Theor Appl Genet (1995) 91:544–552

S. Magnussen · A. Kremer

The beta-binomial model for estimating heritabilities of binary traits

Received: 10 May 1993 / Accepted: 17 January 1995

Abstract Calculations of individual narrow-sense heritability and family mean heritability of a binary trait in stochastically simulated sib trials in completely randomized block experiments showed that in some situations estimates of "realized" heritabilities obtained from the mixed linear threshold model could be improved by application of a proposed beta-binomial model. The proposed model adopts the beta-binomial as the conjugate-prior for the distribution of probabilities of observing the binary trait in a genetic entry. Estimation of the beta parameters allows an estimation of selection response and, by linkage to a threshold model for the individual observations, the desired heritabilities can be obtained. The average bias in the beta-binomial estimates of heritability and family mean heritability was less than 2%. Improvements over existing procedures were especially manifest at heritabilities above 0.3 and at low overall probabilities of observing the trait ($p < 0.30$). The lowest root mean square errors were consistently obtained with the algorithm proposed by Harville and Mee (1984). The beta-binomial framework, although restricted to a single random additive genetic effect, further facilitates general analysis, estimation of selection response, and calculation of reliable family mean heritability. Intraclass correlations can be estimated directly from the beta-binomial parameters.

Key words Simulation · Genetic response · Family mean heritability · Sib analysis

Introduction

Binary traits in animal and crop breeding are often important determinants of product quality and production efficiency or else are indicators of underlying factors that are difficult or costly to measure. Heritabilities of these "traits" will help gauge the potential for genetic improvement and serve as an important population genetic parameter (Wright 1978). Dempster and Lerner (1950) developed an algorithm for calculating the heritability of such binary traits, and Gianola (1979) generalized the derivations. Van Vleck (1971) used the algorithm in a simulation study of sib and parent-offspring analyses of a binary trait and found that the estimated heritabilities were very close to the "true" values as long as the binary trait was expressed in 20–80% of the population and as long as the true heritability was below 0.7. The algorithm is based on the assumption of an underlying polygenic factor with Gaussian-distributed environmental and genetic parts. When the underlying phenotypic value exceeds a certain threshold the outward phenotype is expressed; otherwise the "trait" is not expressed. The threshold value on the underlying scale is linked via the underlying probability density function to the proportion (p) of the population that expresses the trait.

Newer and more versatile methods have been developed for categorical data with more than two response levels (Quaas and Van Vleck 1980; Harville and Mee 1984; Foulley et al. 1987; Magnussen 1990) and for models with a mixture of a quantitative and a binary trait (Foulley et al. 1983; Simianer and Schaeffer 1989).

Shortcomings of the Dempster and Lerner (1950) and Gianola (1979) algorithms include the recognized upward bias of about 0.03 for estimates of individual narrow-sense heritabilities, the dependency on a Gaussian framework, and the lack of a logical linkage to an expression of the expected response to selection. Experience with the Harville and Mee procedure, which is rooted in the same threshold-model framework as the

---

Communicated by D. Van Vleck

S. Magnussen (✉)
Natural Resources Canada, Canadian Forest Service, Petawawa National Forestry Institute, Chalk River, Ontario K0J 1J0, Canada

A. Kremer
Institut national de la recherche agronomique, Laboratoire d'amélioration des arbres forestieres, Pierroton 33610 Cestas, France

Dempster and Lerner algorithm, is still limited and a formal comparison with the Dempster and Lerner algorithm has not been carried out.

Logit-based analysis (Gianola 1980; Stiratelli et al. 1984) and analysis based on intra-class correlations (Landis and Koch 1977) are possible alternatives to the Dempster and Lerner (1950) algorithm for calculating the heritability of a binary trait, although these alternatives have not yet been tested for this particular purpose.

In the present paper, the beta-binomial model is introduced as a model framework for computing heritabilities and selection response for a binary trait. The beta distribution is used as a conjugate-prior to model the probability of observing the trait in genetic entries. The beta-binomial approach makes it possible to estimate selection response, and thus 'realized' heritabilities, via a linkage to a threshold model. The performance of the beta-binomial model was tested in a series of simulated sib-trials in complete blocks and compared to the performance of a series of alternate procedures for estimating heritability of a binary trait.

In a Bayesian context, the beta distribution is the conjugate-prior to the binomial distribution (Santner and Duffy 1989) and, with the aforementioned limitations, the beta-binomial model lends itself easily to a nested or hierarchical analysis of variance (McCullagh and Nelder 1989; Searle et al. 1992).

