Chapter 18: Quantitative Genetics I – Important Concepts

Introduction

In the chapter on Mendel and Morgan, we saw how the transmission of genes from one generation to another follows a precise mathematical formula. The traits discussed in that chapter, however, were discrete traits—peas are either yellow or green, someone either has a disorder or does not have a disorder. But many behavioral traits are not like these clear-cut, have-it-or-don't-have-it phenotypes. People vary from being quite shy to very outgoing. But is shyness a discrete trait or merely a descriptive adjective for one end of a continuous distribution? In this chapter, we will discuss the genetics of quantitative, continuously distributed phenotypes.

Let us note first that genetics has made important—albeit not widely recognized—contributions to quantitative methodology in the social sciences. The concept of regression was initially developed by Sir Francis Galton in his attempt to predict offspring phenotypes from parental phenotypes; it was later expanded and systematized by his colleague, Karl Pearson[1], in the context of evolutionary theory. The analysis of variance was formulated by Sir Ronald A. Fisher[2] to solve genetic problems in agriculture. Finally, the famous American geneticist Sewell Wright developed the technique of path analysis, which is now used widely in psychology, sociology, anthropology, and other social sciences.

---

[1] After whom is named the Pearson product moment correlation.
[2] After whom the F statistic is named.

Continuous Variation

Continuous variation and a single locus

Let us begin the development of a quantitative model by considering a single gene with two alleles, *a* and *A*. Define the *genotypic value* (aka *genetic value*) for a genotype as *the average phenotypic value for all individuals with that genotype*. For example, suppose that the phenotype was IQ, and we measured IQ on a very large number of individuals. Suppose that we also genotyped these individuals for the locus. The genotypic value for genotype *aa* would be the average IQ of all individuals who had genotype *aa*. Hence, the means for genotypes *aa*, *Aa*, and *AA* would be different from one another, but there would still be variation around each genotype. This situation is depicted in Figure 18.1.

[Insert Figure 18.1 about here]

The first point to notice about Figure 18.1 is the variation in IQ around each of the three genotypes, *aa*, *Aa*, and *AA*. Not everyone with genotype *aa*, for instance, has the same IQ. The reasons for this variation within each genotype are unknown. It would include environmental variation as well as the effects of loci other than the one genotyped.

A second important feature about Figure 18.1 is that the means of the distributions for the three genotypes differ. The mean IQ (i.e., the genotypic value) for *aa* is 94, that for *Aa* is 96, and the mean for *AA* is 108. This implies that the locus has some influence on individual differences in IQ.

A third feature of note in Figure 18.1 is that the genotypic value of heterozygote is not equal to the average of the genotypic values of the two homozygotes. The average value of genotypes *aa* and *AA* is (94 + 108)/2 = 101, but the actual genotypic value of *Aa* is 96. This indicates a certain degree of *dominant gene action* for allele *a*. Allele *a* is not completely dominant; otherwise, the genotype value for *Aa* would equal that of *aa*. Hence, the degree of dominance is incomplete.

A fourth feature of importance is that the curves for the three genotypes do not achieve the same height. This is due to the fact that the three genotypes have different frequencies. In the calculations used to generate the figure, it was assumed that the allele frequency for *a* was .4 and the frequency for *A* was .6, giving the genotypic frequencies as .16 (*aa*), .48 (*Aa*), and .36 (*AA*). Consequently, the curve for *Aa* has the highest peak, the one for *AA* has the second highest peak, and that for *aa* has the smallest peak.

A final feature of note is that the phenotypic distribution of IQ in the general population (the solid line in Figure 18.1) looks very much like a normal distribution. The phenotypic distribution is simply the sum of the distributions for the three genotypes. For example, the height of the curve labeled "Total" when IQ equals 90 is the distance from the horizontal axis at 90 to the curve for genotype *aa* plus the distance from the horizontal axis at 90 to the curve for genotype *Aa* plus the distance from the horizontal axis at 90 to the curve for genotype *AA*. Often social scientists mistakenly conclude that the phenotypic distribution must be trimodal because it is the sum of three different distributions.[3]

---

[3] The phenotypic distribution *may* be trimodal, but it will be so only when the means for the three genotypes are very, very different. When single genes exert only a small influence on a phenotype, then

The gene depicted in Figure 18.1 is currently termed a *QTL* for *Q*uantitative *T*rait *L*ocus. Behavioral genetic research devotes considerable effort toward uncovering QTLs for many different traits—intelligence, reading disability, various personality traits, and psychopathology. The mathematical models that quantify the extent to which a QTL contributes to trait variance are not necessary for us to know.

Continuous variation and multiple loci

It is unlikely that one and only one gene contributes to individual differences in IQ, so let us examine the influence of a second genes. Suppose that we could genotype people at another locus for IQ, say the *B* locus with its two alleles, *b* and *B*. We would now have nine genotypic values as illustrated in Table 18.1. Once again, we would compute the mean IQ score for all those with a genotype of *aabb* and then enter this mean into the appropriate cell of the table. Next we could compute the genetic value for all those with genotype *Aabb* and enter this value into the table and so on. The results would—hypothetically at least—be similar to the data given in Table 18.1.

[Insert Table 18.1 about here]

We could also draw curves for each genotype analogous to the curves depicted in Figure 18.1. This time, however, there would be nine normal curves, one for each genotype. We could continue by adding a third locus with two alleles. This would give 27 different genotypes and 27 curves. If we could identify each and every locus that contributes to IQ, then we would probably have a very large number of curves. The variation within each curve would be due to the environment.

---

the phenotypic distribution can appear quite smooth as the present example suggests.

This model is equivalent to he polygenic model introduced in Chapter 6 during the discussion of DCG. At the present time, we cannot identify all the genes for a polygenic phenotype, calculate the types of data in Table 18.1 or plot the data ala Figure 18.1. Hence, these tables and figures are useful for *understanding* quantitative genetics, but they cannot be used for any *practical application* of quantitative genetics to IQ.

Important Quantitative Concepts

Introduction

The following sections are the core of this chapter. They explain in English the six important concepts in quantitative behavioral genetics: (1) heritability, (2) environmentability, (3) genetic correlation, (4) environmental correlation, (5) gene-environment interaction, and (6) gene-environment correlation. Although these six are described at a conceptual level, it is important to recognize that behavioral geneticists try to quantify each of them—i.e., arrive at an actual number to estimate these six quantities and then judge how important this quantity is for a behavioral phenotype. The next chapter presents an introduction to the estimation and testing of these quantities. What follows is an introduction to the concepts behind these six quantities.

I. Heritability and environmentability

The concepts of heritability and environmentability of polygenic traits are central to quantitative analysis in behavioral genetics. Instead of providing formal definitions of these terms, let us begin with a simple thought experiment and then discover the definitions through induction.

