HERITABILITY OF THRESHOLD CHARACTERS*[1]

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WITH AN APPENDIX BY ALAN ROBERTSON

THE amount of selection pressure applied on the phenotypic level to metric characters can be expressed in terms of the selection differential, defined as the extent of superiority of the individuals which are the parents of the next generation (weighted for the relative number of offspring they produce) over the average of their own generation. For characters showing a discontinuous distribution the measurement of selection pressure can be expressed in terms of selection coefficients, that is, the relative number of offspring produced by each of the phenotypically classified categories of individuals in the population.

THE DEGREE OF HERITABILITY

The effective amount of selection pressure, however, in both cases depends on the degree to which the phenotypic variation, on which the selection differential and the selection coefficients are based, is reflected by genetic variation. It seems rather obvious that the changes in the genetic composition of a population from generation to generation, changes of which the shifts in the phenotypic mean will be representative (under a constant environment), are thus a function of the accuracy with which either nature or man recognizes genetic differences on the basis of phenotypic differences between individuals or groups of individuals.

Intimations of this fact were apparent to early geneticists. Many of them recognized that a phenotype represents a combination of genetic and environmental effects, of which only the first would contribute to changes in a population which are attributable to selection. As a single example the viewpoint of YULE (1906) may be cited. In discussing ancestor-offspring correlation, he stated: "A complete theory of heredity should take into account, besides germinal processes, the effect of the environment in modifying the soma obtained from any given type of germ-cell—an effect which is hardly likely to be negligible in the case of such a character as stature."

As early as 1910 WEINBERG (1909, 1910) suggested methods of separating genetic and environmental components of total phenotypic variability, but his contribution to the subject was overlooked for many years, sharing the fate

of his independent discovery of what was subsequently referred to in genetic literature as HARDY's binomial (see STERN 1943).

It was only several years later that WRIGHT (1917 *et seq.*) and FISHER (1918), independently of each other and apparently unaware of WEINBERG's papers, developed comprehensive techniques of dealing with the problem. The first of WRIGHT's papers dealt with a case in which the genetic and the environmental types of variation were separated by experimental rather than statistical means. This is, of course, essentially the procedure used by JOHANNSEN (1903), who, however, did not attempt to assess the relative proportions of the two types of variation in a population.

WRIGHT's material consisted of inbred lines of guinea pigs. His analysis included not only the measurement of the relative amounts of genetic and environmental variation, but also the separation of genetic from environmental correlation between two traits by the use of the technique of covariance (at that time as yet an uncoined term).

A subsequent contribution of WRIGHT (1918) first presented his method of path coefficients, elaborated upon by him later (1921, 1934a), a method which permits the statistical separation of environmental and genetic variation in general populations. Simultaneously and independently FISHER (1918) dealt with the same subject, which he also elaborated upon later (FISHER, IMMER and TEDIN 1932). The developments finding their origin in the respective contributions of WRIGHT and FISHER appeared to proceed to a considerable degree independently of each other. Thus the statistical separation of genetic and environmental correlations between different traits was eventually described by SMITH (1936) basing himself on FISHER's work, and by HAZEL (1943), whose technique was derived from the method of path coefficients combined with the analysis of covariance.

The fraction of the total phenotypic variance due to genetic differences came to be known as the degree of heritability or simply as heritability (LUSH 1945) usually symbolized by h^2. It is, however, necessary to distinguish between two different statistics which may be understood by this term, namely the *total* and the *additive* h^2. The distinction between them may be made clear by reference to *genotypic* and to *additive genetic* values of individuals.

The former (with respect to whatever trait is considered) is defined as the mean of an individual's genetic replicates exposed to the total array of environments. The concept of the additively genetic value is based on the theoretical possibility of assigning particular values to each gene contained in the population. These can be so chosen that the variance of the differences between their sums for each individual (the additive genetic value) and the genotypic values for the same individuals is at a minimum. If the genotypic value of every individual is the same as its additive genetic value, genetic variance is completely additive. If this is not true, the additive portion of the genetic variance is equal to the variance of the additively genetic values; the amount by which the total genotypic variance exceeds this value is the non-additive genetic variance.

The additive heritability then refers to the ratio of the additively genetic

to the total variance. In the absence of non-additive genetic variation, and when there is no correlation between genotypic level of individuals in the population and its environmental variance,—(1) the correlation between the genotypic and phenotypic values is equal to the square root of the heritability, (2) the regression of genotypic value on phenotypic value is linear, and (3) the expectation of phenotypic value of offspring is equal to the mean genotypic value of the parents (excluding deviations from this rule due to sex linkage). It follows that genetic gain from mass selection is equal to the product of the heritability and the mean phenotypic advantage of.the parents over the mean of their generation (the selection differential). A combination of these propositions with a knowledge of the genetic and non-genetic correlations between members of a family leads to simple formulas for computing gains from family selection and from combined family and individual selection on the basis of an index, as well as formulas for computing optimum indexes for this purpose and for simultaneous selection with respect to more than one character at the same time (HAZEL 1943; LUSH 1947).

When some non-additive genetic variance is present, the expectation of the phenotypic value of the offspring is not necessarily equal to the mean genotypic value of the parents. A tendency for the regression of offspring towards the mean of the parental generation is present, so that selection is less effective than it might be expected to be on the basis of the genotypic values of the parents. Furthermore, such gains as are obtained in one generation of selection will not necessarily persist in subsequent generations (LUSH 1945).

The removal of the second restriction noted (independence of the average genotypic value from the environmental variance) may cause the regression of genotype on phenotype to be non-linear. In this situation, the proportion of the phenotypic selection differential which will be realized as a genetic gain will depend on selection intensity and other factors. This will generally lead to a positive or negative bias of *estimate of gain* based on the conditions of strict additiveness.

HERITABILITY OF ALL-OR-NONE CHARACTERS

Most of the applications of the concept of heritability have until recently been made with respect to characters showing a continuous distribution on the phenotypic scale. The importance of this statistic, however, extends equally to traits which, while possibly based on a continuous or normal distribution of genotypic values, may be expressed phenotypically on an all-or-none, or generally speaking discontinuous basis.

It was once more WRIGHT (1920) who first dealt with the general subject. Since the average phenotype of groups of related individuals for such traits can be expressed in terms of percentage incidence, he evolved a transformation of percentage data to a normalized form. The transformation was described by him in detail (1926) under the name of "inverse probability." Essentially the same method was later proposed by BLISS (1935) as the "probit" transformation. It is the latter term which has gained currency in the literature and

under which the statistical properties of transformed distributions were described (for instance by FINNEY 1947).

By the use of his transformation WRIGHT was able to determine the relative amounts of genetic and environmental variation in such traits as digit number in guinea pigs (1934b, 1934c) and flower color in Linanthus (1943). Subsequently LUSH, LAMOREUX and HAZEL (1948) introduced the subject into the realm of animal breeding by considering the heritability of viability in the fowl, a matter which also received attention from ROBERTSON and LERNER (1949).

In general, it seems reasonable to treat characters with an all-or-none phenotypic expression as dependent on one or several more or less continuously variable underlying variates. The value of each underlying variate in a particular individual, according to this view, would depend both on genetic and non-genetic factors, and the appearance of the character would require that some threshold be exceeded. The genotype thus determines a probability of the character appearing, or the proportion of environments in which it will actually come to expression.

This viewpoint permits the determination of heritability of a character measured in terms of its probability of appearance, with values of 0 and 1 assigned to the alternative phenotypic expressions. This scale of measurement will be referred to henceforth as the *p scale*. Heritabilities determined from it may provide a basis for computation of genetic gains expected from selection and for construction of selection indexes. However, in some instances, serious inaccuracies may result, since genetic variance which may be completely additive for the underlying variate may lose this property on the p scale. Since the limiting genotypic values are 0 and 1, it is highly unlikely that a given gene substitution would have the same effect near these limits as in the middle of the range.