Methodology

The data model

Under consideration is a sib analysis of an intrinsically (unobservable) mixed process under independent polygenic and environmental influences in a completely randomized block design. Assume, for now, that, in a given reference population, this process can be described by a Gaussian variable (z) with a mean of zero and a variance of one. Whenever z exceeds a certain threshold value, say z', an outwardly observable character (δ) is expressed. This character is clearly dichotomous (absence/presence); on a binary scale, the character has a value of 1 for presence and 0 for absence.

The linear model for the intrinsic variable z in the simulated sib-trials is

$$z_{ijk} = \mu + \tau_i + \varepsilon_{ijk}, \tag{1}$$

where z_{ijk} is the observation on the kth individual in family i and block j; μ, is the overall mean, τ_i the family effect attributed to family i, and ε_{ijk} the residual effects (plots, block, and 'error'). All effects except the mean are considered as random effects with a known distribution (here Gaussian) with means of zero and variances σ_f^2, and σ_ε^2, respectively.

Transformation of the intrinsic variable z to a binary trait (δ) on the outward scale was done as follows:

$$\delta_{ijk} = \begin{bmatrix} 1 \text{ for } z_{ijk} \le z' \text{ or } \Phi(z_{ijk}) \le p \\ 0 \text{ for } z_{ijk} > z' \text{ or } \Phi(z_{ijk}) > p \end{bmatrix}, \tag{2}$$

where Φ denotes the cumulative probability function of a normal distribution and p denotes the population probability of observing the dichotomous character (δ).

Data were simulated (Ripley 1987) according to the linear model in (1) for a normally distributed intrinsic variable (z) with a total variance of 1.0 in a series of half-sib trials in randomized complete blocks. Family values (τ_i) were simulated as a normal variate with mean of zero and variance of 0.025, 0.050, 0.075, 0.100, or 0.125. Environmental values (ε_{ijk}) were simulated as a single Gaussian variable with mean of zero and variance of $1-\sigma_f^2$.

Seven threshold levels (p) for triggering expression of the binary trait (δ) were superimposed on the simulated data (z_{ijk}). The chosen thresholds were for $p=(0.05, 0.10, 0.15, 0.20, 0.30, 0.40, 0.50)$ which are the probabilities of observing the binary trait in the reference population [i.e. $p=\Phi(\mu)$ or $z'=\Phi^{-1}(p)$].

Simulations were generated for experimental designs with $n_{fam}=(50, 100)$ and $n_{block}=(3, 5)$ and plot size of five observational units. Each of these 140 combinations of design-parameters (σ_f^2, p, n_{fam}, n_{block}) were simulated 1000 times.

Estimating heritabilities from the simulated trials

From each set of simulated data, alternative estimators of individual narrow-sense heritability and family mean heritability were computed and compared. Table 1 provides an overview of the estimators. Estimation procedures are given below.

Estimates of the true heritabilities

As a baseline for comparing the various heritability estimators the individual narrow-sense and family mean heritabilities were com-

Table 1 An overview of alternative heritability estimators

Narrow-sense individual heritability h^2	Family mean heritability h_f^2	Remarks
$h^2(z)$	$h_f^2(z)$	"True" heritabilities derived from simulated intrinsic values (z_{ijk})
$h^2(\text{beta})$	$h_f^2(\text{beta})$ $h_f^2(\Delta p \vert \text{beta})$	Estimates based on beta-distributed family probabilities of observing the binary trait
$h^2(\text{DL})$	n.a.	Estimator derived via the Dempster-Lerner algorithm
$h^2(\rho)$	$h_f^2(\rho)$	Estimates based on intraclass correlations (Landis and Koch)
$h^2(\text{HM})$	$h_f^2(\text{HM})$	Estimates derived via the Harville and Mee procedure
$h^2(\text{logit})$	$h_f^2(\text{logit})$	Estimates based on an analysis of logits (Gianola and Stiratelli et al.)

puted directly from the simulated intrinsic variable z. These estimates were considered to be the "true" values for each of the realized stochastic simulations of a sib-trial. Specifically the "true" individual narrow-sense heritability is

$$\widehat{h}^2(z) = \frac{4\hat{\sigma}_f^2(z)}{\hat{\sigma}_f^2(z) + \sigma_\varepsilon^2(z)}, \tag{3}$$

where the estimated variance components are derived from an analysis of variance (Henderson's Method III, Searle et al. 1992) applied to the variable z_{ijk} and the model in (1). "True" family mean heritability is

$$h_f^2(z) = \frac{\hat{\sigma}_f^2(z)}{\hat{\sigma}_f^2(z) + \sigma_\varepsilon^2(z)/n_{plot} \cdot n_{block}}. \tag{4}$$

Estimates of heritabilities via the beta-binomial model

In the beta-binomial model family-specific probabilities of observing the binary trait are assumed to be distributed as a beta distribution (Johnson and Kotz 1970; Prentice 1986; McCullagh and Nelder 1989; Santner and Duffy 1989; Searle et al. 1992). When the assumption about a beta distribution holds, the posterior distribution of family probabilities conditional on a selection outcome (Santner and Duffy 1989; Searle et al. 1992) can be computed. Changes in the distribution of family probabilities at the phenotypic and additive genetic level due to a selection can then be used to estimate the expected realized gain, and thus the realized heritability, of the trait (Falconer 1981).