Imagine that scores on the behavioral trait of impulsivity are gathered on a

population of individuals. These observed scores will be called the *phenotypic values* of

the individuals. Assume that there was a futuristic genetic technology that could

genotype all of the individuals in this population for all the loci that contribute to

impulsivity. One could then construct a *genotypic value* for each individual. Just as with

one or two loci, the genotypic value for a polygenic trait is defined as the mean

phenotypic value of all those individuals with that genotype in the population. For

example, if Wilbur Waterschmeltzer's genotype for impulsivity is AaBBCCddEeff and

the mean impulsivity score for all individuals in the population who have genotype

AaBBCCddEeff is 43.27, then Wilbur's genotypic value is 43.27.

Imagine another technical advance that would permit us to calculate and quantify

all the environmental experiences in a person's life that could contribute to the person's

level of impulsivity. This would be the *environmental value* for an individual. We

would now have a very large set of data, part of which is illustrated in Table 18.2.

[Insert Table 18.2 about here]

From the data in this hypothetical table, we would compute a correlation

coefficient between the genotypic values and the phenotypic values. Recall that the

square of the correlation coefficient between two variables gives the proportion of

variance in one variable attributable to (i.e., predicted by) the other variable.

Consequently, if we square the correlation coefficient between the genotypic values and

the phenotypic values, we would arrive at the proportion of phenotypic variance

predicted by (or attributable to) genetic variance. This quantity, the square of the

correlation coefficient between genotypic values and phenotypic values, is called

heritability[4].

Thus, heritability is a quantitative index of the importance of genetics for

individual differences in a phenotype. Strictly defined, heritability is *the proportion of*

phenotypic variance attributable to or predicted by genetic variance. Because

heritability is a proportion, it will range from 0 to 1.0. A heritability of 0 means that

genes to no contribute to individual differences in the trait, whereas a heritability of 1.0

means that trait variance is due solely to heredity. A less technical view would define

heritability as *a measure, ranging from 0 to 1.0, of the extent to which observed*

individual differences can be traced in any way to genetic individual differences.

Heritability is usually denoted as h^2, a convention that we will adopt from now on.

Just as we could compute a correlation between genetic values and phenotypic

values, we could also compute correlations between environmental values and

phenotypic values. Squaring this correlation would give us the *environmentability* of the

trait. Environmentability has the same logical meaning as heritability but applies to the

environment instead of the genes. *Environmentability is the proportion of phenotypic*

variance attributable or predicted by environmental variance. It is also *a quantitative*

index, ranging from 0 to 1.0, of the extent to which environmental individual differences

underlie observable, phenotypic individual differences. We will denote

environmentability as e^2.

---

[4] Two assumptions are necessary to define heritability (and later, environmentability) this way. First, it is assumed that the genotypic values are uncorrelated with the environmental values. Second, there is no statistical interaction between genotypic values and environmental values. These assumptions will be discussed later in the chapter.

II. Genetic correlations and environmental correlations

You took the SAT or the ACT to enter college, and you received two subscale scores, one for verbal and the other for quantitative. If a behavioral geneticist were performing a twin study of the SAT or ACT s/he could report heritability (h^2) and environmentability (e^2) for the verbal section and heritability (h^2) and environmentability (e^2) for the verbal section. There is nothing the matter with that, but much more information can be gained by analyzing both phenotypes simultaneously. In this case—i.e., when more than one phenotype is analyzed at the same time—it is termed *multivariate genetic analysis*.

The extra information from multivariate genetic analysis consists of the *genetic correlation* and the *environmental correlation* among the traits. To discover the meaning of these quantities, examine Table 18.3 which updates Table 18.2 to depict two phenotypes—SAT verbal and SAT quantitative.

[Insert Table 18.3 about here]

Once again, we can compute the correlation between the genetic values for verbal and phenotype values for verbal (i.e., h_V) and square this quantity to estimate the heritability for verbal scores (h_V^2). Or, we could square the correlation between the environmental values for quantitative and the phenotypic scores for quantitative to arrive at the environmentability of quantitative (e_Q^2). The genetic correlation, usually denoted by r_g, consists of the *correlation between the genotypic values for two traits*. That is, for the present example,

$r_g = \text{corr}(G_V, G_Q).$

The genetic correlation tells us the extent to which the genotypic values for one trait predict the genotypic values for the second trait. It has the same meaning as any correlation; the only difference is that it applies to genotypic values. Hence, a genetic correlation of 0 implies that the two sets of genotypic values are statistically independent of each other; one cannot predict the genetic values of one trait by knowing the genetic values of the other trait. A genetic correlation approaching 1.0 implies strong predictability; in this case, knowing the genotypic values of one trait strongly predicts the genotypic values of the other trait[5].

The environmental correlation, or r_e, has an analogous definition—it is the *correlation between the environmental values of the two traits*, or, in terms of the current example,

$$r_e = \text{corr}(E_V, E_Q).$$

The environmental correlation informs us how well environmental values for one trait predict environmental values for the other trait.

III. Gene-environment interaction

In nonscientific discussions about the importance of genes in human behavior, we behavioral geneticists often encounter the attitude best described in a quote to the author at a party—"It is not the gene and it is not the environment that is important. It is the *interaction* between the gene and the environment that is crucial." Indeed, the notion of interactionism has been raised almost to the status of dogma in many circles. This is not necessarily bad. But neither is it good, because the simple phrase "gene-environment

---

[5] It is tempting to interpret genetic correlations in terms of the number of genes two traits have in common. However, the situation is more complicated than that (see Carey, 1988).

interaction" has equivocal meetings and the usefulness of interactionism depends on which meaning is being used.

The first and the generic meaning of gene-environment interaction—and the one most often used in the nonscientific literature—defines it as *the fact that both genes and environment contribute to behavior*. This is the *lemonade* concept, the theme of which is central to this book. The second meaning of the term is found most often in the scientific literature within behavioral genetics and interprets the phrase "interaction" in a statistical sense. This meaning implies that the *actual relationship between the environment and a phenotype depends upon the genotype, or equivalently, the actual relationship between a genotype and a phenotype depends on the environment*. The first definition (i.e., lemoade) will be referred to as the *loose* definition and the second as the *strict* or *statistical* definition. For those of you aware of the concept of interaction in the analysis of variance (ANOVA), these definitions are meaningful. For those of you who suspect that ANOVA is a committee devoted to abolishing the NOVA series on PBS, these definitions will be vague and vacuous, so perhaps an example will help.

An Example of gene-environment interaction.

Asian-Americans in both California (Klatsky et al., 1983) and Hawaii (Johnson et al., 1984) who were born in Asia drink much less alcohol than their sons and daughters who were born and raised in the US. The difference in alcohol consumption between the parental and offspring generation is likely to be culture and environment because drinking in most Asian countries follows the same abstemious quality brought over by the immigrants.

We have also learned how the polymorphism for a form of the enzyme aldehyde dehydrogenase (ALDH-2) influences individual differences in alcohol consumption among Asians, even those raised in the US (see Chapter 5). Those with a deficient DNA blueprint for ALDH-2 become ill after consuming alcohol, so their risk of developing alcohol abuse and dependence is diminished.