Similarly the environmental variance of the underlying variate may be independent of the mean genotypic value (the level of incidence or p), but this property may be lost on the p scale. This is apparent from the fact that environmental variance on the p scale, which, of course, is the total variance for any given fixed genotype, is equal to pq, where $q = 1 - p$. For the range of genotypes with p values from, say, .35 to .65, the environmental variance is reasonably constant, but in the extreme ranges of p from 0 to 0.1 and from 0.9 to 1.0, it is nearly directly proportional to p and to q respectively. Finally, the categorical nature of the p scale may obscure the finer degrees of measurable variation (as for example age of death, where viability in a set period of time is considered). All of these circumstances vitiate to some extent the utility of the degree of heritability determined on the p scale.

LUSH, LAMOREUX and HAZEL (1948) held that probit transformation avoids some of the objections to the p scale. As in the previously noted work of WRIGHT, the transformation is based on the concept of an underlying variate with a normal environmental distribution, whose variance is independent of the genotypic level. The heritability on the probit scale is independent of the threshold value, above which the character will be manifested in individuals whose underlying variate exceeds it, whereas on the p scale heritability (pro-

viding the genetic variance is not too large compared to the environmental variance) varies approximately in proportion to $\bar{z}^2/(\bar{p}\bar{q})$, where $\bar{z}$ is the ordinate of a unit standard normal curve cutting off an area equal to $\bar{p}$. The p scale heritability then, in terms of the probability of an individual exhibiting the character, would be low for values of $\bar{p}$ near zero or unity and relatively high for intermediate values. ROBERTSON and LERNER (1949) have shown that the situation is similar where there are a number of underlying variates, although the distribution of h^2 is, of course, somewhat different in form.

The probit transformation may be a satisfactory one for the purpose of comparing heritability values but it is not apparent how it can be used for devising optimum selection indexes nor is it at all convenient in many situations for computing expected rates of gain. It appears useful therefore to investigate the conditions, if any, under which serious errors are likely to result from the use of the convenient p scale and the nature and magnitude of such errors. Such an investigation might disclose that the p scale can be used without hesitation in many situations and with some reservations or with corrections in others. The results of such a study might also suggest in a general way the degree to which a scale can deviate from the optimum one without leading to selection indexes that depart seriously from the optimum or to computations of gain that are much in error. Where calculations of the gain from mass selection based on the p scale lead to correct results there is good reason to conclude that calculations of the gain from family and combined selection will also be accurate, as well as that the usual methods for the computation of optimum indexes will also be applicable to a high degree of approximation. The converse of this statement is, however, not necessarily valid. The present investigation is chiefly concerned with a study of the gains that would result from mass selection on an all-or-none basis in comparison to estimates of gains whose computations are based on heritability determinations on the p scale, as well as with gains that would result could selection be based on direct observation of the underlying variate. The bearing of these findings on indexes for combined individual and family selection is discussed briefly.

THE MATHEMATICAL MODEL

The first step in the proposed investigation involves the comparison of genetic gains computed on the basis of h^2 determined from the p scale with those expected on the postulate of a normally distributed underlying variate. The assumptions involved are that:

1. There is an underlying variate whose value is the sum of a normally distributed environmental component and an independent normally distributed genetic component.

2. The character is present in all those individuals, and only those, in which the underlying variate exceeds a certain threshold value.

3. Gene substitutions have individually small and strictly additive effects on the underlying variate.

There is no claim made here that these conditions actually describe the

situation with respect to all characters of an all-or-none nature. It is, however, reasonable to believe that a model in which additiveness is the property of the underlying variate (on what will henceforth be referred to as the *x scale*) approaches actual situations more closely than one based on additive gene action on the p scale.

Figure 1 illustrates the model postulated. The solid-line curve (b) is a normal curve of unit area representing the distribution of the underlying vari-
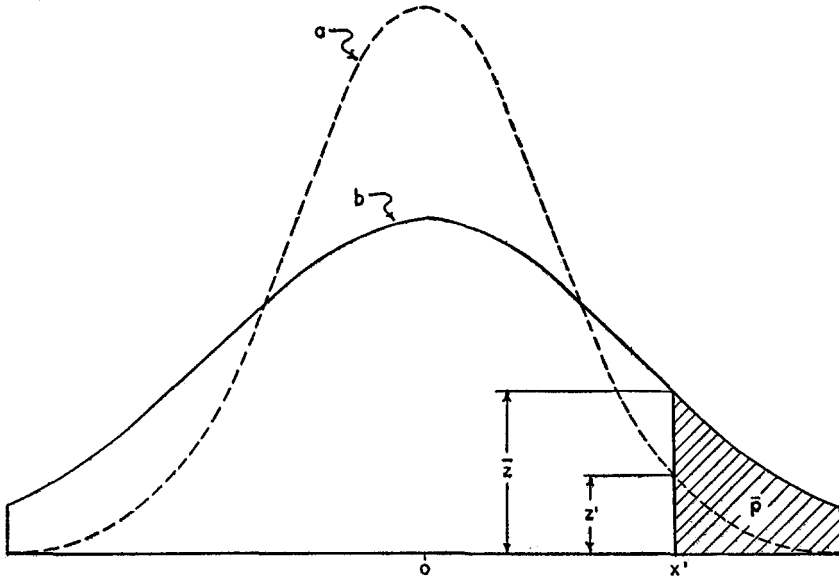

FIGURE 1. The model investigated. The solid line curve *b* represents the distribution of all individuals in the population. The abscissae (x) are values of the underlying variate expressed in standard deviation units. If this variate in any individual exceeds the threshold value x', the character is expressed and $\bar{p}$ is the proportion of all individuals in the population exhibiting the character. The dotted curve a, having the environmental standard deviation $\sqrt{1-\sigma G_x{}^2}$, represents the distribution of x for all individuals possessing the mean genotype of zero on the x scale. If this curve is shifted to the right or left so that its mean is not zero, it then represents the distribution of x for all individuals whose genotype on the x scale is the new mean. The area to the right of x' under any such curve is the corresponding genotype on the p scale.

ate. It is convenient to take the standard deviation of this distribution as the unit of measurement on the x scale. The dotted curve (a) is a normal curve of unit area representing the distribution of all those individuals that possess the *mean* genotypic value; this distribution is therefore the environmental distribution and its variance is $(1-h_x{}^2)$ where $h_x{}^2$ is the heritability of the underlying variate and is also equal to the genetic variance on the x scale. The ordinate drawn to the right of the mean is the threshold value, all individuals to the right of this ordinate exhibiting the character.

The abscissa corresponding to the ordinate cutting off a tail of area $\bar{p}$ where $\bar{p}$ is the incidence of the character in the population is represented by x'. Its value can be obtained from tables of the normal distribution. The height

of the ordinate ($\bar{z}$) can also be similarly obtained. Since the dotted curve has unit area and a standard deviation of $\sqrt{1-h_x^2}$, the area under this curve to the right of the threshold is that of a unit standard normal distribution lying to the right of abscissa $x'/\sqrt{1-h_x^2}$. This area is the proportion of all those individuals possessing the *mean genotype* that exhibit the character. The proportion of all those individuals possessing *some other fixed genotypic value* that exhibit the character can similarly be obtained if one first subtracts from this abscissa the product of the factor $1/\sqrt{1-h_x^2}$ and the difference between the genotypic value in question (on the x scale) and the mean genotypic value. Genetic differences are thus thought of as shifting the mean of the environmental distribution, the threshold remaining fixed. Alternatively, they can equally well be thought of as shifting the threshold abscissa, the mean of the environmental distribution remaining fixed. Using either concept the genotype of an individual on the p scale is the proportion of environments in which it would exhibit the character.

