THE PROBLEM OF GENOTYPE-ENVIRONMENT CORRELATION IN THE ESTIMATION OF HERITABILITY FROM MONOZYGOTIC AND DIZYGOTIC TWINS

ARTHUR R. JENSEN
Institute of Human Learning, University of California, Berkeley, California, USA

It is commonly, but incorrectly, assumed that the presence of genotype × environment covariance must necessarily reduce the heritability (h^2) as estimated from twin data, when the formula used to obtain h^2 makes no assumption about $G \times E$ covariance or assumes that it is zero. But, in fact, $G \times E$ covariance does not always reduce the genetic variance, and it can be shown under some conditions, an increase in the $G \times E$ covariance implies a greater genetic variance. The effect of $G \times E$ covariance on h^2 as estimated from data on MZ and DZ twins, depends jointly upon the degree of assortative mating and the degree of environmental correlation between MZ twins and between DZ twins. A method, based on the solution of a pair of simultaneous quadratic equations, is proposed for estimating the range of h^2 from twin data under varying assumed values for assortative mating, the environmental correlations between MZ and DZ twins, and the $G \times E$ covariance. The solution of three simultaneous equations permits direct estimation of the genetic variance, environmental variance, and $G \times E$ covariance, under varying reasonable assumed values for assortative mating and the MZ and DZ environmental correlations. Examples of the method are based on intelligence tests scores of MZ and DZ twins.

One of the most common methods for estimating the broad heritability, h^2, of metric characters is by comparing the degree of resemblance of MZ twins reared together with the degree of resemblance of DZ twins reared together. Several different "formulas" or indexes of "heritability" have been proposed for use with twin data. But each one exhibits more or less serious shortcomings.

In order to appreciate these shortcomings, it is first necessary to present the basic genetic model from which the definition of heritability is derived. (In explicating the model, all values are expressed as deviations from the population mean.) According to the linear or additive model, an individual's phenotypic value P (i.e., deviation from the population mean) is analyzable into three additive components: a genetic deviation G, plus an environmental deviation E, plus measurement error e. Thus,

$$P = G + E + e\,. \tag{1}$$

Note that the model in this basic form does not include a component attributable to any interaction (i.e., nonadditive effect) of G and E. If in fact there were a substantial interaction of G and E, the strictly additive model would show a poor fit to empirical results. Thus, the adequacy of the additive model is an empirical question which cannot be argued on a priori theoretical grounds. One begins with the simplest possible model and tests its fit to the data.

The measurement errors, e, in Equation 1 are assumed to be random, with a mean of zero. The phenotypic variance of the population σ^2_P is expressed in the usual way:

$$\sigma^2_P = \frac{\Sigma P^2}{N} = \frac{\Sigma(G + E + e)^2}{N}\,, \text{ which when expanded is:}$$

$$\sigma^2_P = \frac{1}{N}(\Sigma G^2 + \Sigma E^2 + 2\,\Sigma GE + 2\,\Sigma Ge + 2\,\Sigma Ee + \Sigma e^2) \tag{3}$$

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or, dividing the expression in parentheses by N,

$$\sigma^2_P = \sigma^2_G + \sigma^2_E + 2\rho_{GE}\,\sigma_G\,\sigma_E + 2\rho_{Ge}\,\sigma_G\,\sigma_e + 2\rho_{Ee}\,\sigma_E\,\sigma_e + \sigma^2_e \tag{4}$$

where ρ is the correlation between G and E, G and e, etc.
Since errors are random and therefore have zero correlation with G and E, the terms including ρ_{Ge} and ρ_{Ee} must be zero and so can be deleted, thus leaving the familiar expression for the phenotypic variance:

$$\sigma^2_P = \sigma^2_G + \sigma^2_E + 2\rho_{GE}\,\sigma_G\,\sigma_E + \sigma^2_e. \tag{5}$$

In this model, the genetic variance σ^2_G is analyzable into two components: additive and nonadditive (dominance) deviations, designated σ^2_A and σ^2_D, respectively. Thus,

$$\sigma^2_G = \sigma^2_A + \sigma^2_D\,. \tag{6}$$

This formulation follows directly from simple Mendelian principles. An individual's genetic value (i.e., genetic deviation) on a given character is represented as the sum of the additive and dominance deviations, i.e., $G = A + D$. For example, consider a polygenic trait with any combination of two possible alleles, A and a, at each of three loci. The phenotypic expression of the trait is enhanced by A and diminished by a. We assign the arbitrary values A = + 1/2 and a = — 1/2. Individuals' genotypic values will differ depending on whether there is dominance. If there is complete dominance of A, then the genetic value of Aa = AA. The genetic value of an individual with the genotype of say, AA, Aa, aa, with and without dominance, is shown in Table 1.

Table 1. *Genetic values of genotype* AA, Aa, aa *with and without dominance*

	Genes			Genetic value	
	AA	Aa	aa	Additive deviation (A)	Dominance deviation (D)
Without dominance	½ + ½ = 1	½ + (— ½) = 0	— ½ + (— ½) = — 1	0	0
With dominance	½ + ½ = 1	½ + ½ = 1	— ½ + (— ½) = — 1	0	+ 1

Without dominance the genetic value of this genotype is $G = A + D = 0 + 0 = 0$. With dominance, $G = 0 + 1 = 1$. For N genotypes the variances of A and D are $\sigma^2_A = \dfrac{\Sigma A^2}{N}$ and $\sigma^2_D = \dfrac{\Sigma D^2}{N}$.

BROAD AND NARROW HERITABILITY

Heritability is defined in terms of this model as the proportion of the phenotypic variance σ^2_P attributable to genetic variance σ^2_G. But since the genetic variance can be partitioned into additive and dominance components, there are two different definitions of heritability: broad heritability h^2_b, which includes all genetic components, and narrow heritability $h_n{}^2$, which includes only the additive variance. Thus,

$$h^2_b = \sigma^2_G \,/\, \sigma^2_P, \tag{7}$$

and

$$h^2{}_n = \sigma^2{}_A / \sigma^2{}_P. \tag{8}$$

When there is no dominance, $h^2{}_b = h^2{}_n$, and with dominance $h^2{}_b > h^2{}_n$. Many geneticists refer to the "narrow heritability" simply as "heritability" and use the term "coefficient of genetic determination" for the broad heritability.