Hence, there is good evidence that both a known gene and a known environmental factor (enculturation in Western society) influence individual differences in alcohol consumption among Asian-Americans. Clearly, this is an example of gene-environment interaction in the loose sense of the term—in *lemonadish* terms, both the ALDH polymorphism and growing up in the US contribute to phenotypic differences in alcohol consumption.

But whether this involves a *statistical* gene-environmental interaction cannot be resolved from these data. To illustrate the point, consider a study that would measure alcohol consumption and ALDH genotypes in both the immigrant and USA-born generations of an Asian-American community. The results would look like either of the two graphs depicted in Figure 18.2.

[Insert Figure 18.2 about here]

The graph on the left-hand side portrays gene-environment interaction in the loose sense. In both the older and the younger generations, ALDH-2 genotypes predict alcohol use—those with the active enzyme drink more than those with the inactive (deficient) form do. Culture has also influenced alcohol use—the younger generation drinks more on average than their parents do. In ANOVA terms, this graph suggests a main effect for ALDH genotype and a main effect for generation. However, the two lines in left panel of

Figure 18.2 are parallel. Hence, there is no *statistical* interaction between genotype and

generation, and hence, no gene-environment interaction in the *statistical* sense of the

term.

The graph on the right illustrates gene-environment interaction in the strict sense.

Here, the two lines are no longer parallel. (Indeed, any statistical interaction involves

testing whether the lines are parallel or not. If the two lines are within sampling error of

being parallel, then there is no statistical interaction. When they are not parallel, then

there is a statistical interaction.) In the older generation, ALDH weakly predicts alcohol

use; the line is close to being flat and there is not much difference in drinking between

those with and those without the active enzyme. In the younger generation, however,

ALDH is a much stronger predictor of alcohol use. This illustrates the phrase "the actual

relationship between a genotype and a phenotype depends on the environment" in the

definition of statistical gene-environment interaction. In this case, the relationship

between ALDH and drinking depends on whether the person was a recent immigrant

from Asia (the older generation) or a person raised in a Western environment (the

younger generation).

How Important is Gene-Environment Interaction?

The answer to this question depends upon which definition of gene-environment

interaction is used. As we learned in Chapter 1, gene-environment interaction in the

loose, lemonade sense of the phrase is *always* important for human behavior. MZ twins

always correlate less than 1.0, so there must always be an important environmental

contribution to human behavior. Also, almost all human behaviors demonstrate some

degree of heritability, so genes are important. Hence, the lemonadish concept that genes and environment *both* contribute to individual differences in human behavior is as close to a universal statement as one can get in the behavioral sciences.

Unfortunately, we have no real idea of how important gene-environment interaction in the statistical sense is for human behavior. The reason is not for want of trying or lack of theory. The biggest problem is a lack of technology to gather adequate empirical data to examine interactions. To test for an interaction, it is necessary to directly measure genotypes or environments (preferably both). However, the ability to do extensive human genotyping is only a recent phenomenon. Also, the environment, especially aspects of family life, is not nearly as easy to measure as one might suspect. Many putative environmental aspects of a household may be more reflections of parental phenotypes than causal environmental inputs into a child's behavior. The classic example of the number of books in a house—often found to correlate with child's academic achievement—may be a better index of parental intelligence and interest in reading than a direct and causal environmental influence on the children.

IV. Gene-environment correlation

Gene-environment correlation occurs when people with high genetic values for a trait experience environments with high values for the trait. The converse, of course, is also true—people with low genetic values experience environments with low values.[6] The effect of such GE correlation is to increase phenotypic variance, i.e., to create a wider range of individual differences.

---

[6] Logically, it is possible to have a negative gene-environment correlation, but few researchers concern themselves with this.

Several different mechanisms can produce GE correlation, two of which will be described here. The first is the joint transmission of genes and environments within families. For example, consider a husband and wife who are quite intelligent themselves and also have considerable intellectual interests. In addition to providing their offspring with favorable genes for intelligence, they may also foster reading skills, curiosity, inquisitiveness, and a host of other factors that environmentally promote high intelligence in their offspring. Couples with lower than average intelligence may provide less favorable environments to their offspring. The result would be GE correlation that Plomin, DeFries & Loehlin (1978) have termed *passive* GE correlation.

A second mechanism is self-selection or environments (Scarr & McCartney, 1983), and it generates what Lindon Eaves[7] (personal communication) has dubbed the *smorgasbord model* of GE correlation. A smorgasbord is a buffet of different breads, cheeses, vegetables, fishes, meats, etc. On the first pass, most people sample a little of everything, but on the second pass, they return for those dishes that they found most tasty. In the course of development, most of us experience a wide variety of different people, academic subjects, work activities, past-times, hobbies, etc.. These are equivalent to the first pass through the buffet. Those people whom we enjoy associating with become our friends, those academic subjects that perked our interest become our majors, those work activities we found enjoyable become our careers, and so on. This is the behavioral equivalent of the second pass through the smorgasbord. If genes influence the type of people we find rewarding to be around (and being around these people alters our

---

[7] The exact origin of the phrase "smorgasbord model" is obscure. It first came into this author's awareness in casual conversation when Lindon Eaves graciously provided him with a ride home, but the concept had

behavior), if genes influence the type of academic subjects we find interesting (and

pursuit of those topics alters our subsequent behavior), and if genes influence the work

we find enjoyable (and if that work causes subsequent changes in our behavior), then GE

correlation will be induced.[8]

How important is GE correlation?

Many of the statements about the effects of GE interaction apply to GE

correlation. There is considerably more theoretical writing about GE correlation than

there are empirical data on the issue. Once again, the lack of data does not reflect a lack

of effort. Rather, there are two major problems in gathering data on GE correlation. The

first is that longitudinal data are required during the active phase of a mechanism that

induces GE correlation. For example, the association between adolescent peer groups

and delinquency is a ripe area for exploring the extent to which such peer groups are self-

selected by teenagers already predisposed towards antisocial behavior versus the extent to

which the peer groups themselves foster problem behavior. The longitudinal twin data

that could help to answer this question are just being gathered. The second and most

difficult problem is the one shared with gene-environment interaction—finding good

measures of the environment.

In a highly underreferenced[9] paper, Eaves et al. (1977) note that the *existence* of

both GE interaction and GE correlation is less important than the specific mechanism that

generates the interaction or correlation and the parameters of the situation before that

---

been extensively bantered about in tea-time conversations involving Lindon Eaves, Nick Martin, Andrew
Heath, Jeffrey Long, and this author.
[8] Self-selection mechanisms are termed "active" GE correlation by Plomin et al. (1978).

mechanism entered the picture. For example, if the correlation between the environments

of relatives (η, in this model) is very low to begin with, then GE interaction can lower the

correlation among relatives and lead to an underappreciation of genetic influences. On

the other hand, if η is low and the smorgasbord model of GE correlation applies, then GE

correlation is included in the estimate of heritability. Here, one may underestimate the

importance of the environment. Finally, if η is low and both GE interaction and the

smorgasbord GE correlation mechanisms begin, then perhaps their statistical impacts will

cancel each other out. Confusing? It should be to the novice in behavioral genetics. The

real challenge of GE interaction and GE correlation does not lie in finding numerical

values that estimate these concepts. Rather, it resides in identifying the *mechanisms* that

generate these statistical quantities in the first place.