For every genotypic value on the x scale there is a corresponding value (that is, proportion of individuals exhibiting the character) on the p scale. The determination of p values corresponding to given x values is easily accomplished by the method outlined above. Some idea of the degree of disparity of the two scales can be obtained by marking off a linear scale in x units and marking the corresponding points on a parallel scale in p units. Figure 2
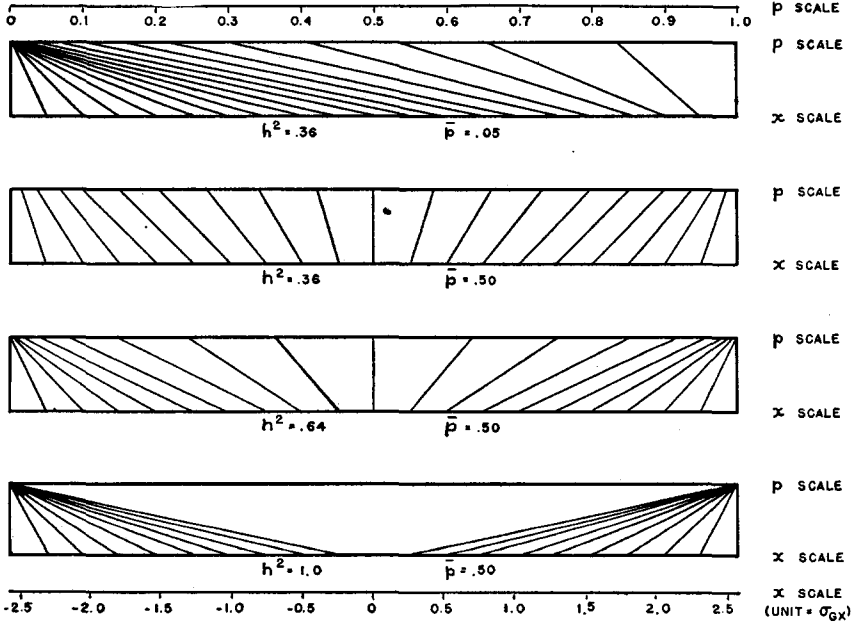

FIGURE 2. Comparison of x and p scales. The slanting lines in each example connect genotypes of the x scale (lower), on which gene differences are postulated to have additive effects, with corresponding genotypes of the probability or p scale (upper). The variability of the spacing of the intersects on the p scale is a measure of the inequality of the effects of gene substitutions expressed on this scale at different genotypic levels.

shows some scale comparisons of this sort for heritabilities of 0.36, 0.64 and 1.0 and for incidences of 0.05 and 0.5. The range shown is ± 2.58 standard deviations of the genetic distribution each side of the mean so that 99 percent of genotypes are included. The distribution of p values for the case in which the underlying variate has a heritability of unity is, as shown, discontinuous. Inspection of these scale comparisons indicates but little disparity where heritability is low and the incidence close to 0.5 but may lead to some doubt as to the applicability of the usual estimation of gains from selection on the basis of heritability determinations where h_x^2 is high or the incidence is near zero or unity.

EMPIRICAL DETERMINATION OF ADDITIVE HERITABILITY ON THE P SCALE

Before proceeding with the discussion of the relationship between heritabilities on the x and p scales it may be worth while to present the method of heritability estimation on the p scale as outlined by ROBERTSON and LERNER (1949) based on actual data, which may also serve to illustrate the relationship between the heritability on the p scale and that on the x scale.

The data used for the present purpose are derived from the material reported on by LERNER and CRUDEN (1948). They computed the heritability of egg production for various periods of time of birds surviving the first laying year in a flock of Single Comb White Leghorns. The roughly normal underlying variate considered here is the total first year egg production (from beginning of lay till September 30th of the second year of life) of the same birds. Thus the heritability estimates computed by the conventional methods represent in our notation h_x^2.

The estimates presented by LERNER AND CRUDEN were based on two separate series of birds hatched in consecutive years. Each series in turn contained two sub-flocks (lines), one selected primarily for egg production, and

TABLE 1

The material used in the analysis (derived from LERNER and CRUDEN 1948).

SERIES	LINE	NUMBER OF SIRES	NUMBER OF DAMS	NUMBER OF DAUGHTERS	AVERAGE GENETIC RELATIONSHIP WITHIN FAMILY	h_x^2 ESTIMATE BY LERNER AND CRUDEN
S	Production	6	22	111	.314	− .034
	Others	11	50	235	.297	.587
	Total	17	72	346	.302	.381
T	Production	10	47	226	.296	.272
	Others	11	46	225	.305	.423
	Total	21	93	451	.301	.358
S+T	Total	38	165	797	.301	.369

the other for various other traits listed in their report. The distribution of birds by sub-flocks and series is shown in table 1.

Several methods of h_x^2 computation were employed by LERNER and CRUDEN. The estimate shown in table 1 is the one based on information from the correlations between both full sisters and half sisters. For our purposes the values of h_x^2 thus reported need some adjustment, because the p scale analyses to be presented were conducted by partitioning the total variance into that between and within sire families. Each sire family was therefore a mixture of full and half sisters. Hence, the genetic relationship within each family was greater than 0.25 (the value for half sisters) and less than 0.5 (the value for full sisters). The average genetic relationship was computed and is also given in table 1. Having obtained this constant the heritability values reported by LERNER and CRUDEN were recomputed on the basis of the usual statistical design.

The raw data were arranged for the p scale analyses in order of magnitude of individual egg production records for each sub-population separately. For combined sub-populations (by series) the data were rearranged in a similar fashion and again for the total group of birds. Nine levels of incidence were analyzed, from 0.1 to 0.9 ranging in intervals of 0.1. Thus for the lowest level the top ten percent of each sample considered were given values of 1 and the rest of the sample, values of 0. For the second level the top 20 percent were assigned values of 1 and the rest values of zero, and so forth.

The formulas derived or cited by ROBERTSON and LERNER (1949) were then applied to the data. The estimates of heritability obtained by this method include a certain amount of non-additive variance and will be designated as h_p^2, as contrasted to h_{pa}^2 for strictly additive and h_{pt}^2 for the total heritability (see following section). By way of recapitulation the essential expressions used were the following ones:

$$h_p{}^2 = \frac{\chi^2 - (N - 1)}{rn_0},$$

where

$$n_0 = \sum n - \frac{\sum n^2}{\sum n} - (N - 1),$$

while   N = number of sires,
     n = number of daughters per sire,
      r = the genetic relationship between the members of a sire family.
and χ^2 = the heterogeneity Chi-squared in the 2 × N table.
The standard error of estimate of h_p^2 is σ_t/r,
where

$$\sigma_t = \frac{[1 + (n - 1)t](1 - t)\sqrt{2}}{\sqrt{n(n - 1)(N - 2)}},$$

and   t = the phenotypic correlation between the members of the sire family
        and equals rh_p^2.

The h_p^2 values may be compared with the h_{pa}^2 transformations from h_x^2 by the method described in the following section $(h_{pa}^2 = \bar{z}^2 h_x^2 / \bar{p}\bar{q})$, which appear in the last line of table 2. It may be seen that in general $h_p^2 > h_{pa}^2$ as

TABLE 2

Heritability estimates.

SERIES	LINE	h_x^2 FROM TABLE 1	RECOMPUTED h_x^2	ESTIMATES FROM BINOMIAL DATA FOR GIVEN LEVEL								
				.1	.2	.3	.4	.5	.6	.7	.8	.9
S	Production	−.034	.030	−.14	−.09	−.11	−.03	−.08	−.10	−.10	.18	.17
	Others	.587	.530	.18	.37	.42	.50	.45	.47	.45	.27	.19
	Total	.381	.358	.08	.23	.26	.33	.28	.29	.28	.24	.18
T	Production	.272	.236	0	−.05	.06	.09	.06	.10	.11	.17	.07
	Others	.423	.472	.22	.35	.55	.49	.33	.31	.22	.23	.35
	Total	.358	.364	.11	.16	.31	.30	.20	.20	.17	.20	.21
S+T	Total	.369	.362	.10	.19	.29	.31	.23	.24	.26	.22	.20
	Standard Error of Estimate			.06	.08	.10	.10	.08	.09	.09	.08	.08
	h_{pa}^2 computed from x scale			.12	.18	.21	.22	.23	.22	.21	.18	.12

would be expected from the fact that some non-additive variance is included in the first. By and large, however, the agreement between h_p^2 and h_{pa}^2 is in the light of the standard errors of estimate reasonably good.