Animal and plant breeders are more interested in $h^2{}_n$, since it is more highly predictive of parent-offspring correlation and hence of responsiveness to selection. Behavioral scientists, especially those concerned with the sources of variation in human behavioral trais, are generally interested more in the broad heritability, i.e., the sum total of all the variance attributable to genetic factors. Evolutionists are more interested in the relative proportions of dominance to additive genetic variance, i.e., $\sigma^2{}_D/\sigma^2{}_A$, since this is an index of the extent to which a trait has been subject to selection. Selection, which always acts on phenotypes, will have its greatest effect on the component of genetic variance which is the most responsible for resemblance between parents and offspring, and this is the additive component $\sigma^2{}_A$. Since directional or stabilizing selection (i.e., eliminating either or both of the extremes of the distribution of phenotypes from the breeding population) reduces $\sigma^2{}_A$, more or less rapidly depending upon the severity of the selection, it increases the ratio $\sigma^2{}_D/\sigma^2{}_A$. Thus, it is sometimes said that selection "uses up" the additive variance in a breeding population.

HERITABILITY FROM TWIN DATA

Monozygotic Twins Reared Apart. Conceptually the simplest method of estimating $h^2{}_b$ is from MZ twins reared apart in uncorrelated envirenments. This condition is difficult to approach in the case of human twins. Aside from the fact that MZ twins reared apart are rare, it is seldom possible to assume that their environments are uncorrelated. They have shared the same maternal prenatal environment and when separated are often placed in different homes within the same social class or even in different branches of the same family. There is also the even more fundamental question of whether MZ twins are typical of the general population of singletons to whom we wish to generalize our estimate of $h^2{}_b$. If for the trait in question the means and variances of MZ twins are not significantly different from the corresponding population values determined on singletons, it is presumptive evidence that inferences from the twin data may be generalized to the general population of non-twins. This presumption is strengthened, of course, if other methods of heritability estimation not based on twins yield highly similar results. Despite the possible empirical difficulties with inferences based on MZ twins reared apart, it is worth noting why, at least in theory, the correlation r_{MZA} between MZ twins reared apart is an estimate of the broad heritability $h^2{}_b$.

First, we must make the reasonable (and testable) assumption that $\sigma^2{}_P = \sigma^2{}_{P'}$, where P and P' are the phenotypic values of the cotwins. ($\sigma^2{}_P$ must be assumed to be equal to the population variance if the inferences are to be generalized to the non-twin population.) Also, in the case of MZ twins the genetic values of the cotwins are identical, i.e., $G = G'$, and therefore the genetic correlation $\rho_{GG'} = 1$. It is also assumed that the twins' environmental deviations E and E' are uncorrelated, and that $\sigma^2{}_E = \sigma^2{}_{E'}$. Also, $\sigma^2{}_E$ must be assumed to equal the population environmental variance if the result is to be generalizable. The total phenotypic variance therefore is as shown in Equation 5. The covariance between N twin pairs is

$$\sigma^2{}_{PP'} = \Sigma(PP')/N \tag{9}$$

and

$$\sigma^2{}_{PP'} = \Sigma\,[(G + E)\,(G + E')]/N, \tag{10}$$

which when expanded is:

$$\sigma^2_{PP'} = \frac{1}{N}(\Sigma G^2 + \Sigma GE' + \Sigma GE + \Sigma EE'), \tag{11}$$

and dividing the expressions in parentheses by N:

$$\sigma^2_{PP'} = \rho_{GG'}\sigma^2_G + \rho_{GE'}\sigma_G\sigma_{E'} + \rho_{GE}\sigma_G\sigma_E + \rho_{EE'}\sigma_E\sigma_{E'}. \tag{12}$$

Since we have assumed that the cotwins were reared in random uncorrelated environments, the correlations ρ_{GE}, $\rho_{GE'}$ and $\rho_{EE'}$ are all zero, and so the last three terms of Eq. 12 drop out, leaving only:

$$\sigma^2_{PP'} = \rho_{GG'}\sigma^2_G \tag{13}$$

and since for MZ twins $\rho_{GG'} = 1$,

$$\sigma^2_{PP'} = \sigma^2_G, \tag{14}$$

which is to say that the MZ twin covariance is equal to the genetic variance. Dividing Eq. 14 by the total variance, we have:

$$\sigma^2_{PP'}/\sigma^2_P = r_{MZA}, \text{ and } \sigma^2_G/\sigma^2_P = h^2_b, \text{ and thus } r_{MZA} = h^2_b. \tag{15}$$

To the extent that all of the above assumptions are not met, of course, r_{MZA} will be a poor estimate of h^2_b. As already noted, the assumption least likely to be met in natural twin data is that $\rho_{EE'} = 0$, and when $\rho_{EE'} > 0$, r_{MZA} will include some environmental variance and will therefore overestimate h^2_b. This is always assumed to be true for MZ twins reared together. Since MZ twins reared together (MZT) are naturally much more plentiful than MZ twins reared apart, they are used more often in determining heritability. But since, as we have seen, MZT share some environmental variance in common, they can be used to estimate heritability only in combination with some other kinship, most simply (and most often) DZ twins reared together.

Heritability from MZ and DZ Twins. Several different indices of heritability have been used in twin research based on the comparison of MZ and DZ twins. All of them are commonly interpreted as broad heritability. The oldest and most frequently used formula is the H index of Holzinger (1929).

$$H = \frac{r_{MZ} - r_{DZ}}{1 - r_{DZ}}. \tag{16}$$

Another formula is the HR index of Nichols (1965), which was proposed as an improvement of the H index:

$$HR = \frac{2(r_{MZ} - r_{DZ})}{r_{MZ}}. \tag{17}$$

A simple formula well know to geneticists, but used surprisingly little in twin research, is

$$h^2 = 2(r_{MZ} - r_{DZ}). \tag{18}$$

An elaboration of Eq. 18 (which unrealistically assumes random mating by parents of the DZ twins) to take account of assortative mating was proposed by Jensen (1967):

$$h^2 = \frac{r_{MZ} - r_{DZ}}{\rho_{GMZ} - \rho_{GDZ}} \tag{19}$$

where ρ_G is the theoretical genetic correlation (for MZ and DZ twins, respectively), which is always unity for MZ twins and assumes a range of values greater than 0.50 for DZ twins as a function of the degree of positive assortation mating.