The Twin and Adoption Methods

Family Correlations

Even with the marvelous technology of modern genetics, it is not possible to

directly measure genotypic values for polygenic traits. And it stretches imagination to

suppose that we can measure environmental values for all those varying factors that

influence a trait. Instead, we observe only phenotypes in relatives.

Table 18.4 illustrates the type of data that behavioral geneticists gather. The

family is the unit of observation and the phenotypic scores for the different classes of

relatives are the variables. For the data in Table 18.4, we would compute the correlation

---

[9] Underreferenced in the sense that few contemporary researchers refer to that paper any more, not in the
sense that it contains few references.

between the variables "parental IQ" and "child IQ" giving a parent-offspring correlation[10].

[Insert Table 18.4 about here]

But correlations among the relationships in ordinary nuclear families cannot be used to estimate heritability. Behavioral similarity between, say, parents and offspring, may be due to any of three factors: (1) shared genes; (2) shared environments; and (3) some combination of shared genes and shared environments. Consequently, behavioral scientists usually study two special types of relatives to tease apart the influence of shared genes from that of shared environment. These two special populations are twins and adoptees. Each is discussed in turn.

The Twin Method: Rationale

Monozygotic (MZ) or *identical* twins are the result of the fertilization of a single egg. The cells from this zygote[11] divide and divide, but early in the course of development, some cells physically separate and begin development as an independent embryo. The reasons for the separation are currently unknown. Because the two individuals start out with the same genes, they are effectively genetic clones of each other and any differences between the members of an identical twin pair must be due to the environment. Included in the environment is the fact that one twin may have developed from more cells than the other since it is suspected that the original separation is seldom

---

[10] The reader familiar with data analysis should realize that because families do not have the same number of offspring, family data is usually not "rectangular." There are methods to take care of such data sets but they are too advanced for this text. The interested reader should consult Neale and Cardon (1992).
[11] A *zygote* is "scientificese" for a fertilized egg.

an equal 50-50 split. MZ twins look so alike that people who do not know them well

often confuse them.

Dizytogic (DZ) or *fraternal* twins result when a woman double ovulates and each

egg is independently fertilized. Genetically, DZ twins are as alike as ordinary siblings,

sharing on average 50% of their genes, and look alike as ordinary brothers and sisters

Differences between the members of a fraternal twin pair will be due to both the

environment and also to the different alleles that each member inherits.

Consequently, the logic of the twin method is quite simple. If genes contribute to

a trait, then MZ twins should be more similar to each other than DZ twins. Thus, the

striking physical similarity of MZ twins in terms of height, facial features, body shape,

hair color, eye color, etc., suggests that genes influence individual differences in these

traits because fraternal twins are as alike in their physical features as ordinary siblings.

The Twin Method: Assumptions

The central assumption of the twin method is often called the *equal environments

assumption*. This assumption states that *environmental factors do not make MZ twins

more similar than they make DZ twins similar*. To violate this assumption, two very

important phenomenon must *both* occur: (1) environmental factors must treat MZ twins

more similarly than DZ twins; and (2) that similarity in treatment *must* make a difference

in the *phenotype under study*. An example can help to illustrate. Parents often dress

identical twin children in similar outfits. All of us have seen a pair of identical twin girls

outfitted in the same dress or a pair of young MZ boys both wearing a sailor suit. Parents

frequently dress their DZ twins in identical attire but not nearly with the frequency of

parents with MZ twins. Consequently, if the phenotype under study were "fashion in young children," then the equal environments assumption would be violated and we should not use the twin method to estimate heritability.

Let us take this example a bit further. Suppose that the phenotype under study was adult shyness. The first facet of the equal environments is violated because the MZ twins in the sample will have been dressed more alike as children than the DZ twins. However, the second facet of the equal environments assumption—that similarity in treatment makes a difference in the phenotype under study—would probably not be true. If it were true, then being dressed as a child in, say, a cowboy outfit as opposed to a sailor suit would have an important influence on adult shyness. Hence, for the phenotype of childhood fashion, the equal environments assumption would be violated, but for the phenotype of adult shyness, the assumption may be valid.

Potential difficulties with the equal environments assumption begin shortly after fertilization. Because they result from independent fertilizations, DZ twins develop separate umbilical cords, amniotic sacks, chorions,[12] and placentas. (In a significant proportion of DZ twins, particularly those where each member implants close to the other, the placentas will fuse together during development, leading to a single afterbirth.) Consequently, DZ twins are always dichorionic and diamniotic. Although they may be crowded in the womb, they usually have independent blood supplies from the mother.

The intrauterine status of MZ twins, however, depends on the time of the splitting of the blastomere. When the split is very early, the twins may implant separately in the

---

[12] Two separate "sacks" enclose a fetus. The first of these is the amniotic sack containing the amniotic fluid and the second is the chorionic sack surrounded by a layer of cells referred to as the chorion.

uterus and follow the same developmental pattern of DZ twins—separate amniotic sacks, chorions, and placentas.[13] This pattern occurs in about 25% to 35% of MZ twins. When the split occurs later, then the two twins develop within a single chorion and are called *monochorionic*. The remaining 65% to 75% of MZ twins follow this pattern. When the split is very late, then the monochorionic twins may actually share a single amniotic sack.[14]

The result of these intrauterine differences between MZ and DZ twins is poorly understood. At first glance, it may appear that development within the same chorion might lead to greater MZ twin similarity, but some twin experts argue that it might also increase differences between MZ twins. Often, development within a single chorion leads to crowding and unequal distribution of blood to the twins. Unfortunately, there are few empirical data on this topic. Sokol et al. (1995) report that monochorionic MZ twins were more similar than MZ dichorionic twins on some childhood personality measures but not on measures of cognitive ability. Perhaps intrauterine effects are trait-dependent.

The second major way in which the assumption may be violated is in parental and peer treatment of MZ and DZ twins. Here, empirical data on the equal environments assumption suggests that the assumption is very robust. That is, for most substantive human behaviors studies thus far, the effects of violating the assumption are very minor. It is quite true that as children MZ twins are often called by rhyming or alliterative names (e.g., Johnnie and Donnie), that they are dressed alike more frequently than DZ twins,

---

[13] Like DZ twins, the two placentas may fuse during development.
[14] The fact the DZ twins are always dichorionic lead to the erroneous conclusion among many obstetricians that fraternal twins always had two afterbirths while identical twins only had a single afterbirth. Not long ago, I interviewed a mother of an opposite-sex DZ pair who swore that her son and daughter were identical twins because the doctor told her so on the basis of a single afterbirth.

and that in general parents treat them more as a unit than they do fraternal twins. However, several different types of data suggest that this treatment does not influence substantive phenotypic traits later in life.