COMPARISON OF HERITABILITIES ON THE P AND THE X SCALES

There is no distinction between additive and total heritability on the underlying or x scale because, in accordance with the postulates listed, gene differences have strictly additive effects on this scale. On the p scale, however, some of the genetic variance is non-additive and the non-additive portion may be very large where heritability on the x scale is very high or the incidence close to 0 or 1.0. It is therefore desirable for the purposes of the present investigation to compute both the additive heritability (additive portion of genetic variance divided by total variance) and the total heritability (total genetic variance divided by total variance).

For the special case in which $h_x^2 = 1.0$, expressions giving the total and the additive heritabilities on the p scale are easily obtained. Here, all of the variance on the p scale is genetic and is equal to $\bar{p}\bar{q}$. Multiplication of this value by the square of the correlation (r_{xp}^2) between p and x will yield the additively genetic variance. This correlation is the covariance divided by the square root of the product of the variances of p and x; since p equals zero for values of x less than x' and equals unity for values of x greater than x', the covariance can be expressed as follows:

$$\frac{\int_{-\infty}^{+\infty} (0) y x \, dx + \int_{x'}^{+\infty} (1) y x \, dx}{\int_{-\infty}^{+\infty} y \, dx} - \bar{x}\bar{p}.$$

The first term in the numerator is zero as is also the product $\bar{x}\bar{p}$ because the mean value of x is zero. The denominator of the fraction is equal to unity. The remaining integral, and hence the covariance, is simply

$$\left[-\frac{1}{\sqrt{2\pi}} e^{-x^2/2} \right]_{x'}^{\infty} = \bar{z}.$$

The variance of x is unity and that of p is $\bar{p}\bar{q}$, so the expression for r_{xp} becomes $\bar{z}/\sqrt{\bar{p}\bar{q}}$. The product of the square of this correlation and the total genetic variance $(\bar{p}\bar{q})$, is equal to $(\bar{z}^2/\bar{p}\bar{q})\bar{p}\bar{q}$ or to $\bar{z}^2$, which is thus the additively genetic variance. The additive heritability on the p scale (h_{pa}^2) is then for this case equal to $\bar{z}^2/\bar{p}\bar{q}$.

This expression in a more general form,

$$h_{pa}^2 = \bar{z}^2 h_x^2/\bar{p}\bar{q},$$

can be used irrespective of the value of h_x^2. The rationale behind the deduction can be briefed as follows. Were the genotypic values on the two scales linearly related, the genetic variance on the p scale would be equal to that on the x scale multiplied by the constant term $(dp/dx)^2$. The relation is not linear and dp/dx is not constant, but the use of the mean value of dp/dx will yield the additive genetic variance.[2] The height of the ordinate at x', which for a population of individuals all possessing the mean genotype is z' (fig. 1), is the value of dp/dx. The weighted mean value of z' for all genotypes in the population is also the value of z which corresponds to $\bar{p}$ and is indicated by $\bar{z}$. The additive genetic variance on the p scale $(\sigma_{G_{pa}}^2)$ is then: $\sigma_{G_x}^2 \bar{z}^2 = \bar{z}^2 h_x^2$. The additive heritability on the p scale is this value divided by the total variance which, in a dichotomous distribution, is $\bar{p}\bar{q}$. We have then, $h_{pa}^2 = h_x^2 \bar{z}^2/\bar{p}\bar{q}$.

The total heritability of character incidence on the p scale (h_{pt}^2) may be approximated in the following manner. The genetic distribution in terms of the x scale, which has been postulated to be normal in form and has a variance of h_x^2, is divided into a number of segments by ordinates spaced along the x axis. The genotype on the p scale of every individual within a segment is considered, as an approximation, to correspond to the mean genotype on the x scale of all individuals represented by this segment. The genetic variance of these p values, each weighted in proportion to the area of the corresponding segment, is then an approximation to total genetic variance. The quotient of this value by $\bar{p}\bar{q}$, the total variance, is an approximation to total heritability on the p scale. This heritability calculation can be carried to any desired degree of accuracy by choosing an appropriate number of segments. The results obtained by this method (the "segmental" method of table 3) in the present study are based on a division of the genetic distribution into 20 equal segments and a further division of the end segments into five equal sub-segments each, making 28 segments in all.

[2] See Appendix for a demonstration that the use of this mean value yields an exact expression for the additive genetic variance.

TABLE 3

*Heritabilities and genetic variances on the x and p scales.**

h_x^2	VARIATE	METHOD OF CALCULATION	INCIDENCE					
			.05 .95	.1 .9	.2 .8	.3 .7	.4 .6	.5
0–1	h_{pa}^2/h_x^2	$\bar{z}^2/\bar{p}\bar{q}$	.224	.342	.490	.576	.622	.637
.36	$\sigma_{G_{pt}}^2$	Segmental —	.00583	.0145	.0320	.0463	.0555	.0586
.36	$\sigma_{G_{pa}}^2$	$(\sigma_{G_{pt}}^2)(r_{xp}^2)$—segmental	.00378	.0111	.0282	.0436	.0538	.0574
.36	$\sigma_{G_{pa}}^2$	$\bar{z}^2 h_x^2$ —	.00383	.0111	.0282	.0435	.0537	.0573
.36	$\sigma_{G_{pe}}^2/\sigma_{G_{pt}}^2$	$1-r_{xp}^2$ —segmental	.353	.236	.120	.0600	.0294	.0205
.36	h_{pt}^2	$(\sigma_{G_{pt}}^2)(r_{xp}^2)$—segmental	.123	.161	.200	.221	.231	.235
.36	h_{pa}^2	$\bar{z}^2 h_x^2/(\bar{p}\bar{q})$ —	.0806	.123	.176	.207	.224	.229
.64	$\sigma_{G_{pt}}^2$	Segmental —	.0146	.0319	—	.0896	—	.111
.64	$\sigma_{G_{pa}}^2$	$(\sigma_{G_{pt}}^2)(r_{xp}^2)$—segmental	.00677	.0197	—	.0776	—	.102
.64	$\sigma_{G_{pa}}^2$	$\bar{z}^2 h_x^2$ —	.00681	.0197	.0502	.0774	.0955	.102
.64	$\sigma_{G_{pe}}^2/\sigma_{G_{pt}}^2$	$1-r_{xp}^2$ —segmental	.535	.383	—	.134	—	.0760
.64	h_{pt}^2	$\sigma_{G_{pt}}^2/(\bar{p}\bar{q})$ —segmental	.307	.355	—	.427	—	.442
.64	h_{pa}^2	$\bar{z}^2 h_x^2/(\bar{p}\bar{q})$ —	.143	.219	.314	.368	.398	.407
1.0	$\sigma_{G_{pt}}^2$	$\bar{p}\bar{q}$ —	.0475	.09	.16	.21	.24	.25
1.0	$\sigma_{G_{pa}}^2$	$\bar{z}^2$ —	.0106	.0308	.0784	.1209	.1493	.1592
1.0	$\sigma_{G_{pe}}^2/\sigma_{G_{pt}}^2$	$1-\bar{z}^2/(\bar{p}\bar{q})$ —	.7761	.6578	.5101	.4243	.3781	.3634

* Empty cells represent non-essential values.