The assumption of beta-distributed family effects was tested at the phenotypic level by computing the expected beta distribution of phenotypic family probabilities and comparing it with the distribution of the values obtained directly from the simulations. The parameters of the expected beta distribution were obtained from the average trait probability $\bar{p}$ in the population sample and the sample variance of the phenotypic family probabilities $\hat{\sigma}_{pf}^2(\delta)$ derived as the observed variance of family probabilities of observing the binary trait (see equation 6). Inference about "goodness of fit" was based on a two-sample Kolmogorov-Smirnov test (Siegel 1956). All simulated distributions were consistent with the assumptions ($P > 0.10$). A typical example of an observed/simulated and expected distribution is shown in Fig. 1

To derive the beta-binomial-based heritability estimates, three sets of beta parameters are needed: one for the phenotypic family probabilities, one for the family probabilities, and finally one for the additive genetic probabilities. The beta parameters can be obtained once estimates of the variances associated with each of the three types of probabilities are known. To obtain these variance components the following probabilistic model for the simulated binary data (δ_{ijk}) was used

$$p_{ijk} = p + p_i + p_{ijk}, \tag{5}$$

where p_{ijk} is the probability of observing the binary trait (δ) on the kth individual in the jth block of family i, p is the overall population probability (fixed effect), and the remaining ps are the random contributions due to the family effect (subscript i), and residual effects (subscript ijk), respectively. From this model the three [phenotypic (pf), family (f), and additive genetic (a)] variance components $\sigma_{pf}^2(\delta)$, $\sigma_f^2(\delta)$, and $\sigma_a^2(\delta)$ were obtained via a one-way analysis of variance performed on the binary data (δ_{ijk}).

Phenotypic family probabilities [$p_{(pf)i}$] are assumed to follow a beta distribution:

$$p_{(pf)i} = \sum_{jk} \frac{P_{ijk}}{n_{i..}} \text{ with } p_{(pf)i} \sim Beta\,(\alpha_{pf}, \beta_{pf}), \tag{6}$$

where $n_{i..}$ is the number of observations in family i. Family probabilities [$p_{(f)i}$] are defined as the sum of the overall mean (p) and an additive family effect:

$$p_{(f)i} = p + p_i \text{ with } p_{(f)i} \sim Beta\,(\alpha_f, \beta_f). \tag{7}$$

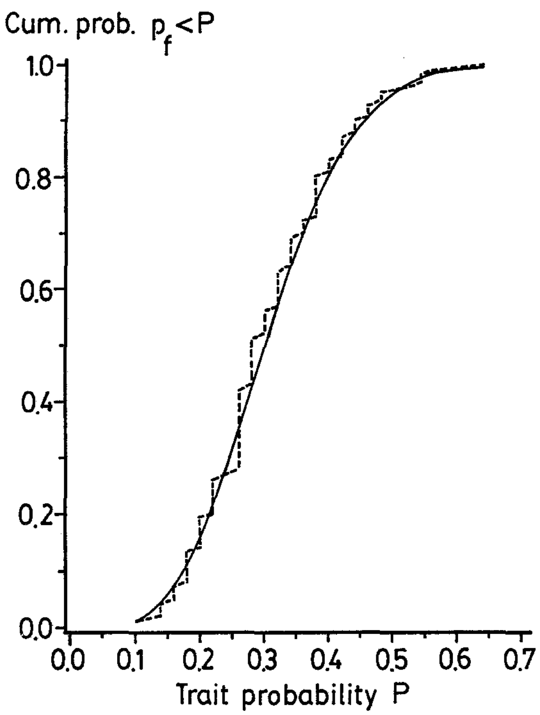


Fig. 1 Simulated ("observed") cumulative probability distribution of family mean probabilities of observing the binary trait (*broken line*) versus the expected beta distribution (*solid line*). $n_{fam} = 100$, $n_{block} = 5$, $n_{plot} = 5$ obsevational units, "true" heritability $h^2(z) = 0.2$, $\bar{p} = 0.3$

Assuming that the families consist of half-sibs the following conceptual model is used for the additive genetic family probabilities [$p_{(a)i}$]:

$$p_{(a)i} = p + 0.5p_i \text{ with } p_{(a)i} \sim Beta\,(\alpha_a, \beta_a). \tag{8}$$