The first line of evidence is that *actual* zygosity predicts behavioral similarity better than *perceived* zygosity (Scarr , 1968; Scarr and Carter-Saltzman, 1979). In the past, many parents of twins were misinformed or made erroneous conclusions on their own part about the zygosity of their offspring[15]. Consequently some parents raised their DZ twins as MZ twins while other treated their MZ offspring as DZ pairs. Their biological zygosity rather than their rearing zygosity better predict the behavioral similarity of these twins.

A second line of evidence relies on the fact that even though *on average* parents of MZ children treat them more alike than parents of DZ children, there is still strong variability in the way parents of MZ pairs treat their children. Some parents accentuate their MZ offspring's similarity by making certain that they have the same hairstyle, clothing, brand of bicycle, etc. Other parents will actually go out of their way to avoid treated their MZ children as a unit and deliberately try to "individualize" them. However, those MZ twins treated as a unit were no more similar in their adolescent and adult behavior than those who were deliberately individualized (Kendler et al., 1993; Loehlin and Nichols, 1976).

The final and best line of evidence comes from studies of twins raised apart. These twins are not raised in completely random environments, but they are certainly not

---

[15] A persistent myth, held even by some MDs, was that identical twins have one afterbirth while fraternal twins have two afterbirths. What is true is that DZ twins always have two chorions (a sac enclosing the

subject to the subtle treatments of being dressed alike as those twins who are raised day-

in and day-out in the same household for all their childhood and early adolescence. As

one scholar of twins raised apart, James Shields, put it, "The importance of studying

separated twins is to demonstrate that the microenvironment of daily living in the same

household is not solely responsible for the great similarity observed in twins raised

together" (Shields, personal communication, 1976).

If sibling resemblance were due mainly to the environment and if violation of the

equal environments assumption were the major reason why MZ twins raised together

correlated higher than DZ twins raised together, then two predictions can be made about

separated twins. First, the correlation for separated twins should be small and close to 0;

it should certainly be less than the correlation for siblings raised together. Second, the

correlation for MZ twins raised apart should be no different than the correlation for DZ

twins raised apart. The available data on twins raised apart are inconsistent with both of

these predictions (Bouchard et al., 1990). First, for almost all traits that have been

studied, the correlations for twins raised apart have been substantial and significant.

Second, MZ twins raised apart are consistently *more* similar than biological siblings and

DZ twins who are raised together. Finally, MZ twins raised apart correlate higher than

DZ twins raised apart.

Taken together, all these lines of evidence suggest that the equal environments

assumption meets the definition of a robust assumption. A *robust assumption* is one that

might actually be violated, but the effect of violating the assumption is so small that the

estimates and substantive conclusions are not altered. For example, Newtonian physics is

---

amnion and amniotic fluid) while MZ twins may have either one or two chorions. Either type of twins can

incorrect, but one can use Newtonian principles to build a bridge or design a skyscraper. In these situations, the assumptions of Newtonian physics are robust even though they are technically wrong.

The Adoption Method: Rationale

The logic of the adoption method is as simple as the logic of the twin method, provided that nonfamilial adoptions are used. When parents adopt and raise a child to whom they are not genetically related, any similarity between the parents and child must have something to do with the environment. Similarly, when there are two adoptive children raised in the same family, then sibling resemblance between the two must also be environmental in nature.

When children are adopted shortly after their birth, then shared genes are the only reason they would show similarity with their biological relatives. Thus, correlations between adoptees and their genetic relatives give evidence for heritability.

The Adoption Method: Assumptions

There are two critical assumptions about the adoption method—the absence of selective placement and the representativeness of the adoptive families. Selective placement occurs most often when adoption agencies deliberately try to place adoptees with adoptive parents who resemble the adoptee's genetic parents. Like the equal environments assumption in the twin method, the critical issue is not whether selective placement occurs—it does—but whether the selective placement influences the trait in question.

---

have one or two afterbirths.

Some contemporary adoption studies report strong selective placement for race/ethnicity and for religion. Placement for religion is seldom done deliberately. It is mostly a secondary consequence of different religious denominations supporting their own adoption agencies. Catholic Social Services, for example, deal mostly with Catholic unwed mothers and place children into Catholic homes. Similar venues occur for other religiously affiliated agencies. Hence, one must be cautious in interpreting adoption data on phenotypes that may correlate strongly with religious affiliation (e.g., attitudes toward abortion.)

There is moderate selective placement for physical characteristics, especially height. The rationale here is to avoid placing a child into home where the child might "stick out like a sore thumb." For behavioral traits, the empirical evidence suggests selective placement is usually—but not always—small or nonexistent. It is hard to make generalizations about selective placement for behavioral traits because different adoption studies work with different adoption agencies. For example, selective placement on parental education was weak in two recent studies, one from Texas (Horn, Loehlin & Willerman, 1979) and the other from Colorado (Plomin & DeFries, 1985), yet was significant in a third study from Minnesota (Scarr & Weinberg, 1978, 1994). Typically adoption researchers test for selective placement and, if it is present, adjust their statistical methods to account for it.

The second assumption about the adoption method concerns the representativeness of the adoptive families. Adoptive parents are screened—sometimes intensively—on issues of positive mental and physical health, the ability to financially support a child, and the probability of providing a safe and secure home for the child.

Researchers mistakenly assume that the screening process is *for* wealth, *for* positive

mental health, etc. Instead, the process is *against* extreme poverty and *against* serious

psychopathology. As a result, mean income of adoptive families is not very different

from average income in the general population—it is just that the lower tail of the income

distribution is missing.

Selection against psychopathology is a more serious matter. Parental alcoholism,

criminal behavior, psychosis, drug abuse, and several other factors can exclude a family

from adopting a child. As a result, there may be a restriction in range in the

environments provided by adoptive parents, making it very difficult to detect a

correlation between adoptees and their adoptive relatives. Hence, one should be cautious

in interpreting low correlations among adoptive relatives as evidence for a lack of family

environmental influence on the trait. Once again, one must consider restriction in range

on a trait by trait basis. It may be very important for phenotypes like antisocial behavior,

but rather weak for personality traits.

The Family Environment

The behavioral genetic definition of "family environment" sows untold confusion

among social scientists, so it is important to discuss it. According to almost all social

science research, the term "family environment" refers to the physical, psychological, and

social state of the household and the members within it. Physical attributes include the

physical area of the house, number of books or computers, adequacy of provisions, etc.

Psychological variables tap such constructs as parental warmth, psychopathology, and

sibling interactions, while social variables measure parental education, religion, etc. This definition of family environment is termed the *substantive* definition.

In contrast, the definition of family environment used in behavioral genetic research is a *statistical* definition[16]. According to this definition, the family environment consists of *all those factors that make relatives similar on a phenotype*. The phrases "all those factors" and "make relatives similar" are very important. *Both* of these must be present for a causal factor to be considered part of the statistical family environment.