The segmental method can be extended so as to provide a technique of computing the proportions of the total variance that are additive and non-additive, and hence also of a determination of h_{pa}^2 alternative to the exact one described above. The alternate determination provides a check on the degree of approximation involved in the segmental procedures. The proportion of the total genetic variance that is additive is equal to the square of the correlation between the additively genetic values and the genotypic values on the p scale of the individuals in the population (r_{xp}^2). This correlation is the same (under the condition that all gene substitutions have small individual effects) as that between corresponding genotypic values on the x and p scales because, under the condition mentioned, the effects of all possible gene substitutions for any given genetic background, and hence also the mean effect on the p scale for all genetic backgrounds, will be in direct proportion to the effects on the x scale. An approximation to this latter correlation can ·be obtained by dividing the covariance of the genetic values on the p and x scales of the segments of the genetic distribution by the square root of the product of the variances of these values. The product of the square of this correlation and the total genetic variance is then an approximation to the additively genetic variance,

$$\sigma_{G_{pa}}^2 = \sigma_{G_{pt}}^2 r_{xp}^2,$$

while the remainder of $\sigma_{G_{pt}}^2$ is the non-additive variance. The latter, on the assumption that individual gene differences have small effects, is epistatic and may be indicated by $\sigma_{G_{pe}}^2$. We may, then, write:

$$\sigma_{G_{pe}}^2 / \sigma_{G_{pt}}^2 = 1 - r_{xp}^2.$$

This method of estimating epistatic variance is probably more accurate than that based on the difference between the approximately determined total variance and the exactly computed additive variance.
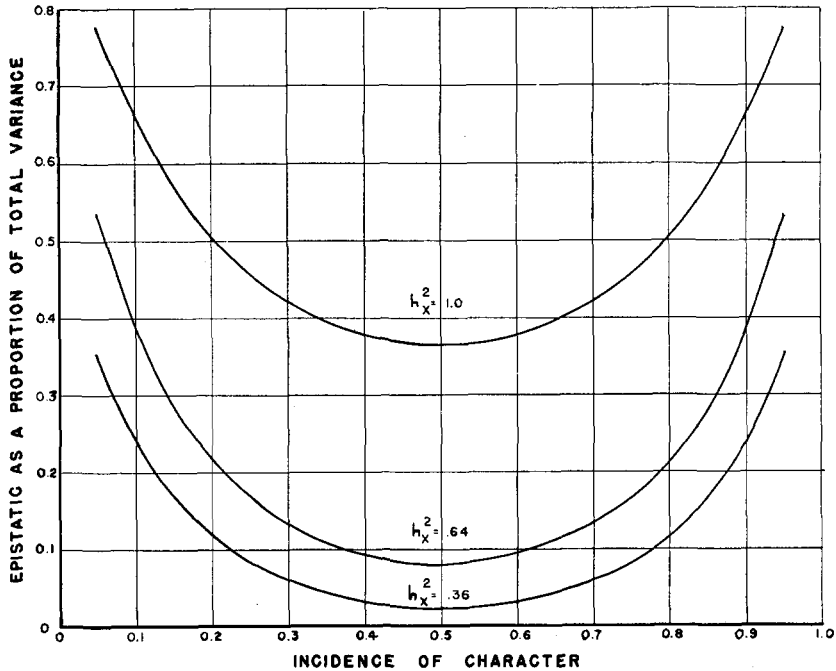

FIGURE 3. The proportion of total genetic variance that is epistatic, expressed as a function of character incidence in the population.

Table 3 presents the p scale estimates of variances and heritabilities obtained by the methods described for heritabilities of 0.36, 0.64, and 1.0 at varying levels of incidence. Very close agreement between the two sets of values of $\sigma_{G_{pa}}^2$ (segmental versus the exact) may be noted for h_x^2 of 0.36 and 0.64. Approximate methods are not required where h_x^2 approaches zero or unity.

Ratios of epistatic to total genetic variance on the p scale are also listed in table 3 and shown graphically in figure 3. It may be noted that the epistatic variance is trivial where the heritability is low and the incidence near 0.5 but may constitute most of the genetic variance under other conditions, having a value of over 77 percent of the total variance under the most extreme condition listed. It will be shown later that serious errors resulting from the use of heritability on the p scale in estimating gains from mass selection occur

only when epistatic variance forms a considerable proportion of the total genetic variance and when selection intensity is relatively high.

THE PROPORTION OF EPISTATIC VARIANCE INCLUDED IN HERITABILITY ESTIMATES

Intraclass correlations between full and half sibs are commonly used to estimate the degree of heritability of a trait (that is, the h_x^2 values in table 1). Since non-additive genetic variance does not contribute as much to such correlations as additive genetic variance does, such estimates will tend to be of a magnitude intermediate between additive and total heritability. The classic investigation of the contribution made by non-additive genetic variance to the correlation between relatives was carried out by WRIGHT (1935). In the model studied by him the values of a secondary character depend on the deviations of a primary character from an optimum. Here (as in our model) the genetic variance of the secondary character is partly epistatic even when that of the primary is completely additive. WRIGHT found, for the case of many genes, exclusively epistatic genetic variance, and no environmental variance, that the correlation between full sibs is one quarter; the value under the same conditions for half sibs is one sixteenth. These values are to be multiplied by the square of the heritability (h^4) of the primary character where environmental variance is present. However heritability on the secondary scale, all non-additive (epistatic) under the conditions mentioned, is the square of heritability on the primary scale, or h^4. Thus the correlation between full sibs on the secondary scale is one quarter of the heritability on the same scale; similarly the correlation between half sibs on the secondary scale is one sixteenth of the same value. If therefore the correlation between full sibs is doubled to obtain a heritability estimate (as is appropriate in the case of additive genetic variance) the result is only half the actual heritability. Likewise quadrupling the half-sib correlation yields one quarter the actual heritability. Finally, the variance components due to different dams mated to the same sire yields a heritability estimate including the proportion $4[(1/4)—(1/16)]$ or three quarters of the genetic variance.

For the model considered here, the fraction of the non-additive variance included in the estimates of heritability can be readily determined from a comparison of the previously calculated values for additive and total heritability with the estimates obtained from the three variance components noted in the previous paragraph. The total variance is equal to $\bar{p}\bar{q}$. If the p scale variance within full sib families is subtracted from this total, the variance component due to different sires and dams (intraclass full sib correlation) is obtained. Similarly the difference between the total variance and that within half sib families yields the variance component due to different sires (intraclass half sib correlation). Finally the difference between the two components thus obtained is the component due to different dams mated to the same sire.

The problem then calls for the determination of intra-family variances on the p scale. For full sibs in a population under random mating and in equilibrium the variance of family means on the x scale equals one-half of the genetic variance ($\sigma_G^2/2$). If the mean abscissae of segments of a unit normal

curve are multiplied by the square root of this variance, the true mean geno-
types on the x scale of a representative array of families are obtained. The
difference between such a mean value of any family and the threshold abscissa
for the whole population is the effective threshold, in terms of total standard
deviation units, for the family in question. However, the total variance
within the family is only $(\sigma_G^2/2)+\sigma_E^2$ (where σ_E^2 represents environmental
variance). Hence in order to obtain the equivalent threshold abscissa for a
standard unit normal curve, the computed threshold abscissa for the family
must be divided by $\sqrt{(\sigma_G^2/2)+\sigma_E^2}$. The area of the normal curve (p) cor-
responding to this standardized abscissa is, of course, the incidence of the
character in the particular family, while $p(1-p)$ is the total variance within
the family. Such variances can be computed for the whole array of families
and their mean (weighted for the relative frequency of each kind of family
in the population) is the total variance within full sib families. The analogous
value for half sib families is obtained in a similar fashion. There the variance
of family means on the x scale is $\sigma_G^2/4$, and the variance within families
$(3/4)\sigma_G^2+\sigma_E^2$.