Sample estimates of the above probabilities were obtained from the simulated data as

$$\hat{p}_{(pf)i} = \sum_{jk} \delta_{ijk}/n_{i..} = \hat{p}_{(f)i}$$

$$\bar{p} = \sum_i \hat{p}_{(pf)i}/n_{fam}$$

$$\hat{p}_i = \hat{p}_{(pf)i} - \bar{p}. \tag{9}$$

Estimates of the three sets of beta parameters were obtained from the following standard formulae (Johnson and Kotz 1970, p. 44, equations 19 and 20):

$$\hat{\alpha}_t = \bar{p} \times (1 - \bar{p})/\hat{\sigma}_t^2(\delta) \times \hat{\omega} - \bar{p}$$

$$\hat{\beta}_t = (1 - \bar{p})/\bar{p} \times \hat{\alpha}_t, \tag{10}$$

where the subscript t denotes the type of parameter [$t = f$ (family), pf (phenotypic family mean), or a (additive genetic)] and $\hat{\sigma}_t^2(\delta)$ is the corresponding variance component estimated from the analysis of variance of the simulated binary trait (δ) applied to the model in (5) (Henderson's Method III, Searle et al. 1992). The ω in (10) is a weighting (shrinkage) factor that adjusts for the proportionality between family mean probabilities and their variances [the binomial variance $p_i(1 - p_i)$]. To be specific, ω is the square of the ratio of the variance of the unweighted sample probability ($\bar{p}$) to the variance of a weighted sample mean (p'). The weights used to construct p' were the inverses of the standard deviation of a family mean probability.

Equation (11) provides the computational details

$$\hat{\omega} = \left[\frac{\bar{p} \times (1-\bar{p})}{\bar{p}' \times (1-\bar{p}')}\right]^2 \text{ and}$$

$$p' = \sum_i \hat{p}_{(pf)i} \cdot [\hat{p}_{(pf)i} \cdot (1-\hat{p}_{(pf)i})]^{-0.5} \cdot \sum_i [\hat{p}_{(pf)i} \cdot (1-\hat{p}_{(pf)i})]^{0.5}, \qquad (11)$$

where the last multiplier in the expression for p' is the sum of the applied weights.

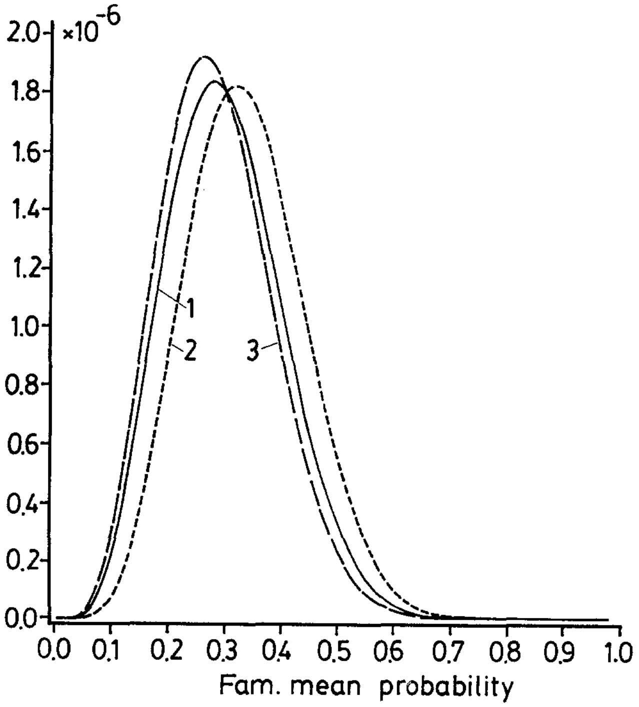
Note, in presence of significant block effects this formulae for computing the beta parameters should be applied to the within-block estimates of the population mean and family mean probabilities and subsequently averaged across blocks. In this study, block effects were zero, and given the balanced nature of the data the across-block estimates were used as outlined.

With estimates of the beta distribution parameters for the phenotypic (*pf*), family (*f*), and additive genetic (*a*) mean probabilities, estimates of the posterior mean after a selection can be obtained (see equation 119, p. 345 in Searle et al. 1992). For example, after selecting either all progenies expressing the binary trait ($\delta = 1$) or all progenies without the trait ($\delta = 0$) the conditional mean family probability in the selected population is expected to be

$$\bar{p}_{t|\delta} = \frac{\delta + \hat{\alpha}_t}{1 + \hat{\alpha}_t + \hat{\beta}_t}, \qquad (12)$$

where $t = (f, pf, a)$ denotes the effect under consideration. The shift in mean due to this selection (i.e. $\bar{p}_{t|\delta} - \bar{p}$) is considered to be a response to selection from which the expected realized heritability of the trait under selection (Falconer 1981) can be estimated. Figure 2 illustrates the impact of selection of all progenies with the binary trait on the distribution of family mean probabilities in one of the simulated trials.