Literally, the term "all those factors" does not restrict causal variables from physically occurring inside the family unit. If living in the same neighborhood makes siblings similar to one another, then the factor of "living in the same neighborhood" is part of the statistical family environment. Other factors relevant for siblings might include going to the same (or very similar) schools, having overlapping groups of friends, and sharing the same religion.

Likewise, the phrase "make relatives similar" is crucial for understanding the statistical family environment. If siblings share the same religion but religion does not make siblings similar on the personality trait of sociability, then "sharing the same religion" is *not* part of the statistical family environment for sociability.

---

[16] The statistical definition has its origin in ANOVA techniques developed for genetic analysis in agronomy. As applied to humans, each human family would be a single cell in a very large one-way ANOVA. The scores for the individuals within a family are the within-group numbers for a cell in the ANOVA. Hence, the within-family variance component reflects all those factors that make relatives of a family different from one another. The between-group variance component taps factors that make members of a family similar to one another but different from other families. With genetically informative designs (twins, adoptees), one can estimate a within-family environmental variance component and a between-family environmental variance component (see Jinks & Fulker, 1970). The between-family environmental variance is the quantity that behavioral geneticists have taken to calling the "family environment."

Hence, if some factor physically occurs within the family household but does not make siblings similar, then it is *not* part of the statistical family environment. For example, consider Papa Smith, a rather authoritarian and argumentative chap whose in-your-face style and rigid views of right and wrong influence both of his sons. Aaron Smith, something of a chip off the old block, argues right back and the continual verbal tete-a-tete between him and Papa reinforce Aaron's tendency to express his views openly. Zeke Smith, on the other hand, is very intimidated by Papa and becomes submissive in his public persona. In this scenario, Papa Smith is definitely part of the family environment (in the substantive use of the term) of the Smith brothers. However, Papa is not part of the statistical family environment for Aaron and Zeke because his influence is to make them phenotypically different from each other. Although Papa is a shared experience for Aaron and Zeke, he does not make them similar.

For some phenotypes—most notably personality traits—the effects of the statistical family environment for siblings are very small. This finding has often been interpreted as implying that parents have no effect on their children's personality. Although this conclusion *may* be true, the data are not sufficient to prove it. All the data say is that the net effect of being raised in the same family does not, on average, make siblings similar to one another in personality. For each Papa Smith in a population there may be a corresponding Papa Jones whose influence on his children is to make them similar. The differences induced by the Papa Smiths may cancel out the similarities created by the Papa Joneses resulting in a negligible statistical family environment. The effect of the *substantive* family environment, on the other hand, may be quite important for personality development.

Just as social scientists sometimes use the term *nonfamily environment* to refer to factors outside the household and its members, behavioral geneticists will use that term but again in a statistical sense. The statistical nonfamilial environment is defined as *all those factors that make relatives different from one another*. Papa Smith is part of the statistical nonfamily environment. If the neighborhood peer group makes siblings similar for juvenile delinquency, then they are part of the statistical family environment but the substantive nonfamily environment.

At this point, the insightful reader might ask why behavioral geneticists have not developed a new vocabulary to avoid the equivocation and all the resulting confusion. The short answer is that behavioral geneticists have indeed used different terminology and have tried to be explicit about their definitions, but the subtle differences in meaning have not caught on in the broader social science literature. Table 18.5 presents the statistical concepts, their definitions, and the terms used most often in behavioral genetic research to refer to these statistical quantities. It is important to commit them to memory. Both the general literature in behavioral genetics and the remaining chapters in this book will use these terms.

[Insert Table 18.5 about here]

Comments on Heritability and Environmentability Estimation

In the next chapter we will learn simple methods for obtaining numerical estimates of the concepts described above as well as the assumptions behind the estimation process. Before embarking on that, we must gain insight into the accuracy of

these quantitative estimates and the various factors that influence them. The important

points about heritability and environmentability arelisted below.

With humans, h^2 and e^2 are measured with some accuracy but not with complete

accuracy. Temperature can be measured accurately with a thermometer. Estimations of

h^2 and e^2 have the accuracy of going outside in the middle of the day and guestimating the

temperature from the warmth or coldness without a thermometer. On a frigid, winter day

you may be hard pressed to distinguish a temperature of 0 degrees F from 10 degrees or

even –10 degrees, but you know darn well that it is not 40 degrees, 70 degrees or 100

degrees. On a blistering summer day, you may not be able to tell 95 degrees from 105

degrees, but you can estimate with confidence that it is not 70 degrees, 40 degrees or 0

degrees. Heritability and environmentability cannot be estimated well to second digit but

they can be placed into categories of low (arbitrarily taken here as 0 to .30), moderate

(.30 to .60), and high (.60 to 1.0).

Heritability and environmentability are population concepts and statistics that

apply only weakly to individuals. An h^2 of .40 for achievement motivation does *not* mean

that 40% of your achievement motivation is due to your genes and the remaining 60%

was generated by your environment. Your own level of achievement motivation could be

determined almost completely by your genes or almost completely by your environment

and yet be totally consistent with an h^2 of .40. The heritability simply means that

averaged over an entire population, 40% of observed individual differences in

achievement motivation are attributable to genetic individual differences. For similar

reasons, an extremely deviant score for a heritable does not necessarily imply that a

person is a genetic "deviant." Antisocial behavior has a moderate heritability, but that

fact alone does not imply that a chronic felon has an extreme genotype for antisocial behavior[17].

Heritability depends on the range of environments and environmentability depends upon the range of genotypes. A simple example can illustrate this principle. Farmer Jones buys corn seed that consists of a wide variety of different genotypes. She plants each seed in the same soil, gives it the same amount of fertilizer and water, and makes certain each plant receives identical amounts of sunlight. At the end of the growing season, some corn plants are taller than others. All differences in height must be must to differences in the genotypes of the corn because each seed and plant received the same environmental treatment. The heritability of height for Farmer Jones's corn plants would be 1.0. If Farmer Smith bought corn seeds that were genetically identical but then planted them at different depths in different soils and provided them with differing amounts of water, fertilizer, and sunlight, then the environmentability for height in her crop would be 1.0. Thus, h^2 and e^2 have a yin-yang relationship. Decrease one and the other increases; increase one and the other decreases. This can lead to some counter-intuitive conclusions. Providing equal schooling for all children is a laudable social goal, but it could increase the heritability of academic achievement.

---

[17] The difference between population statistics and their predictability for an individual is difficult for those without a strong quantitative background so perhaps an analogy will help. Suppose that I gathered a random sample of 500 adult males and 500 adult females and measured their height. How much would wager that the average height of the males was significantly greater than the average height of the females? If you had no personal scruples about betting and if you knew about statistics, you should beg, borrow—but not steal—as much money as you could for your wager. The odds that you will win are greater than a billion to one. Now suppose that I picked a random male from this sample. How much would you bet that he is taller than the average for the whole sample of 1,000 people? Would you bet the farm on this? Of course not! There is much more uncertainty guessing about an individual than there is in guessing about population statistics (the mean heights of males and females).