The differences between these four heritability indices and their shortcomings as estimates of h^2 as defined by the model can best be seen when the formulas are expressed in the algebraically equivalent form of variance components. Note that all of the formulas contain $r_{MZ} - r_{DZ}$ in the numerator. In terms of variance components, this is

$$r_{MZ} = [1\sigma^2_G + \rho_{EE'}\,\sigma^2_E + 2\,\rho_{GE}\,\sigma_G\,\sigma_E]/\sigma^2_P \tag{20}$$

$$r_{DZ} = [\rho_{GDZ}\,\sigma^2_G + \rho_{EE'}\,\sigma^2_E + 2\,\rho_{GE}\,\sigma_G\,\sigma_E]/\sigma^2_P \tag{21}$$

$$r_{MZ} - r_{DZ} = [(1 - \rho_{GDZ})\,\sigma^2_G]/\sigma^2_P \tag{22}$$

If in Eq. 20 and 21 we assume equal environmental correlations for MZ and DZ twins (i.e., $\rho_{EE'} = \rho'_{EE'}$), and equal MZ and DZ genotype-environment correlations, ρ_{GE}, then

$$r_{MZ} - r_{DZ} = [(1 - \rho_{GDZ})\,\sigma^2_G]/\sigma^2_P. \tag{23}$$

If the theoretical genetic correlation between DZ twins is 1/2, as would be the case under random mating, and if there were no dominance, then $r_{MZ} - r_{DZ}$ would be equal to $1/2\ \sigma^2_G/\sigma^2_P$, and so twice this amount would be h^2. Thus we see that Eq. 20 estimates h^2 only under these restrictive assumptions, viz.:

(*a*) equality of environmental correlations for MZ and DZ twins;
(*b*) equality of genotype×environment correlations for MZ and DZ twins;
(*c*) no dominance; and
(*d*) random mating.

If there is positive assortative mating, $\rho_{GDZ} > 1/2$, and therefore $2(r_{MZ} - r_{DZ})$ underestimates h^2. Eq. 19 differs from Eq. 18 by taking account of assortative mating. The denominator of Eq. 19 is the same as $1 - \rho_{GDZ}$, and if $\rho_{GDZ} > 1/2$, it is equivalent to $k(r_{MZ} - r_{DZ})$, where $k > 2$. Thus, all of the formulas except Eq. 19 underestimate h^2 when there is assortative mating. But Eq. 19 is no better than the rest with respect to the above assumptions *a*, *b* and *c*.

Holzinger's *H* and Nichols's *HR*, as can be seen by expressing their denominators as variance components, are in no way algebraically equivalent to $h^2 = \sigma^2_G/\sigma^2_P$. The Nichols formula (Eq. 17), under assumptions *a* to *d* assuming $\rho_{GDZ} = 1/2$ is:

$$HR = \frac{\sigma^2_G}{\sigma^2_G + \rho_{EE'}\,\sigma^2_E + 2\,\rho_{GE}\,\sigma_G\,\sigma_E}. \tag{24}$$

Since the denominator is simply the covariance for the MZ twins, it is lacking two variance components that are contained in the total variance which is the denominator in the definitional formula for h^2, viz., $(1 - \rho_{EE'})\,\sigma^2_E$ and σ^2_{error}. *HR*, with its deficient denominator, therefore overestimates h^2. It should be abandoned as an estimate of h^2. Also, it should be noted that the absence of the error variance in the denominator of Eq. 24 means that *HR* is, in effect, corrected for attenuation (i.e., unreliability of measurement).

Holzinger's *H*, under assumptions *a* to *d*, is algebraically equivalent to:

$$H = \frac{1/2\ \sigma^2_G}{1/2\ \sigma^2_G + (1 - \rho_{EE'})\,\sigma^2_E + \sigma^2_{error}}. \tag{25}$$

Thus *H* is practically uninterpretable, even under the restrictive assumptions; it is the ratio of half the genetic variance to half the genetic variance plus some indeterminate fraction of the environmental variance plus error variance. It is an utterly confused formulation and should never be used as an index of heritability.

Thus we see that only Eq. 20 and 21 can be regarded as estimates of h^2 under assumptions *a* to *d*, and Eq. 21 does not depend upon assumption *d* if there is information on assortative mating, although the effect of assortative mating on the estimate of the DZ genetic correlation ρ_{GDZ} will depend upon the particular genetic model of assortative mating that one uses to obtain ρ_{GDZ}.

To the extent that assumptions *a* to *d* are not met, the MZ-DZ twin method cannot provide an estimate, and certainly not a point estimate, of h^2. So we must now examine each of these assumptions to determine how they might be taken into account to obtain the most defensible heritability information from twin data.

(a) Environmental Correlations for MZ and DZ Twins

In estimating the heritability of behavioral characteristics such as mental abilities and personality traits, which are substantially influenced by environmental factors, the similarity of twins' environments becomes a question of crucial importance. If MZ pairs are subjected to more similar environmental influences relevant to the trait in question than are DZ pairs, any method of heritability estimation based on the difference $r_{MZ} - r_{DZ}$ will spuriously inflate h^2 with environmental variance. Unfortunately, the fact is that we do not know if there is a difference in the environmental correlations of MZ and DZ twins; or, if there is a difference, how large it is.

It seems clear that MZ twins share more similar social environments than do DZ twins (Smith 1965). MZ twins are more often mistaken for one another by relatives and friends, they are more often dressed alike, spend more time with each other, experience a closer relationship, engage in more similar activities, have more similar diets, likes and dislikes, and so on. This apparently is true even when parents are not aware of the genetic distinction between MZ and DZ twins or are mistaken as to which type of twins they have.