Fig. 2 Expected beta probability density distributions of family mean probabilities. $n_{fam} = 100$, $n_{block} = 5$, $n_{plot} = 5$ obervational units, "true" heritability $h^2(z) = 0.2$. 1 Unconditional distribution (no selection), $\bar{p} = 0.30$; 2 Conditional on selecting all progenies with $\delta = 1$, $\bar{p}_{f|\delta=1} = 0.34$; and 3 Conditional on selecting all progenies with $\delta = 0$, $\bar{p}_{f|\delta=1} = 0.29$



Before estimating the realized heritability for this type of selection, the selection intensity (*i*) and the expected response in the underlying intrinsic character *z* must be estimated. The selection response ($\bar{p}_{t|\delta} - \bar{p}$) on the binary scale can be transformed into the expected response on the underlying continuous variable (*z*) via the inverse of its cumulative distribution function (i.e., Φ^{-1} for the normal case). An estimate of the implicit selection intensity is easily obtained from the threshold of the intrinsic character [($z' = \Phi^{-1}(\bar{p})$ for the Gaussian case, see 2] and the standard method for calculating the mean of the selected population on a standardized (mean zero and variance one) scale (Falconer 1981). For the standardized normal case *i* is calculated as

$$\hat{i} = \frac{1}{\sqrt{2\pi} \times \bar{p}} \times exp\left(-\frac{z'^2}{2}\right). \qquad (13)$$

Given the above estimates of selection response and selection intensity the beta-binomial estimate of the "realized" individual narrow-sense heritability was computed as

$$\widehat{h^2}(beta) = [\Phi^{-1}(\bar{p}_{a|\delta}) - \Phi^{-1}(\bar{p})]/\hat{i}. \qquad (14)$$

The beta distribution parameters were also used to compute the family mean heritability by taking the ratio of the additive family variance to that of the phenotypic family variance:

$$\widehat{h^2_f}(beta) = \frac{\hat{\alpha}_f \times \hat{\beta}_f \times (\hat{\alpha}_{pf} + \hat{\beta}_{pf})^2 \times (\hat{\alpha}_{pf} + \hat{\beta}_{pf} + 1)}{\hat{\alpha}_{pf} \times \hat{\beta}_{pf} \times (\hat{\alpha}_f + \hat{\beta}_f)^2 \times (\hat{\alpha}_f + \hat{\beta}_f + 1)}. \qquad (15)$$

Equation (15) uses the formulae for the variance of a beta-distributed variable (here family effect and phenotypic family values, Johnson and Kotz 1970, p. 40).

An alternative formulation of the family mean heritability rooted in the beta-binomial model is derived from the expected ratio of the "realized" selection response in family mean probability ($\bar{p}_{f|\delta} - \bar{p}$) to the selection response at the phenotypic family mean level ($\bar{p}_{pf|\delta} - \bar{p}$). Transforming this ratio of responses to the scale of the intrinsic variable *z* via the inverse to its cumulative distribution function yields an estimate of the realized family mean heritability:

$$h^2_f(\Delta p|beta) = \frac{\Phi^{-1}(\bar{p}_{f|\delta=1}) - \Phi^{-1}(\bar{p})}{\Phi^{-1}(\bar{p}_{pf|\delta=1}) - \Phi^{-1}(\bar{p})}. \qquad (16)$$

Estimates of heritability via the Dempster and Lerner algorithm

For a binary trait that is the observable expression of an underlying normally distributed (mean 0 and variance of one) intrinsic character (here *z*), Dempster and Lerner (1950) suggested the following estimator of the individual narrow-sense heritability which Gianola (1979) showed to be a special case of the more general solution

$$\widehat{h^2}(DL) = 4\hat{\cdot}\sigma^2_f(\delta) \times [\phi(z')]^{-2} \qquad (17)$$

where ϕ denotes the Gaussian probability density function evaluated at the threshold z' for expression at the binary scale [$z' = \Phi^{-1}(p)$], and $\sigma^2_{fam}(\delta)$ is the estimate of the family variance component obtained from a conventional analysis of variance (Henderson's method III, Searle et al. 1992) of the binary trait applied to the model in (5).

Family mean heritabilities could not be derived using the Dempster and Lerner algorithm because it does not include estimation of unbiased variance components for the intrinsic character (*z*).

