

$$V_A = 2v_{F_2} - (V_{B_1} + V_{B_2}) = 2(40.350) - (17.352 + 34.288) = 29.060$$

The phenotypic variance is the sum of all the variances

$$V_P = V_A + V_D + V_E = 29.060 + 2.426 + 8.864 = 40.350$$

4. Heritability is the ratio of the additive to the phenotypic variance. Using data from Answer 4 above  $V_A = 29.060$  and  $V_P = 40.350$ , heritability is

$$H = 29.060/40.350 = 0.72$$

With such a high correspondence of genotype to phenotype selection for days to heading in wheat should proceed fairly rapidly.

## STUDY GUIDE

### I. Qualitative Traits

- A. Usually affected by a single pair of alleles; sometimes two pairs of alleles.
- B. Also called Mendelian or discontinuous traits.
- C. Phenotypic expression not greatly affected by the environment.
- D. Fall into easily identifiable classes.
- E. Examples:

Dwarf vs. tall in peas, polled vs. horned in cattle, white vs. colored fur in domestic cats.

### II. Quantitative Traits

- A. Affected by several genes, multiple alleles, each with small effects (polygenes) and the environment.
- B. Generally, does not fall into identifiable classes.
- C. Measured on a continuous scale: inches, centimeters, grams, pounds (except for threshold traits).
- D. Examples:

Skin color in humans, I.Q., blood pressure, body weight in pigs, height of trees, speed in race horses, egg production of chickens, wheat kernel color, and ear length of corn.

### III. Statistical Measures for Quantitative Traits

- A. Mean ( $\bar{x}$ ) = the average or magnitude of central value  
$$\bar{x} = \frac{\sum X}{n}$$
 where  $\sum$  = sum of, and  $n$  = number of observations.
- B. Variance ( $s^2$ )

A measure of the variation of a population

$$s^2 = \frac{\sum (X - \bar{x})^2}{n - 1}$$

Usually a quantitative trait forms a normal frequency distribution curve.

C. Standard deviation(s)

Square root of the variance

$$s = \sqrt{\frac{\Sigma(X - \bar{x})^2}{n - 1}}$$

IV. Genetic Basis of Quantitative Inheritance

A. With additivity of alleles

1.  $F_1$  is intermediate between the two homozygous parental lines.
2. Average of  $F_2$  is similar to  $F_1$ .
3.  $F_2$  variation is greater than  $F_1$  variation.
4. Traits may be modified by environment.
5. Allelic effects may be summed over total genotype to produce genetic effect on individual phenotype.

B. Dominance

Complete dominance  $Aa = AA$ . Deviation of heterozygotes from midpoint between homozygotes.

C. Multiplicative action

When alleles affect trait by multiplying not adding effects.  
Example: body growth.

V. Estimation of Genetic and Environmental Variances from Cross of Homozygous Lines

A. Interpretation of variances of parental lines,  $F_1$ ,  $F_2$ , and back-crosses.

$$V_{P_1} = V_E \text{ (environmental variance)}$$

$$V_{P_2} = V_E$$

$$V_{F_1} = V_E$$

$$V_P \text{ (phenotypic variance)} = V_{F_2} = V_G + V_E \text{ (genotypic + environmental variances)}$$

$$V_G \text{ (genotypic variance)} = V_A + V_D \text{ (additive + dominance variances)}$$

$$V_{B_1} \text{ (backcross of } F_1 \text{ to } P_1) = VA + VD + V_E$$

$$V_{B_2} \text{ (backcross of } F_1 \text{ to } P_2) = 1/2 VA + VD + VE$$

B. Environmental variances,  $V_E$

$$(V_{P_1} + V_{P_2} - V_{F_1})/3 = V_E$$

C. Genotypic variance, all the genetic variance,  $V_G$

$$V_{F_2} - V_E = V_G$$

D. Additive genetic variance, variance of additive effect alleles, "A"

$$2 \sigma_{F_2}^2 - (V_{B_1} + V_{B_2}) = V_A$$

E. Dominance variance,  $V_D$

Due to deviation of heterozygotes from midpoint between homozygotes.

$$(V_{B_1} + V_{B_2}) - V_{F_2} - V_E = V_D$$

F. Phenotypic variance,  $V_P$

The genotypic plus environmental variances in the population. Expressed as variance of the phenotypic observations of the  $F_2$  population.

## VI. Heritability

Provides a measure of genetic influence on the phenotype. Ratio of the additive genetic variance to the total variance.

$$H = \frac{V_A}{V_P}$$

## VII. Threshold Traits

A. Definition

All-or-none traits affected by several genes and the environment. Threshold is determined by genotypic and environmental effects. Trait is expressed when threshold is exceeded and not expressed when below the threshold.

Examples: Hatchability of bird embryos, disease and mortality, appearance or non-appearance of a tooth, number of toes in guinea pigs, harelip and cleft palate in humans.

## B. Expressivity

The variation of a threshold trait when expressed.

Example: Diabetes mellitus when present varies from mild to severe form of disease.