

Quantitative Genetics

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Quantitative genetics is the assessment of variation due to genetic and environmental factors. It can be used to identify the sources of similarity between relatives.

Introduction

Genetic analysis of quantitative traits is applied in a wide variety of substantive research areas. Many human characteristics exhibit continuous variation, such as height, weight, intelligence or personality, but the application of quantitative genetics is not limited to traits thought to be continuous. For example, eye color may be assessed in a qualitative fashion – blue versus brown versus green – but there exists substantial variation within these categories that could be measured and therefore quantified. Even apparently binary characteristics, such as having had heart disease or not, may best be understood in terms of an underlying continuum of predisposition to heart disease, rather than as a qualitative outcome. Conversely, a truly qualitative, monogenic trait could be considered as a quantitative trait that has a single source of variation. Therefore, almost all genetics is, at its base, concerned with quantitative traits. Comprehension of the basic elements of a quantitative genetic model requires some understanding of how we measure variation, or individual differences.

Population Variability

Perhaps the most elementary statistic used to describe a population is the mean, usually symbolized as μ , which is calculated by summing the scores (x_i) of individuals $i = 1, \dots, N$, and dividing this total by N , the number of scores in the sample. There are several ways of describing the variation in a sample, such as noting its maximum and minimum, or particular percentiles. Probably the most useful for statistical purposes is the variance, which is calculated as the average of the squared distances from the mean:

$$s^2 = \frac{\sum_{i=1}^N (x_i - \mu)^2}{N} \quad (1)$$

This quantity was shown by Fisher (1922) to have several valuable properties, of which the most notable are that (i) with infinite sample size it is unbiased, that is, correct, and (ii) it is the most accurate of all measures that have criterion (i). Therefore, these statistics provide a natural foundation for a

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quantitative genetic model of individual differences. Much quantitative genetics in both humans and inbred laboratory animals concerns the computation of variances and the subsequent partitioning of variance into different genetic and environmental components. The seminal work in this area was also by Fisher (1918) and was further developed by Mather and Jinks (1971).

A single locus model

Suppose that an autosomal genetic locus has only two alleles, A and a . Since humans are diploid, the possible genotypes at this locus are the homozygotes AA and aa and the heterozygote Aa . Let the means of individuals grouped according to these genotypes be d , $-d$ and h respectively (in this notation d is used for additive deviation and h for heterosis, that is, dominance). This model is shown in **Figure 1**. Variation due to this locus is a function of both the size of the deviations d and h and of the relative frequencies of the genotypes AA , Aa and aa in the population. Under Hardy–Weinberg equilibrium, we expect that if allele A has frequency p , and allele a has frequency $q = 1 - p$, then the genotypes AA , Aa and aa will have, respectively, frequencies p^2 , $2pq$ and q^2 in the population. Therefore, the mean of the population, which is the sum of the products of the deviations and their frequencies, is:

$$\mu = dp^2 + 2pqh - dq^2 \quad (2)$$

$$= dp^2 + 2pqh - (1 - 2p + p^2)d \quad (3)$$

$$= (p - q)d + 2pqh \quad (4)$$

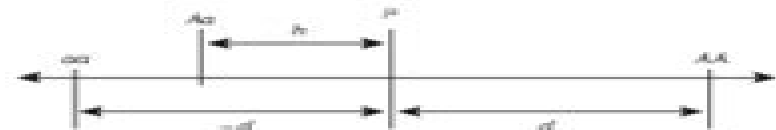


Figure 1 Deviations d and h shown as departures from population mean μ for genotypes AA , Aa and aa at a quantitative genetic locus. (Adapted from Mather and Jinks, 1977, p. 32.)

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