Estimates of heritabilities via intraclass correlations

Landis and Koch (1977) described a method for computing variance components, and thus heritabilities, of binary traits based on an estimation of the intraclass correlation (ρ). Given estimates of the two beta parameters α_{pf} and β_{pf} (cf. above) an estimate of ρ is found as (Prentice 1986; Searle et al. 1992):

$$\hat{\rho} = \frac{1}{(\hat{\alpha}_{pf} + \hat{\beta}_{pf} + 1)}. \tag{18}$$

From the estimate of ρ and following the procedures outlined by Landis and Koch (1977) the following heritabilities (individual narrow-sense and family mean heritability) were computed

$$\hat{h}^2(\hat{\rho}) = 4 \times \hat{\rho} \tag{19}$$

$$\hat{h}^2_f(\hat{\rho}) = \frac{\hat{\rho}}{\hat{\rho} + (1 - \hat{\rho})/(n_{block} \cdot n_{plot})}. \tag{20}$$

Estimates of heritabilities via the Harville and Mee algorithm

Harville and Mee (1984) developed an iterative algorithm for estimating random effects and the additive genetic variance for ordered categorical data. The algorithm is based directly on the model for the intrinsic character and its assumed distribution. Estimation proceeds as iterative generalized least squares for a mixed model applied to maximum-likelihood estimators of the effects on the intrinsic scale (see also Henderson 1984). Full details of the method is given in their publication. For the simulated trials in this study the Harville and Mee algorithm applies directly to the model in (1). Estimation is thus restricted to the overall mean (μ a fixed effect) and the family effects τ_i and the ratio γ of the family variance $[\sigma^2_{fam}(z)]$ to the residual non-family variance $[1 - \sigma^2_{fam}(z)]$. Once γ is estimated the heritabilities can be obtained by straighforward calculus. For this simple model, and given the balanced nature of the simulated binary data (δ_{ijk}), the main steps in the Harville and Mee procedure concerned with the estimation of μ, τ, and γ are relatively simple (Harville and Mee 1984). Iterations were stopped when two successive heritability estimates $[h^2(HM)]$ differed by less than 0.01. Less than 50 iterations (average 36) sufficed to satisfy this criterion in all simulated scenarios. From the final estimates the family heritability was obtained as:

$$\hat{h}^2_f(HM) = \frac{\hat{\sigma}^2_a}{\hat{\sigma}^2_a + 1/(n_{block} \cdot n_{plot})}. \tag{21}$$

Heritability estimates based on logits

Gianola (1980) and others (for example, Foulley et al. 1983, 1987) proposed to evaluate the genetic merit for a binary trait in sires by a logit model. In this model the logits of plot probabilities are assumed to follow a mixed linear model

$$\log\left(\frac{p_{ij}}{1 - p_{ij}}\right) = \mu + \tau_i + \xi_{ij}, \tag{22}$$

where p_{ij} is the probability of observing the binary trait in plot ij ($p_{ij} = \Sigma_k \delta_{ijk}/n_{ij}$) and the parameters on the right side are the grand mean (fixed effect), the family effects (random), and the plot effect, respectively. Application of the Harville and Mee procedure for estimation of the family variance component and heritability of the logits in model (22) is feasible after simple substitutions of probability and cumulative probability functions (see also Stiratelli et al. 1984; Foulley et al. 1987; Breslow and Clayton 1993) and transformations to logits. Instead of the Gaussian cdf. (ϕ) and pdf. (Φ) the logit cdf. $\phi_{\text{logit}}(\bullet) = \exp(\bullet)/[1 + \exp(\bullet)]^2$ and $\Phi_{\text{logit}}(\bullet) = 1 - 1/[1 + \exp(\bullet)]$ were used, respectively, where $\bullet$ denotes the logit of family/plot/experimental probability. Starting values of the grand mean and family effects were

$$\hat{\mu}^0 = \log\left(\frac{\sum_{ijk} \delta_{ijk} + c}{n_{...} - \sum_{ijk} \delta_{ijk} + c}\right) \tag{23}$$

and

$$\hat{\tau}^0 + \hat{\mu}^0 = \log\left(\frac{\sum_{jk} \delta_{ijk} + c}{n_{i...} - \sum_{jk} \delta_{ijk} + c}\right), \tag{24}$$

where c is a correction needed to ensure solutions for extreme family probabilities (zero and one). A c value of $0.75/n$, where n is the appropriate sample size, was chosen as the overall "best" value (least amount of bias).

It is well known that the antilog of the mean of logarithmic-transformed variables differs from the arithmetic mean (McCullagh and Nelder 1989; Breslow and Clayton 1993) by an amount equal to one-half of the variance of the log-transformed variable. This attenuation (bias) will inflate the family variance and thus the heritability estimate. Breslow and Clayton (1993) suggest a heuristic remedy to counter the bias in estimated random effects. Inspired by their approach, the following final adjustment (explained below) to the log-transformed version of the Harville-Mee estimator was introduced

$$\hat{h}^2(\text{log}it) = \frac{4\hat{\gamma}_{\text{log}it}}{(1 + \hat{\gamma}_{\text{log}it})} \times \left\{ \frac{\frac{\pi^2}{3}}{\left[\left(\hat{\mu}^0_{\text{log}it}\right)^2 \left(1 - \frac{2\hat{\gamma}_{\text{log}it}}{1 + \hat{\gamma}_{\text{log}it}}\right) + \frac{\pi^2}{3}\right]} \right\}, \tag{25}$$

where $\hat{\gamma}_{\text{log}it}$ corresponds to the logit-based version of γ in the Harville-Mee algorithm.