It is often argued, on the other hand, that MZ twins have less similar prenatal environments than DZ twins, and that the prenatal differences can affect later mental as well as physical characteristics (Price 1950). Intrapair MZ twin differences in height and other skeletal measurements have been found to be correlated with the intrapair IQ differences (Burks 1940). The largest MZ twin difference in IQ (24 IQ points) reported in the literature (the case of Gladys and Helen in Newman et al. 1937), also ahowed the largest difference in fingerprints, a difference which must have arisen before the fourth month after conception, since fingerprints are completely formed by that point in fetal development (see Jensen 1972).

MZ twins often share the same placenta, chorion, and amnion, and mutual circulation typicallr results in an imbalance in the twins' circulation which can cause differences in fetal growth, in birtr weights, and in stillbirths. Some of these effects of severe prenatal competition, such as intrapaiy differences in birth weight, have been found to be correlated with later IQ differences, the heavieh twin usually having a slightly higher IQ (Babson et al. 1964, Scarr 1969, Willerman and Churchill 1967). Breland (1974), however, has argued, on the basis of her review of the literature and new evidence, that these prenatal effects have negligible influence on intrapair twin differences in intelligence in representative samples where twins have not been selected for extreme differences in birth weight or intelligence. Breland found no significant difference in the intrapair IQ differences of large samples of MZ twins who were concordant and MZ twins who were discordant for handedness. MZ twins who split late in their development are the most prone to severe fetal competition due to mutual circulation, etc., and are also more likely to be mirror images of one another and to be discordant for handedness. Breland concluded: "While discordance or concordance for handedness is a crude estimator of the time of splitting of one fertilized egg into two identical twins, it is one of the few indicators readily observable after birth. Since there were no significant differences between MZ sets concordant for handedness as compared to MZ twins discordant for handedness, it can be concluded that heritability estimates of ability based on the comparison of MZ to DZ twin sets selected from school populations are not appreciably biased by the differential prenatal environments of the identical twin sets" (p. 108).

The prenatal environments of MZ and DZ twins are the same in several respects. The two twins pass through fetal development in the same uterus, at the same time, so that maternal age, health, and parity are constant. And the two individuals usually have the same neonatal care. It is not until the social environment becomes an important influence in development that the postnatal environmental correlation for MZ twins undoubtedly is greater than for DZ pairs. Whether this MZ-DZ difference in postnatal environmental correlations is balanced by prenatal differences going in the opposite direction, we cannot say for sure, though it seems very unlikely that the differences in pre- and postnatal environmental correlations for MZ and DZ twins would perfectly cancel each other out. And so we are left with some uncertainty about the MZ-DZ difference in the net environmental correlation, and no presently available evidence permits us to establish this difference. Under these conditions, the only reasonable solution is to recognize the uncertainty on this point and to take it into account in deriving estimates of heritability from twin data. This can be done by hypothesizing a likely range of values for the environmental correlations and determining the effect of this variation on our estimates of h^2.

(b) Correlation of Genotypes and Environments

It has been claimed, incorrectly as we shall see, that " a necessary and sufficient condition for the applicability of heritability analysis is the absence of genotype-environment correlation. This condition is rarely, if ever, met for behavioral traits in human populations " (Layzer 1974, p. 1259). Although it is undoubtedly true that the absence of genotype-environment correlation is rare in natural populations, the fact of $G \times E$ correlation certainly does not preclude the estimation of h^2 from twin data. However, unless there has been rigid experimental control explicitly designed to minimize $G \times E$ correlation, this source of variance must properly be takes into account in estimating heritability. Another more common misconception is that the larger the $G \times E$ correlation, ρ_{GE}, the smaller must be h^2, as one might intuitively expect from thinking of the GE covariance as subtracting from the genetic variance. It will be shown that under certain conditions just the opposite is true, and that a larger $G \times E$ correlation depends upon a larger proportion of genetic variance and consequently implies higher heritability. Also, since the covariance of genotypes and environments is equal to $2\,\rho_{GE}\,\sigma_G\,\sigma_E$, the maximum size of the covariance, for any value of ρ_{GE}, is attained when $\sigma_G = \sigma_E$, and under this condition it can be shown that $\sigma^2_G + \sigma^2_E$ is equal to the covariance. If $\sigma_G \neq \sigma_E$, the covariance must be less than $\sigma^2_G + \sigma^2_E$. (This follows from $(G + E)^2 = G^2 + 2GE + E^2$, and if $G = E$, then $G^2 + E^2 = 2GE$.) Therefore, the $G \times E$ covariance can never overwhelm the additive components of genetic and environmental variance.

In quantitative genetics the environmental variance is defined as that part of the phenotypic variance (not including error variance) which is *independent* of the genotype. The correlation between genetic and environmental factors can be divided theoretically between that part of the correlation which is attributable to *selection* of environmental factors by the genotype and that part which is attributable to *imposed* environmental factors. In many natural environments, some features of the environment that become important to an individual gain their salience because of the individual's genetically determined propensities. An intellectually gifted child, for example, is apt to spend more time engaged in intellectually stimulating activities than the average child. MZ twins reared apart in random environments would share in common their genetic variance plus only that part of the $G \times E$ covariance which is dependent on or selected by the genotype.