Heritability and environmentability within populations are uninformative about differences between populations. In the above example, suppose that Farmer Smith's plants were on average 4 inches taller than Farmer Jones's corn. To what extent is this due to the genotypes of the seeds or to the environments of the corn plants? There is no way to tell. Hence, even under conditions of perfect heritability and environmentability within populations, it is not possible to determine the reasons why one population differs from another. There was significant heritability for contracting tuberculosis (TB) in the late 1800s and early 1900s and there may well heritability for it today. That is, heritability of TB may be significant in yesteryears and may also be significant today. Does that imply that the difference in TB prevalence between the initial and terminal years of the 20th century is due to genetic differences? Of course not. The great reduction in TB prevalence over the century is better explained by antibiotics and public health measures than by the death of most genetically susceptible people in the interim. To give concrete applications of this general principle, we would conclude: (1) The fact that a trait is heritable in both males and females does not imply that genes contribute to mean sex differences; and (2) heritability of a trait within ethnic groups does not imply that mean ethnic differences are due to genes.

References

Bouchard, T. J. J., Lykken, D. T., McGue, M., Segal, N. L., & Tellegen, A. (1990). Sources of human psychological differences: The Minnesota study of twins reared apart. Science, 250, 223-250.

Carey, G. (1988). Inference about genetic correlations. Behavior Genetics, 18, 329-338.

Eaves, L. J., Last, K., Martin, N. G., & Jinks, J. L. (1977). A progressive approach to non-additivity and genotype-environmental covariance in the analysis of human differences. British Journal of Mathematical and Statistical Psychology, 30, 1-42.

Eaves, L. J., Eysenck, H. J., & Martin, N. G. (1989). Genes, culture and personality: An empirical approach. San Diego CA: Academic Press.

Horn, J. M., Loehlin, J. C., & Willerman, L. (1979). Intellectual resemblance among adoptive and biological relatives: The Texas Adoption Project. Behavior Genetics, 9, 117-207.

Jinks, J. L., & Fulker, D. W. (1970). Comparison of biometrical, genetical, MAVA, and classical approaches to the analysis of human behavior. Psychological Bulletin, 73, 311-349.

Johnson, R. C., Nagoshi, C. T., Schwitters, S. Y., Bowman, K. S., Ahern, F. M., & Wilson, J. R. (1984). Further investigation of racial/ethnic differences uib flushing in response to alcohol. Behavior Genetics, 14, 171-178.

Kendler, K. S., Neale, M. C., Kessler, R. C., Heath, A. C., & Eaves, L. J. (1993). A test of the equal-environment assumption in twin studies of psychiatric illness. Behavior Genetics, 23, 21-27.

Klatsky, A. L., Siegelaub, A. B., Landy, C., & Friedman, G. (1983). Racial patterns of alcohol beverage use. Alcohol. Clin. Exp. Res., 7, 372-377.

Loehlin, J. C., & Nichols, R. C. (1976). Heredity, environment, and personality: A study of 850 sets of twins. Austin TX: University of Texas Press.

Lytton, H., Martin, N. G., & Eaves, L. (1977). Environmental and genetical causes of variation in ethological aspects of behavior in two-year old buys. Social Biology, 24, 200-211.

Neale, M. C., & Cardon, L. R. (1992). Methodology for genetic studies of twins and families. Dordrecht,Netherlands: Kluwer.

Plomin, R., DeFries, J. C., & Loehlin, J. C. (1978). Gene-environment interaction and correlation in the analysis of human behavior. Psychological Bulletin, 84, 309-322.

Plomin, R., & DeFries, J. C. (1985). Origins of individual differences in infancy: The Colorado adoption project. Orlando, FL: Academic Press.

Scarr, S. (1968). Environmental bias in twin studies. Eugenics Quarterly, 15, 34-40.

Scarr, S., & Weinberg, R. A. (1978). The influence of "family background" on intellectual attainment. American Sociological Review, 43, 674-692.

Scarr, S., & Carter-Saltzman, L. (1979). Twin method: Defense of a critical assumption. Behavior Genetics, 9, 527-542.

Scarr, S., & McCartney, K. (1983). How people make their own environments: A theory of genotype --> environment effects. Child Development, 54, 424-435.

Scarr, S., & Weinberg, R. A. (1994). Educational and occupational achievements of brothers and sisters in adoptive and biologically related families. Behav Genet, 24(4), 301-325.

Sokol, D. K., Moore, C. A., Rose, R. J., Williams, C. J., Reed, T., & Christian, J. C. (1995). Intrapair differences in personality and cognitive ability among young monozygotic twins distinguished by chorion type. Behav Genet, 25(5), 457-466.

Table 18.1 Genotypes and genotypic values for two loci that contribute to individual differences in a continuous trait.						
	bb		*Bb*		*BB*	Mean
AA	*AAbb*	101	*AABb*	106	*AABB*	111	108
Aa	*Aabb*	89	*AaBb*	94	*AaBB*	99	96
aa	*aabb*	87	*aaBb*	92	*aaBB*	97	94
Mean		93		98		103	100

Table 18.2. Hypothetical data set containing the genetic, environmental, and phenotypic values for individuals.			
	Genetic Value $= G$	Environ-mental Value $= E$	Phenotypic Value $= P$
Abernathy Abercrombie	113	96	107
Beulah Bellwacker	92	74	77
...	.	.	.
Zelda Zwackelbee	118	104	118

Table 18.3. Hypothetical data set containing the genetic, environmental, and phenotypic values for individuals on two traits.						
	Verbal			Quantitative		
	Genetic Value $= G_V$	Environ-mental Value $= E_V$	Phenotypic Value $= P_V$	Genetic Value $= G_Q$	Environ-mental Value $= E_Q$	Phenotypic Value $= P_Q$
Abernathy Abercrombie	590	620	610	630	620	620
Beulah Bellwacker	410	380	400	510	470	490
...	.	.	.			
Zelda Zwackelbee	630	540	580	520	590	540