A logit-transformed normal variable with a mean of zero and a variance of one would have a variance of $\pi^2/3$ (see for example, McCullagh and Nelder 1989); a shift in the mean of the random vairable away from zero would attenuate the variance of the logit-transformed variable by an amount proportional to the square of the average logit. The adjustment factor is then the ratio of the expected variance of a logit-transformed standard normal variable to the inflated variance due to a shift in the mean. Thus the final form of the adjustment takes into consideration that the attenuation is half the variance of the random effects, and also that the attenuation bias is in the non-genetic fraction of the total variance. Family mean heritability of logits was estimated as outlined for the Harville and Mee procedure.

Results

The goal was to see how well the various estimates of heritability approximate the "true" heritability estimates, i.e., the minimum-variance unbiased estimates derived directly from the simulated intrinsic variable (z). A comparison of estimates of genetic merit was beyond the scope of this study. The comparisons were made only across the various levels of the "main effects" (i.e., the five heritability levels, the seven probability levels, and the four sample sizes). More detailed analyses of contrasts and ratios of heritability estimates performed for each combination of heritability, probability, and sample size did not reveal any trends not already captured by these "main effects".

The proposed beta-binomial algorithm for calculating "realized" individual narrow-sense heritability, either directly $[h^2(\text{beta})]$ or via the intraclass corelation, $[h^2(\rho)]$ tracked, on average, the "true" heritabilities closer than any of the alternatives (Table 2). With an average overall discrenpancy between the beta estimates

Table 2 Average estimates of individual narrow-sense heritabilities (h^2) and family mean heritability for various values of given h^2. In parentheses are the average standard deviations of the estimates

Estimate	$h^2 = 0.1$	$h^2 = 0.2$	$h^2 = 0.3$	$h^2 = 0.4$	$h^2 = 0.5$	Mean
$h^2(z)$[a]	0.111 (0.046)	0.204 (0.062)	0.302 (0.076)	0.400 (0.090)	0.498 (0.101)	0.303 (0.075)
h^2(beta)[b]	0.138 (0.069)	0.217 (0.087)	0.304 (0.100)	0.390 (0.115)	0.477 (0.127)	0.305 0.100)
h^2(DL)[c]	0.143 (0.083)	0.226 (0.107)	0.322 (0.130)	0.425 (0.153)	0.539 (0.173)	0.331 (0.129)
h^2(HM)[d]	0.113 (0.042)	0.188 (0.061)	0.278 (0.081)	0.370 (0.091)	0.461 (0.103)	0.282 (0.072)
$h^2(\rho)$[e]	0.125 (0.031)	0.217 (0.062)	0.307 (0.051)	0.400 (0.061)	0.495 (0.069)	0.309 (0.051)
h^2(logit)[f]	0.180 (0.036)	0.238 (0.049)	0.322 (0.059)	0.400 (0.072)	0.472 (0.078)	0.322 (0.058)
$h_f^2(z)$[g]	0.428 (0.105)	0.577 (0.085)	0.668 (0.068)	0.729 (0.057)	0.773 (0.048)	0.635 (0.073)
h_f^2(beta)[h]	0.260 (0.031)	0.479 (0.031)	0.651 (0.031)	0.758 (0.032)	0.859 (0.032)	0.601 (0.031)
$h_f^2(\Delta \text{p}\vert\text{beta})$[i]	0.404 (0.125)	0.561 (0.119)	0.670 (0.108)	0.758 (0.099)	0.828 (0.089)	0.644 (0.108)
h_f^2(HM)[j]	0.516 (0.096)	0.645 (0.082)	0.735 (0.064)	0.792 (0.049)	0.830 (0.041)	0.704 (0.066)
$h_f^2(\rho)$[k]	0.303 (0.073)	0.496 (0.064)	0.621 (0.058)	0.715 (0.053)	0.785 (0.048)	0.584 (0.059)
h_f^2(logit)[l]	0.694 (0.073)	0.677 (0.081)	0.702 (0.074)	0.727 (0.076)	0.743 (0.069)	0.709 (0.075)
Corr1[m]	0.488	0.604	0.657	0.690	0.701	0.628
Corr2[n]	0.502	0.608	0.657	0.683	0.691	0.628

[a] "True" estimate of narrow-sense heritability as per simulated intrinsic variable z (see 3)