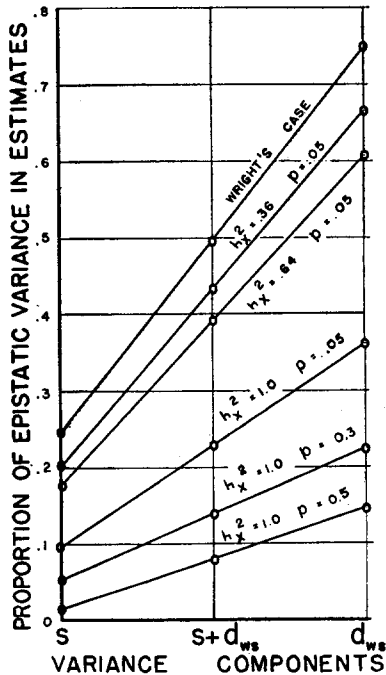It should be noted that when the differences between the total and the

FIGURE 4. The proportion of the epistatic variance included in the numerator of heritability
estimates based on variance components. Here *s* represents the component due to different sires,
$s+d_{ws}$ represents the component due to the combined effect of different sires and dams, and d_{ws}
represents the component due to different dams mated to the same sire.

within family variances are small, slight errors in the determination of the latter are considerably magnified in the final results. Some gain in accuracy can be achieved by computing directly the variance of the incidences in the array of families about their own mean, a method identical to the one described if the product of the mean incidence of the array of families by its complement is used as the total variance. This mean incidence is slightly different from the mean incidence in the population because of the approximation involved in taking the means of each segment as representative of all families within the segment. The modified method is sufficiently accurate for our purposes where the epistatic variance is not too small a proportion of the total variance. The calculations have been made, therefore, for heritabilities on the x scale of 0.36 and 0.64 with a population incidence of 0.05, and for an $h_x^2 = 1.0$ with population incidences of 0.05, 0.3 and 0.5. For the last three cases the determination of the proportion of epistatic variance included in the three kinds of estimates is probably correct within one to three parts in a hundred; for the first two the errors may be as high as one part in ten or twenty.

Figure 4 shows the proportions of epistatic variance, calculated in the manner just described, for the three types of heritability estimates considered. The proportions for WRIGHT's model are shown for comparison. Although the proportions for the latter model do not depend on heritability, those on the p scale increase with decreased heritability, at least where the incidence is near zero or unity. The possibility is suggested that the maximum proportions may equal the corresponding ones for WRIGHT's model. The components due to differences between sires by and large contain but little epistatic variance; those due to differences between dams mated to a sire include considerably more of it. In general it appears that in this model heritability estimates, based on the methods discussed, are likely to lie closer to additive heritabilities than to total heritabilities. Exceptions occur where the epistatic component is small and hence relatively less important.

THE COMPARISON OF CALCULATED AND ACTUAL GAINS FROM MASS SELECTION

In mass selection the estimated gain based on heritability on the p scale is simply the product of this heritability and the difference between the proportion of parental individuals exhibiting the character and the incidence in the unselected parental generation. Calculations have been made, based on the selection as parents of all individuals exhibiting the character, for h_x^2 of 0.36, 0.64, and 1.00 and for a range of incidences in the parental generation from 0.05 to 0.95. For each case two heritabilities on the p scale have been used, the additive heritability and the additive plus one quarter of the epistatic heritability. In view of the results shown in figure 4, most cases would be expected to fall within these limits. For comparison with these values, the actual gains that would be achieved have been calculated on the following basis.

The expected mean phenotypic value of the offspring of a group of parents

is equal to the parents' mean genetic values on the x scale, provided there is no correlation between the genetic values of the parents and the numbers of offspring they produce. In terms of the p scale the genetic gain from mass selection may be defined as the increase in the expected incidence of the offspring over the incidence in the parental generation (the selected parents being mated at random). An assumption to be made here is that the genetic variance on the x scale of such offspring is equal to that of the parental generation.

The mean abscissa of individuals possessing the character in a population with an incidence of p_p is equal to $\bar{z}/p_p$. Should all such individuals (or a ran-
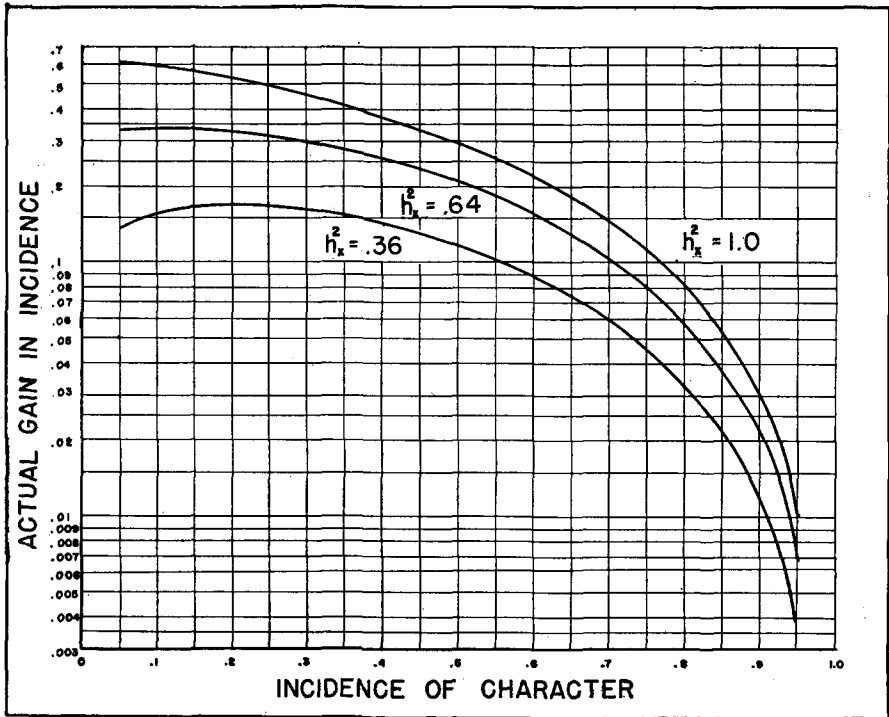
FIGURE 5. Actual gain in character incidence due to use as parents of all individuals exhibiting the character, expressed as a function of character incidence in the population.

dom sample of them) be used as the parents of the next generation, the mean genetic advantage of the selected animals and hence the genetic gain on the x scale is $h_x^2\bar{z}/p_p$. Subtraction of this gain from the distance between the threshold and the mean of the parental generation gives the corresponding threshold distance for the offspring. The area to the right of this threshold is then the incidence of the character in the offspring (p_0) and the difference $p_0 - p_p$ is the genetic gain on the p scale.

The gains thus computed under the conditions noted are illustrated in figure 5. It may be seen that mass selection *for* a character of high incidence (or *against* one of low incidence) leads to slight gains only. This is due to very low effective heritability, combined with the fact that selection intensity here

is also perforce low. For intermediate incidence the gains obtained are relatively high, but further shift in incidence does not produce much greater gains, because the decrease in effective heritability offsets and eventually overcomes the increase in selection intensity incident on this shift.