Most geneticists maintain that this genotypically selected part of the $G \times E$ covariance should be regarded as a part of the genetic variance, and hence of the broad heritability, since it is an integral part of the mechanism through which the genotype becomes expressed in the phenotype. Roberts (1967), p. 218) has stated the point very clearly: " The genotype may influence the phenotype either by means of biochemical or other processes, labeled for convenience as 'development,' or by means of influencing the animal's choice of environment. But this second pathway, just as much as the first, is a genetic one; formally it matters not one whit whether the effects of the genes are mediated

through the external environment or directly through say, the ribosomes." Indeed, some aspects of $G \times E$ correlation would be practically impossible to eliminate except through the most rigid experimental control, and in the case of mental activities even this is doubtful. For example, musically talented persons tend to think about music and run through scores in their mind's ear, even while engaged in other activities. The same has been said about mathematicians, chess players, and writers. Thus, genotypic differences are magnified in the phenotypes through genotypically conditioned differential selection of environmental factors. In natural environments this could contribute the largest share of the $G \times E$ covariance in mental abilities. MZ twins will naturally share more of this source of variance in common than DZ twins. And in any heritability estimate based on $r_{MZ} - r_{DZ}$, this genotypically selected part of the $G \times E$ covariance will be included in the genetic variance, as it should be. The difference between the covariances of MZ twins reared together (MZT) and reared apart (MZA) in uncorrelated environments can be seen in these terms as follows:

$$r_{MZT}\,\sigma^2_P = \sigma^2_G + \rho_{EE'}\,\sigma^2_E + 2\,\rho_{GIE}\,\sigma_G\,\sigma_{IE} + 2\,\rho_{GSE}\,\sigma_G\,\sigma_{SE} \tag{26}$$

$$r_{MZA}\,\sigma^2_P = \sigma^2_G + \ldots\ldots + \ldots\ldots\ldots\ldots + 2\,\rho_{GSE}\,\sigma_G\,\sigma_{SE} \tag{27}$$

$$\text{Difference} = \rho_{EE'}\,\sigma^2_E + 2\,\rho_{GIE}\,\sigma_G\,\sigma_{IE}$$

(where the subscription *IE* signifies "imposed environment" and *SE* signifies "selected environment.")

Since the *MZA* $\rho_{EE'}$ and ρ_{GIE} are both assumed to be zero, the second and third expressions in Eq. 27 drop out. As previously noted, the last term in the above equation, i.e., the $G \times E$ covariance involving genotypically determined self-selection of environmental factors, should be included with σ^2_G; it is shown here separately only for expository purposes.

The imposed part of the $G \times E$ correlation is attributable to those features of the environment which the individual himself does not control and which are, so to speak, imposed on the individual regardless of his genotype. Examples are the quality of the child's home, the socioeconomic status, intelligence and educational level of the child's parents, and the like. These are the features of the family environment which are common to all the siblings within the family. The $G \times E$ covariance resulting from this aspect of the environment theoretically should be the same for MZ as for DZ twins (or other siblings), and should not be confounded with the genetic variance. The twin method of estimating heritability actually offers the possibility of separately estimating also the $G \times E$ correlation, as we shall see.

Finally, it should be noted that some $G \times E$ correlations can be negative. For example, scholastically less able children often receive more than the usual amount of help and encouragement from parents and tutors. Studies have shown substantial negative correlations between children's IQs and the amount of time spent doing homework and amount of parental tutoring, especially in the early school years. Thus scores on tests which reflect scholastic knowledge and skills might show a negative $G \times E$ covariance component. Negative $G \times E$ covariance of course reduces the total phenotypic variance in the population. In this connection it is interesting that culturally and scholastically loaded mental tests usually show smaller variance than nonscholastic culture-fair types of tests.

(c) *Dominance*

Since MZ twins have identical genotypes, the MZ covariance of course includes all of the genetic variance attributable to additive and dominance deviations. The genetic covariance for MZ twins therefore is equal to the total genetic variance, i.e.,

$$\rho_{GG'}\,\sigma^2_G = \rho_{AA'}\,\sigma^2_A + \rho_{DD'}\,\sigma^2_D = \sigma^2_A + \sigma^2_D = \sigma^2_G \tag{28}$$

where $\rho_{GG'}$ is the total genetic correlation between MZ twins, $\rho_{AA'}$ is the correlation between additive deviations, and $\rho_{DD'}$ is the correlation between dominance deviation. For MZ twins these correlations obviously must all be unity.

The situation is more complex for DZ twins, who are genetically equivalent to full siblings. Consider a single gene locus with alleles **A** and **a** having relative frequencies p and q in the population (where $p + q = 1$). The average relative frequencies of individuals having each of the possible genotypes resulting from the mating in the parental generation are then given by $(p\mathbf{A} + q\mathbf{a})^2 = p^2\mathbf{AA} + 2pq\mathbf{Aa} + q^2\mathbf{aa}$. The genetic correlation between any two persons is defined as the probability at any one gene locus of their sharing the same genetic effect *by descent from a common ancestor.* (For polygenic traits, the genetic correlation is the average probability over all loci contributing to the trait's variance.) (The derivation of this definition of genetic correlation from genetical theory is fully explicated by Crow and Kimura 1970, pp. 130-140.) Additive deviations contributing to the genetic variance (i.e., the additive variance) are attributable to *single* alleles **A** and **a**, each allele adding its own effect to any combination, while dominance deviations depend upon a *pair* of alleles, **Aa**, where the combination is more (or less) than the additive effects of **A** + **a**.

The additive genetic correlation between two siblings, then, is the probability that they will have one allele in common from the same parent. Since there are two alleles at the parental locus, the probability that both siblings have each received the same allele from the same parent is 1/2. So the additive genetic correlation, $\rho'_{AA'}$, between siblings is 1/2. Since dominance depends upon the interaction of two alleles, the probability that two siblings have received the same first allele from the father is 1/2 and the probability that they have both received the same second allele from the mother is also 1/2, so that the joint probability of the two siblings sharing both alleles by descent is $1/2 \times 1/2 = 1/4$. And thus the sibling correlation between dominance effects, $\rho'_{DD'}$, is 1/4.