Table 18.4. Organization of data for computing the correlation between parent and offspring.		
Family	**Parent IQ**	**Child IQ**
Athabaska	107	104
Bottomwinger	77	98
.		
Zakmeister	118	102

| Table 18.5. Terminology used in the behavioral genetic literature to refer to the statistical quantities of between-family environmental variance and within-family environmental variance.		
Statistical Quantity:	**Definition:**	**Terms used in the behavioral genetic literature:**
Between-family environmental variance (the statistical family environment)	All factors that make relatives similar to one another on a phenotype = the extent to which being raised in together makes relatives similar.	Family environment Shared environment Common environment Shared, family environment
Within-family environmental variance (the statistical nonfamily environment)	All factors that make relatives different from one another on a phenotype = the extent to which idiosyncratic experiences make relatives different.	Non-family environment Nonshared environment Unique environment Idiosyncratic environment
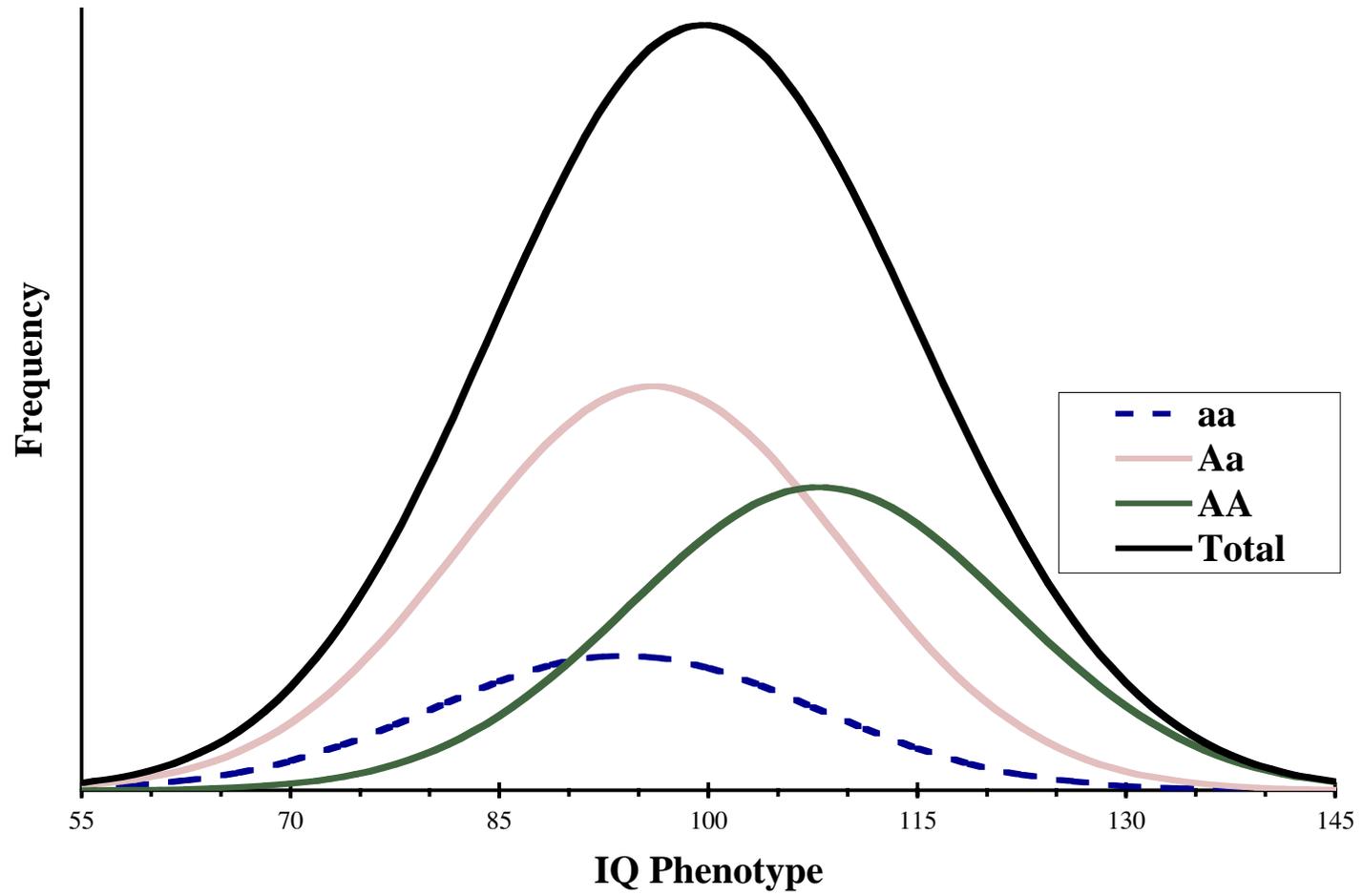
Figure 18.1. Distributions for three genotypes at a single locus and the resulting phenotypic distribution (Total).
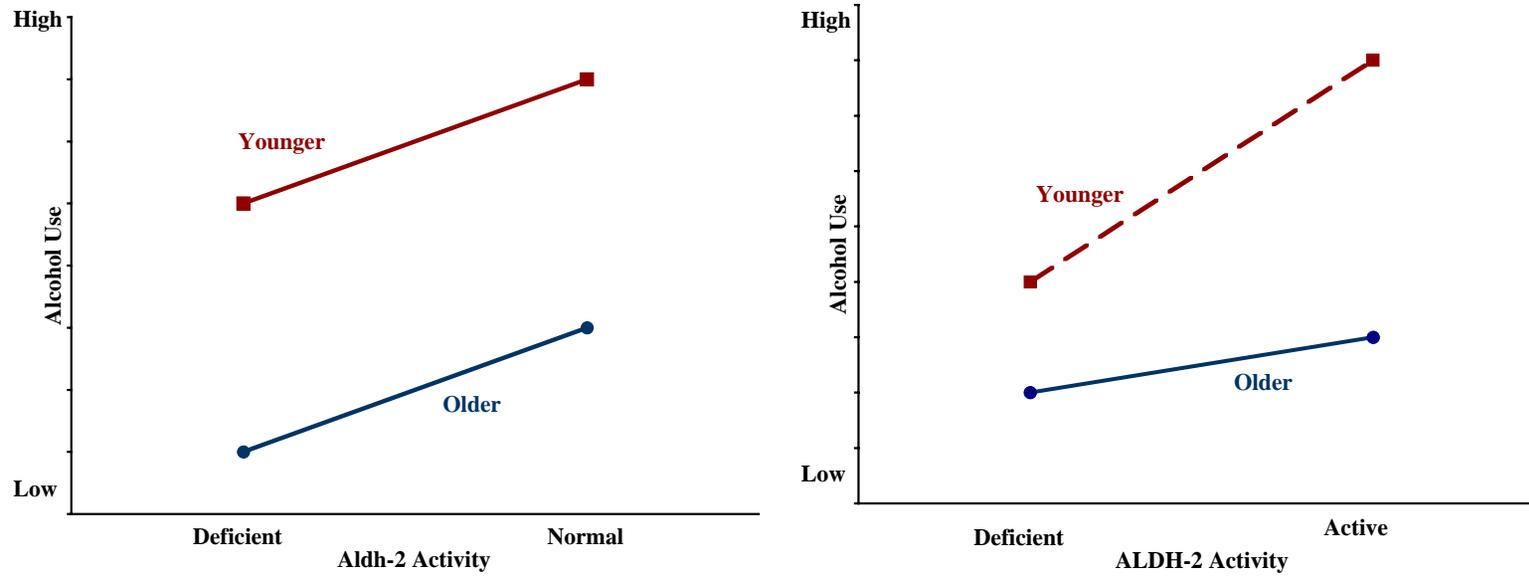
Figure 18.2. Illustration of gene-environment interaction. Both the left and right panels illustrate gene-environment interaction in the loose sense. Only the right panel illustrates gene-environment interaction in the statistical sense.