The percentage by which the gains computed on the basis of additive heritabilities, and additive plus a quarter epistatic heritabilities, under- or
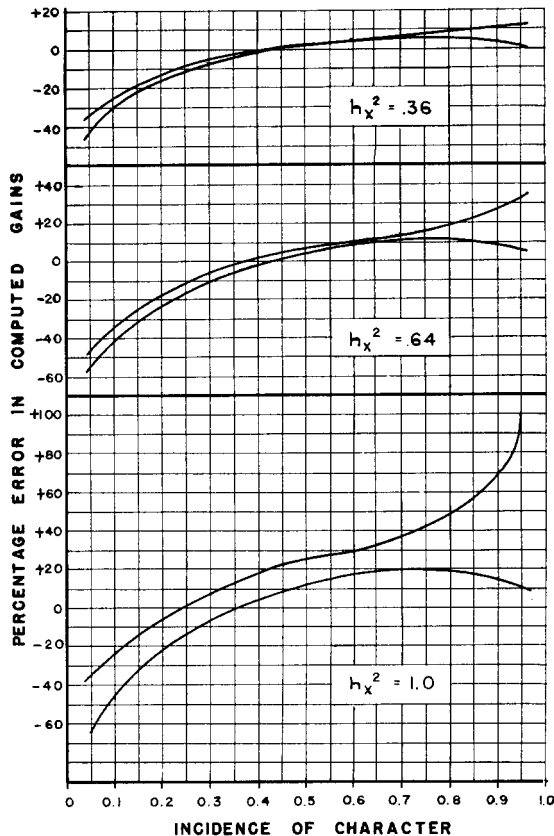

FIGURE 6. Errors in estimated gains based on p-scale heritabilities expressed as a percentage of actual gains, for the case where all individuals exhibiting the character are used as parents. The lower curve for each value of h_x^2 is based on additive heritability and the upper curve on additive plus one quarter of epistatic heritability.

over-estimate the actual gains are shown in figure 6. It will be noted that the actual rate of gain in the selection for a rare character may be more than twice that calculated on the basis of p scale heritabilities. On the other hand, in the case of characters that are already very frequent in the population the rate of gain is likely to be somewhat less than that calculated, but the discrepancy is relatively small except where the epistatic variance is very great. High epistatic variance, on the model discussed, occurs only where the heritability on the x scale is high or the incidence is very close to zero or unity.

If the selection intensity for the character is lower than the proportion of

individuals exhibiting it, it is necessary to use as parents of the next generation some individuals that do not exhibit the character. In this case the gain computed on the basis of p scale heritabilities is the weighted mean of the products of the heritability by the phenotypic advantages on the p scale of individuals exhibiting and not exhibiting the character. The latter values are
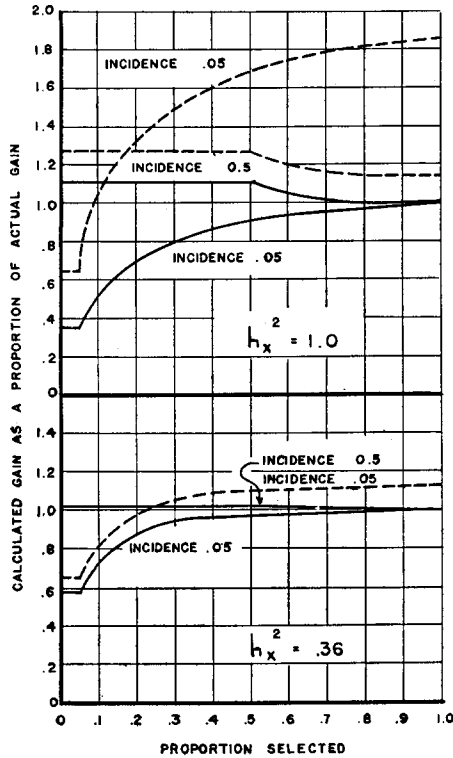

FIGURE 7. Errors in estimated gains based on p-scale heritabilities expressed as a proportion of actual gains, for cases where all individuals exhibiting the character and various proportions of individuals not exhibiting the character are used as parents. The solid line curve for each value of h_x^2 is based on additive heritability and the broken curve on additive plus one quarter of epistatic heritability. The two curves for $h_x^2 = .36$ and p=.5 are indistinguishable from each other on the scale portrayed.

of course $1-\bar{p}$ and $\bar{p}$ respectively, while the weighting is the proportion of parental individuals falling into the two classes. In computing the actual gain, the change in threshold value is computed in a similar fashion. Figure 7 shows the discrepancies between the actual gains and those expected on the basis of the p scale for certain cases where all individuals in the population exhibiting the character and various proportions not exhibiting the character are used as parents. These curves, as well as those in figure 6, suggest that calculations on the basis of additive heritability will always yield accurate estimates of the gain where the selection intensity is very low.

THE EFFICIENCY OF MASS SELECTION OF THRESHOLD CHARACTERS

If the proportion of individuals required as parents is exactly equal to the incidence of the character in the population so that all such individuals and no others are used as parents in a mass selection program, the ensuing improvement is exactly the same, in the case of the model being discussed, as
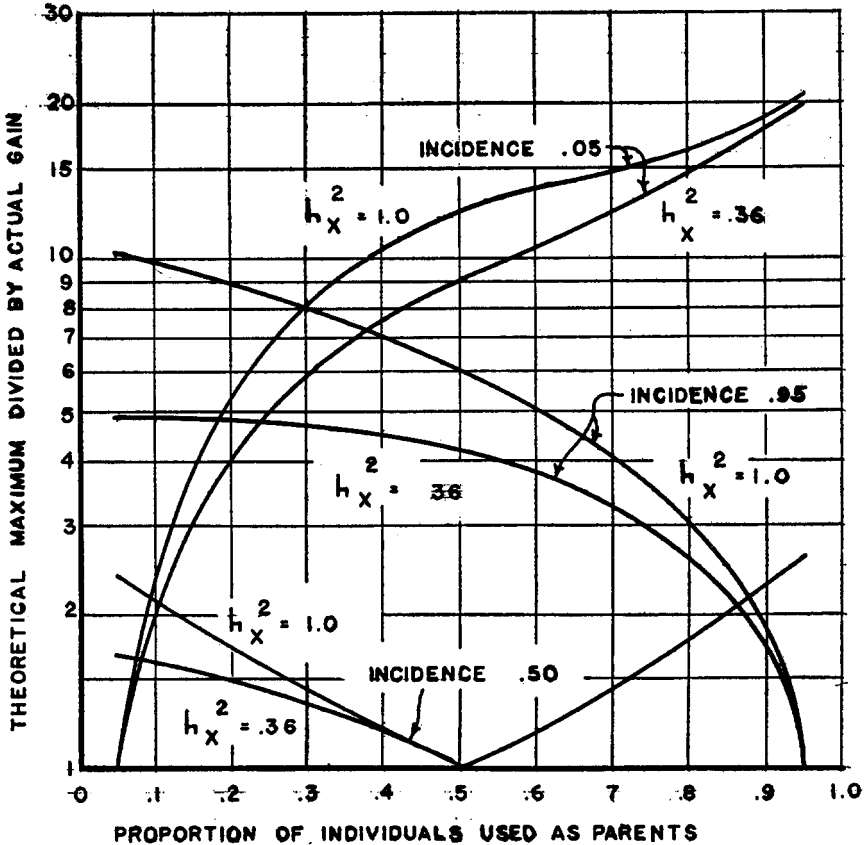

FIGURE 8. Gains that can be achieved on the basis of the all-or-none classification expressed as the reciprocal of the proportion of the gains that could theoretically be achieved on the basis of the underlying variate, for different proportions of individuals in the population that are used as parents.

though selection were based directly on the underlying variate. On the other hand, selection directly on the basis of the underlying variate would produce greater gains when a larger number of parents is required than the number of animals in the population which exhibit the character, or when a smaller number than that exhibiting the character is selected at random. The proportion of the maximum gains theoretically obtainable which are realized when selection on the p scale is practiced is shown in figure 8 for a series of different conditions.

It may be seen that under some circumstances, notably where strong selec-

tion can be applied in favor of a very common character (corresponding to the case of strong selection against a rare defect), or weak selection for a rare character, selection on the all-or-none basis is exceedingly inefficient. In such cases some indication of relative genetic values among individuals exhibiting or not exhibiting the character, as the case may be, would be valuable in improving the efficiency of mass selection. An example of an attempt to improve selection efficiency by such means is that of LUSH, LAMOREUX and HAZEL (1948)[3] to utilize age at death, rather than merely the fact of death during the first production year, as an indication of genetic value for viability. Where such attempt is unsuccessful, as in the case of the authors just cited, family selection may be many times as effective as mass selection and the utility of the latter may be negligible by contrast.