The sibling genetic covariance therefore is

$$\rho'_{GG'}\sigma^2_G = \rho'_{AA'}\sigma^2_A + \rho'_{DD'}\sigma^2_D = 1/2\,\sigma^2_A + 1/4\,\sigma^2_D \tag{30}$$

If now we subtract the genetic covariance of DZ twins from the genetic covariance of MZ twins, we have,

$$\begin{aligned}
\text{MZ: } \rho_{GG'}\sigma^2_G &= \sigma^2_A + \sigma^2_D \\
\text{DZ: } \rho'_{GG'}\sigma^2_G &= 1/2\,\sigma^2_A + 1/4\,\sigma^2_D \\
\text{Difference: } &= 1/2\,\sigma^2_A + 3/4\,\sigma^2_D
\end{aligned}$$

So we see that heritability formulas based on $r_{MZ} - r_{DZ}$ will inflate the total genetic variance to the extent thet dominance deviations contribute to the trait's variance, since with dominance the theoretical genetic correlation between DZ twins will be something less than 1/2. If there were complete dominance of **A** and the relative frequencies of **A** and **a** were each 1/2, for example, the genetic sibling (or DZ twin) correlation would be 5/12 (or 0.417). Since complete dominance at all loci for polygenic traits is unlikely, the actual genetic correlation for DZ twins most probably lies between 0.42 and 0.50, assuming random mating. Nevertheless, the possible presence of dominance adds some uncertainty to the estimates of h^2 based on MZ-DZ twin comparisons. The only solution to this ambiguity is to give up thinking of h^2 as a point estimate and regard it as a probable range of values which may be estimated by assuming a range of reasonable values for the DZ genetic correlation, the limits of which would be 0.42 and 0.50 (under the assumptions that $p = q$ and that mating is random). If $p \neq q$ and q is the relative frequency of the recessive allele, the sibling (or DZ) genetic correlation is $(1 + 3q)/(4 + 4q)$. Since there is considerable positive assortative mating for socially salient traits such as intelligence, this effect must also be taken into consideration.

(d) *Assortative Mating*

Assortative mating (i.e., positive correlation between the parental genotypes) for a given trait has several genetic effects:

1. It does not change gene frequencies in the population, but it changes the frequencies of genotypes, i.e., combinations of genes; it increases the frequency of those gentoypes which make for more extreme values in the trait and decreases the frequency of genotypes which make for more average values.
2. Since it does not change gene frequencies, it has no effect on the population mean when there is no dominance, and it increases the population mean when there is directional dominance for higher values.
3. It increases the additive genetic variance.
4. It increases the variance between family means.
5. It has an almost negligible effect on the variance within families.
6. It increases parent-child correlation.
7. It increases sibling (and DZ twin) correlation.

Because of No. 7, it is therefore necessary to consider assortative mating in estimating $\rho_{GG'}$, the genetic correlation between DZ twins.

After one generation of assortative mating (i.e., assuming previous generations have mated at random), the sibling additive genetic correlation is:

$$\rho_{AA'} = \frac{1 + r_g}{2 + r_g} \tag{31}$$

where r_g is the correlation between the mates' breeding values. (The breeding value of a genotype is its additive genetic effect, i.e., the sum of the independent values of each of the alleles. r_g is estimated by $r_{pp'}\, h^2_n$, i.e., the product of the phenotypic correlation between mates and the narrow heritability.

With each generation of assortative mating, the additive genetic variance is increased at a negatively accelerated rate from generation to generation and finally becomes stabilized at some equilibrium level. Most human traits which have been subject to assortative mating for several generations, like height and intelligence, are probably close to equilibrium. (The equilibrium level will of course be raised if there is an increase in the degree of assortative mating.) Assuming a constant degree of assortative mating over n generations, the additive genetic sibling correlation at equilibrium is

$$\rho'_{AA'} = \frac{\frac{1}{2} + \frac{r_g}{1 - r_g}}{1 + \frac{r_g}{1 - r_g}}, \quad \lim_{n \to \infty} \rho'_{AA'} = \frac{1}{2}(1 + r_g). \tag{32}$$

Thus some reasonable range of values between the limits of $\rho_{AA'}$ and $\rho'_{AA'}$ should be entered into the $\rho'_{AA'}$ of Eq. 30 for traits in which it is likely that there is assortative mating. For intelligence, most coefficients of assortative mating (i.e., parental phenotypic correlations) that have been reported are in the range from about 0.40 to 0.60. Estimates of h_n^2 are mostly between 0.50 and 0.70, so that the most likely range for r_g (i.e., the correlation between mates' breeding values) would be about 0.20 to 0.40.

For polygenic traits, especially if the number of loci is large, it is safe to assume that assortative mating has a negligible effect on the dominance variance. Actually, assortative mating very slightly decreases σ^2_D (see Crow and Kimura 1970, p. 156).

So, what we are left with by this analysis is the necessity for estimating the theoretic genetic correlation $\rho'_{GG'}$ between DZ twins, taking into account the opposite effects of dominance and assortative mating. A range of likely values for $\rho_{GG'}$ is safest. But one should be able to make an informed guess

as to the most probable central tendency of this range in the case of IQ, based on independent estimates of the proportion of dominance variance and of assortative mating. Assuming equal frequencies for A and a, even with complete dominance the additive genetic variance is twice as great as the dominance variance, and under this condition (now assuming random mating) the DZ genetic correlation would be $\rho'_{GG'} = (1/2)(2/3) + (1/4)(1/3) = 5/12 = 0.42$, since $\sigma^2_A + \sigma^2_D =$ unity (i.e., the total genetic variance) and the additive and dominance variance are in the ratio of 2 to 1. So the limits of $\rho'_{GG'}$, from no dominance to complete dominance, are 0.50 to 0.42. Under assortative mating at equilibrium (which affects only the additive genetic variance), assuming, say, $r_g = 0.30$, the limits of $\rho'_{GG'}$, from no dominance to complete dominance, would be 0.65 to 0.52. Since Jinks and Fulker (1970, p. 343) have concluded on the basis of the available evidence that there is a high level of dominance for IQ, $\rho'_{GG'}$ is probably closer to 0.52 than to 0.65. The highest likely degree of assortative mating for IQ (i.e., $r_g = 0.40$), under complete dominance would result in $\rho_{GG'} = 0.55$.

INTERACTION BETWEEN ENVIRONMENTAL CORRELATION, G × E COVARIANCE, AND ASSORTATIVE MATING IN DETERMING h^2

By entering a range of hypothetical values into the unknown parameters of the equations for the covariance of MZ twins and DZ twins and solving the simultaneous equations for the genetic and environmental variances, we can gain an insight into how different environmental correlations for MZ and DZ twins, $G \times E$ covariance, and assortative mating jointly affect the heritability. (The method for estimating variance components from twin data by solving simultaneous equations was originally proposed by Robin Hogarth (1974), as was also the form of the graphical representation of the results used in Figs. 1-3.) For this demonstration I shall use empirical estimates of the MZ and DZ twin correlations, but my immediate aim is more to illustrate some theoretical points than to estimate the heritability of IQ on the basis of this particular analysis.

Erlenmeyer-Kimling and Jarvik (1963), in their well-known review of kinship correlations for intelligence, found the median of MZ twin (reared together) correlations in 14 studies to be 0.87. The median correlation in 11 studies of like-sex DZ twins is 0.56. Many of these studies unfortunately do not report the standard deviations of the mental test scores in the twin samples. If we assume that the variance of MZ and DZ twins are equal and that each is also equal to the population variance, we can estimate the covariances for MZ and DZ twins. (For a detailed discussion of the assumption that $\sigma^2_{MZ} = \sigma^2_{DZ}$ see Christian et al. 1974.) Since the population standard deviation of IQ is 15, the variance is $15^2 = 225$. The covariance for MZ twins, then, is $r_{MZ}\sigma^2_P = 0.87 \times 225 = 195.75$; and for DZ twins it is $0.56 \times 225 = 126.00$. Thus we have two equations:

$$\text{MZ Covariance: } r_{MZ}\sigma^2_P = \rho_{GG'}\sigma^2_G + \rho_{EE'}\sigma^2_E + 2\rho_{GE}\sigma_G\sigma_E = 195.75 \quad (33)$$

$$\text{DZ Covariance: } r_{DZ}\sigma^2_P = \rho'_{GG'}\sigma^2_G + \rho'_{EE'}\sigma^2_E + 2\rho_{GE}\sigma_G\sigma_E = 126.00 \quad (34)$$

In order to solve the equations for σ^2_G and σ^2_E, we must enter hypothetical values for the coefficients $\rho_{GG'}$, ρ_{EE} and ρ_{GE}.

The genetic correlation for MZ twins, $\rho_{GG'}$, is of course always 1. The genetic correlation for DZ twins can take a range of values depending upon dominance and assortative mating. For the present illustration we will assume no dominance and only one generation of assortative mating. We will set the assortative mating coefficient at three values: 0, 0.20, and 0.40. The corresponding DZ genetic correlations under these conditions (see Eq. 31) are 0.50, 0.54, and 0.58.

The environmental correlation $\rho_{EE'}$ for MZ twins is set at one value: 0.90, which is about the upper probable limit for this correlation. The environmental correlation $\rho_{EE'}$ for DZ twins ranges from 0.90 down to 0.70, and thus differs from the MZ environmental correlation over a range from 0 to 0.20.

The genotype-environment correlation ρ_{GE} is allowed to vary over a tenfold range from 0.04 to 0.40. The value of 0.40 seems a reasonable upper bound estimate of ρ_{GE}, since the highest reported multiple

correlation between a host of fine-grained environmental indices and the IQs of adopted children is 0.42 (Burks 1928). Burks's environmental ratings were based on an elaborate index which took 4 to 8 hours of home visits by the examiner to complete, and it includes also the IQs of the adopting parents. Since it seems most unlikely that children's genotypes for intelligence would be more highly correlated with their environments than are their measured IQs, 0.40 seems a good upper-bound estimate of ρ_{GE}.

With these various values entered into the equations for the MZ and DZ covariances, we can then solve the equations for the two unknowns σ^2_G and σ^2_E. (The solutions are easily accomplished by a computer routine for solving simultaneous quadratic equations.) The results of solving these equations, entering the range of values indicated, are shown in Figs. 1 to 3.

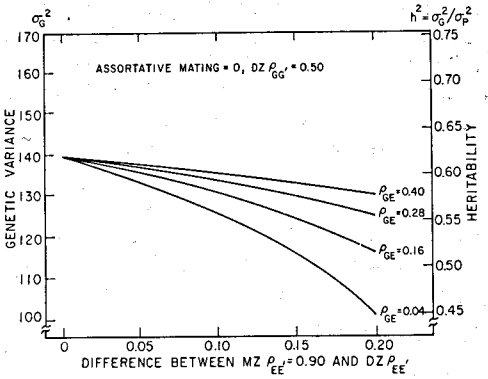

Fig. 1

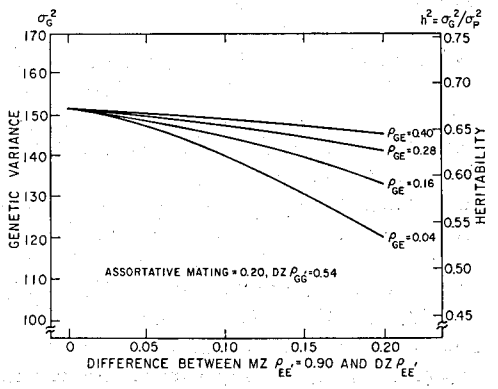

Fig. 2

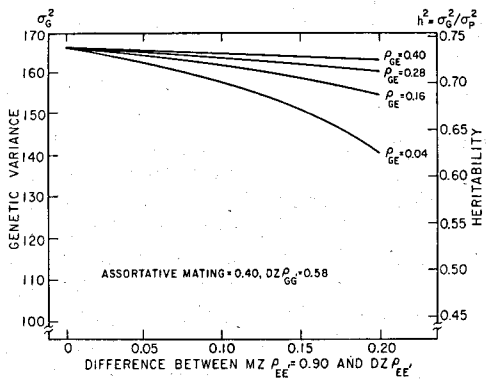

Fig. 3

Figs. 1-3. Genetic variance and broad heritability based on MZ-DZ twin comparison under different assumed values of the parameters $\rho_{EE'}$ (environmental correlation between twins), ρ_{GE} (correlation between genotypes and environments), and assortative mating.