The errors resulting from the use of heritability on the p scale would, in general, be of less importance in family selection than in individual selection. The larger the number of individuals per family, the less important would be the discrepancy between computed and attained gains. One reason why this is true is that the magnitude of that portion of the genetic variance of family means attributable to genetic variance within families, a portion which includes most of the epistatic variance, is only $(1/n)$ times as great as the within family genetic variance. Another reason is that environmental variance of different families does not cover as wide a range because the total genetic variance of family means is less than that of individuals.

In combined individual and family selection, therefore, extra weight should be assigned to the individual in those situations in which actual gains from mass selection are greater than those calculated on the basis of $h_p{}^2$ (see figure 6); conversely extra weight should be given to the family where expected gains from mass selection are less than calculated. Calculation of the precise allowances to be made are beyond the scope of this paper but would be useful to the extent to which the model being investigated is approximated by actual cases of economically important threshold characters.

SUMMARY

A convenient scale of measurement of the genotype of individuals with respect to all-or-none polygenic characters is that of the *probability of expression* of the character in relation to the total array of environments. On such a scale, however, the genetic variance is almost certain to be partly, and under some circumstances is largely, non-additive in nature. In addition the environmental variance, expressed on the same scale, is not independent of the genotypic level. An investigation is made of the effects of these disturbing features on estimates of gain from mass selection computed on the basis of heritability determinations.

On the basis of a simple mathematical model the non-genetic variance is shown to vary from a negligible proportion of the total genetic variance to

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[3] An excellent discussion of the principles of selection for all-or-none traits will be found in the concluding sections of the paper by LUSH, LAMOREUX and HAZEL. It is oriented on what we have called the expected rather than the actual gains in our previous section.

more than three quarters of the total for the range of circumstances considered (figure 3). Only a small proportion of the non-additive variance would usually be included in heritability estimates based on full and half sib correlations (figure 4). Estimates of gain calculated on the basis of heritability determinations would constitute good approximations of the gain that should actually be achieved where the heritability of the variate that is postulated to underlie the appearance of the character is moderately low and the character incidence is not too close to zero or unity; under other conditions the gain may be grossly over- or under-estimated (figure 6). Gains calculated on the basis of the additive heritability, for all the situations studied, are accurate where the selection pressure is very low (figure 7). Under some circumstances gains could be achieved at a much greater rate could classification of the phenotype be made on a graduated rather than on an all-or-none basis (figure 8).

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APPENDIX

PROOF THAT THE ADDITIVE HERITABILITY ON THE P SCALE IS GIVEN BY THE EXPRESSION $\bar{z}^2 h_x^2 / \bar{p} \bar{q}$.

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As indicated in the text, we may expect the additive genetic value on the p scale, (G_{pa}) of a genotype having the value G_x on the x scale to be a linear function of x. We may write, therefore:

$$G_{pa} = a + bG_x.$$

Since G_{pa} is to be a least squares estimate of the genotypic value on the p scale, G_{pt}, b is the linear regression coefficient of G_{pt} on G_x. Further the variance of G_{pa} will equal the variance of G_x multiplied by b^2, or:

$$\sigma_{G_{pa}}^2 = b^2 \sigma_{G_x}^2.$$

The linear regression coefficient, as is well known, is the covariance divided by the variance of the independent variate, so the expression above may be written simply:

$$\sigma_{G_{pa}}^2 = \frac{\text{cov}^2 (G_x G_{pt})}{\sigma_{G_x}^4} \sigma_{G_x}^2 = \left(\frac{\text{cov} (G_x G_{pt})}{\sigma_{G_x}} \right)^2. \tag{1}$$

The value G_{pt} corresponding to any value G_x may be written as follows:

$$G_{pt} = \frac{1}{\sqrt{2\pi}\, \sigma_E} \int_{t - G_x}^{\infty} e^{-x^2/2\sigma_E^2} dx \tag{2}$$

where t is the theshold value and G_x has the frequency distribution:

$$f(G_x) = \frac{1}{\sqrt{2\pi}\, \sigma_{G_x}} e^{-G_x^2/2\sigma_{G_x}^2}. \tag{3}$$

To simplify the symbols we may write:

$$G_x = x_1 \quad \text{and} \quad \sigma_{G_x} = \sigma_1.$$

The covariance may then be written as follows:

$$\text{cov} = M(x_1 G_{pt}) = \frac{\displaystyle\int_{-\infty}^{+\infty} f(x_1) x_1 G_{pt} dx_1}{\displaystyle\int_{-\infty}^{+\infty} f(x_1) dx_1} .$$

The denominator equals unity. The numerator may be integrated by parts, letting $f(x_1)x_1 dx_1 = dv$ and $G_{pt} = u$. Then:

$$v = \int \frac{x_1}{\sqrt{2\pi}\,\sigma_1} e^{-x_1{}^2/2\sigma_1{}^2} dx_1 = -\sigma_1{}^2 f(x_1)$$

and

$$du = dG_{pt}$$

and since the upper limit of expression (2) is fixed, this equals:

$$du = \frac{1}{\sqrt{2\pi}\,\sigma_E} e^{-(t-x_1)^2/2\sigma_E{}^2} dx_1.$$

Therefore:

$$\text{cov} = [uv]_{-\infty}^{+\infty} - \int_{-\infty}^{+\infty} v\,du$$

$$= -\left[G_{pt}\sigma_1{}^2 f(x_1) \right]_{-\infty}^{+\infty} - \int_{-\infty}^{+\infty} \frac{-\sigma_1{}^2 f(x_1)}{\sqrt{2\pi}\,\sigma_E} e^{-(t-x_1)^2/2\sigma_E{}^2} dx_1$$

and since the first term is zero at both limits this equals (substituting from (3))

$$\frac{\sigma_1}{2\pi\sigma_E} \int_{-\infty}^{+\infty} e^{-A/2} dx_1 \qquad (4)$$

where

$$A = \frac{x_1{}^2}{\sigma_1{}^2} + \frac{(t-x_1)^2}{\sigma_E{}^2} = \frac{x_1{}^2}{\sigma_1{}^2} + \frac{1}{\sigma_E{}^2}(t^2 - 2tx_1 + x_1{}^2)$$

$$= \frac{1}{\sigma_E{}^2}\left[\frac{\sigma_1{}^2 + \sigma_E{}^2}{\sigma_1{}^2} x_1{}^2 - 2tx_1 + t^2 \right]$$

which may be written, since $\sigma_1{}^2 + \sigma_E{}^2 = 1$

$$= \frac{1}{\sigma_E{}^2}\left[\frac{x_1}{\sigma_1} - t\sigma_1 \right]^2 + \frac{1}{\sigma_E{}^2}\left[t^2 - t^2\sigma_1{}^2 \right]$$

$$= \frac{1}{\sigma_1{}^2\sigma_E{}^2}(x_1 - t\sigma_1{}^2)^2 + t^2.$$

Substituting in (4):

$$\text{cov} = \frac{\sigma_1}{2\pi\sigma_E}\, e^{-t^2/2} \int_{-\infty}^{+\infty} e^{-(x_1 - t^2\sigma_1^2)^2/2\sigma_1^2\sigma_E^2}\, dx_1$$

$$= \frac{\sigma_1}{2\pi\sigma_E}\, e^{-t^2/2} \sqrt{2\pi}\, \sigma_1\sigma_E = \sigma_1^2\, \frac{1}{\sqrt{2\pi}}\, e^{-t^2/2} = \bar{z}\sigma_1^2.$$

Substituting in (1)

$$\sigma_{Gpa}{}^2 = \left(\frac{\bar{z}\sigma_1^2}{\sigma_1^2} \right)^2 = \bar{z}^2\sigma_1^2 = \bar{z}^2 h_x{}^2.$$

Dividing the additive genetic variance by the total genetic variance:

$$h_{pa}{}^2 = \frac{\bar{z}^2 h_x{}^2}{\bar{p}\bar{q}}.$$