Comparing these Figures, we see that the genetic variance increases as a function of the degree of assortative mating, under all values of the other parameters. But the effect of $G \times E$ covariance depends both upon the degree of assortative mating (having less effect the higher the assortative mating) and upon the discrepancy between the MZ and DZ environmental correlation - the greater this discrepancy, the greater is the effect of the genotype-environment correlation, ρ_{GE}. But it may seem surprising and counter-intuitive that the greater the genotype-environment correlation, the greater is the genetic variance and the higher the heritability. This illustration belies the claim that h^2 cannot be estimated if there is a genotype-environment correlation, or the notion that h^2 is inversely related to the magnitude of the genotype-environment correlation.

ESTIMATING σ^2_G, σ^2_E AND ρ_{GE} FROM TWIN DATA

The foregoing analysis is intended simply to illustrate the interrelationships among the various parameters in determining h^2. Actually we can go a step further and estimate also ρ_{GE} from twin data, by solving three simultaneous equations with three unknowns: σ^2_G, σ^2_E, and ρ_{GE}. In addition to Eq. 33 and 34, we use the equation for the total variance of IQ, omitting error variance (assumed here to be 5%) so that the estimated parameters will in effect be corrected for attenuation (i.e., unreliability of measurement).

$$\sigma^2_P = \sigma^2_G + \sigma^2_E + 2\,\rho_{GE}\,\sigma_G\,\sigma_E = 213.75\,. \tag{35}$$

Eq. 33, 34, and 35 were solved simultaneously with every possible combination of a wide range of values of the DZ genetic correlation $\rho'_{GG'}$ and of the MZ and DZ environmental correlation, $\rho_{EE'}$ and $\rho'_{EE'}$, respectively. The values assigned to $\rho'_{GG'}$ were 0.50, 0.54, 0.58, 0.60, and 0.70. The values assigned to $\rho_{EE'}$ for MZ twins were 0.90, 0.80, and 0.70. The values of $\rho'_{EE'}$ for DZ twins were 0.90, 0.80, 0.70, and 0.60. All combinations of these parameters make $5 \times 3 \times 4 = 60$ sets of simultaneous equations. Not all combinations of these parameters yield possible solutions, and some of the remaining solutions, though mathematically possible, are genetically impossible — e.g., negative values of σ^2_G and σ^2_E. If we stipulate on theoretical grounds that the correlation between genotypes and environments in the population must be non-negative, then only one of the entire 60 sets of equations yields a *realistic* solution. * The combination of hypothetical values in the solution are environmental correlations of 0.70 for both MZ and DZ twins, and a DZ twin genetic correlation of 0.50. The resulting estimates are $\sigma^2_G = 139.50$, $\sigma^2_E = 60.00$, $\rho_{GE} = +0.078$. Thus $h^2 = 0.65$, $\sigma^2_E/\sigma^2_P = 0.28$, and $\mathrm{Cov}_{GE}/\sigma^2_P = 0.07$.

Attention is directed to the DZ genetic correlation of 0.50, since with the known high degree of assortative mating for intelligence, this genetic correlation would have to be greater than 0.50 unless there were some dominance variance. This finding, therefore, is consistent with other evidence for dominance in IQ variance (e.g., inbreeding depression: Schull and Neel 1965). The dominance lowers the DZ $\rho'_{GG'}$ to about the same extent that assortative mating raises $\rho'_{GG'}$, with a resultant value of ρ'_{GG} close to 0.50. If we assume an assortative mating coefficient, r_a, of 0.30, then there must be nearly complete dominance.

The 12 other sets of equations which yielded genetically possible solutions (i.e., with σ^2_G and σ^2_E non-negative) gave negative values of ρ_{GE}, ranging from -0.03 to -0.49 and averaging -0.24. Values of h^2 for these solutions ranged from 0.25 to 1.27, with an average h^2 of 0.84. (Note that it is theoretically possible for h^2 to be greater than 1 if there is a sufficiently large negative $G \times E$ correlation.) But several of these solutions, especially those yielding the most extreme negative values of ρ_{GE}, are based on rather unlikely combinations of the theoretical parameters, such as much higher environmental correlations for DZ than for MZ twins. It seems more likely that in the total population the correlation between genotypes and environments should be positive for measurements such as IQ, and so the one solution yielding a positive value of ρ_{GE} seems to me the most reasonable outcome of this analysis. Whether ρ_{GE} is positive or negative could be answered definitively only by overdetermining the solution by using more equations based on covariances for other kinships, e.g., siblings, parent-child, half-sibs, cousins, and unrelated children reared together.

CONCLUSION

Because of the uncertainty about the environmental correlations for MZ and DZ twins and the amounts of dominance and assortative mating, which would affect the genetic correlation between DZ twins,

* A few other mathematically possible solutions emerge but can be discarded as quite unrealistic in that they are clearly contradicted by evidence from other types of kinship studies of IQ heritability, e. g., adopted children.

t is best, in estimating the variance components σ^2_G, σ^2_E, and ρ_{GE}, from MZ and DZ twin data, to ssume a range of reasonable values for the uncertain parameters and see which of these, in all possible ombinations, leads to possible solutions of simultaneous equations based on a genetic model. If number of different values of the variance components emerge in the solutions, the estimates of enetic and environmental variance and the $G \times E$ covariance must then be described as being within ome range of values. And that is probably the most that can reasonably be done with data on MZ nd DZ twins.

When this procedure was applied to the median values of the correlations for IQ reported in 14 studies of MZ and 11 studies of like-sex DZ twins, only one realistic solution (when restricted by the assumption that all the estimated variance components should be positive) emerged. It attributes 65% of the variance to genetic factors, 28% to environmental factors, and 7% to the covariance between genetic and environmental factors. The analysis also indicates equal environmental correlations for MZ and DZ twins (with respect to environmental influences on IQ) and suggests (but cannot precisely estimate) a substantial amount of dominance in the genetic determination of IQ differences.

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Prof. A.A. Jensen, Institute of Human Learning, University of California, Berkeley, California 94720, USA.