[b] Beta-binomial estimate of narrow-sense heritability (see 14)

[c] The Dempster-Lerner estimate of narrow-sense heritability (see 17)

[d] The Harville-Mee-derived estimate of narrow-sense heritability

[e] The narrow-sense heritability estimate derived via the intraclass correlation (see 19)

[f] Logit-based estimate of narrow-sense heritability (see 25)

[g] "True" estimate of family mean heritability as per simulated intrinsic variable z (see 4)

[h] Beta-binomial estimate of family mean heritability (see 15)

[i] Beta-binomial estimate of family mean heritability (see 16)

[j] The Harville-Mee-based estimate of family mean heritability (see 21)

[k] The estimate of family mean heritability derived via an estimate of the intraclass correlation (see 20)

[l] Logit-based estimate of family mean heritability

[m] Average correlation between the beta estimate h^2(beta) and "true" heritability $h^2(z)$

[n] Average corelation between the Dempster-Lerner estimate h^2(DL) and "true" heritability $h^2(z)$

of individual narrow-sense heritability and the "true" values of only 0.002, the improvement over the next-best alternative was statically significant (t-test, $t > 3.00$, $P < 0.01$). The superiority of the new algorithm was most pronounced for intermediate levels of heritability (0.3–0.5) and low probabilities ($p < 0.3$). When applied in these heritability and probability ranges the beta-binomial approach reduced the bias by approximately five percentage points when compared to the best of the alternatives. The standard deviations of the individual narrow-sense heritabilities derived from the beta-model framework were also comparable to the standard deviations of the "true" heritabilities. However, heritability estimates from the Harville-Mee-based method had standard deviations almost identical (within 10%) to the true values.

Comparison of family mean heritabilities across the levels of individual narrow-sense heritabilities gave a more muted picture with no single estimation procedure emerging as truly superior (Table 2). Most promising were the estimates based on the ratio of selection responses at the family level to the response at the phenotypic family level [$h_f^2(\Delta \text{p}|\text{beta})$]; although the estimator gave results too high for $h^2 \geq 0.3$ and too low for $h^2 \leq 0.2$. The average bias encountered with this estimator across levels of individual narrow-sense heritabilities was about 5%. Estimates derived from the intraclass correlations [$h_f^2(\rho)$] were quite close (within 4%) to the "true" values when h^2 was above 0.2, but a large bias (> 15%) was manifest for lower heritabilities. The Harville and Mee procedure produced the best estimates for low probabilities ($p < 0.2$) but other estimates were less attractive than the best beta-binomial-based estimates. Logit-based estimates of the family mean heritabilities [h_f^2(logit)] were generally among the poorest.

Single estimates of the individual narrow-sense heritabilities (1000 for each combination of heritability and probability) derived from the beta-binomial model were significantly ($P < 0.001$) correlated with the "true" estimates (Corrl in Table 2). The average correlation between the "true" heritabilities and the beta-binomial estimates was 0.63 ($P < 0.001$). As a typical example of the correlation pattern found with alternative estimators, Table 2 gives the average correlations between the Dempster-Lerner estimates [h^2(DL)] and the "true" estimates. Harville-Mee-based estimates were, as a rule, more strongly correlated with the "true" values (average correlation of 0.82).

A comparison of heritability estimates across levels of the probability of trait expression on the binary scale is provided in Table 3. Estimates based on either the beta-binomial model or the Dempster-Lerner algorithm were overall better (less bias) than any of the estimators despite some obvious shortcomings of the latter in cases with low trait probability ($\bar{p} \leq 0.2$). Beta-binomial estimates were superrior for $\bar{p} < 0.30$, but slightly worse than the Dempster-Lerner estimates for $\bar{p} \geq 0.30$ (differences were not statistically significant; t-test: $t < 1.8$, $P > 0.07$). In the low probability range of trait expression, the Dempster-Lerner algorithm yields some of the most biased estimates. As a rule the bias increased in most estimators as the probability of the trait expression decreased. For $\bar{p} \geq 0.30$ the superior Dempster-Lerner-based estimates were within a few percentage points (1–3%) of the expected values. Beta- and Dempster-Lerner-based estimators tended to have a higher standard deviation than observed in either the "true" estimates or in any of the alternatives.

Table 3 Average estimates of individual narrow-sense heritability (h^2) and family mean heritability (h_f^2) for various numbers of families and numbers of replications (blocks). In parentheses are the average standard deviations of the estimates (symbols and legends as in Table 2)

Estimator	$n_{fam} = 100$ $n_{block} = 5$	$n_{fam} = 100$ $n_{block} = 3$	$n_{fam} = 50$ $n_{block} = 5$	$n_{fam} = 50$ $n_{block} = 3$
$h^2(z)$	0.300 (0.049)	0.303 (0.076)	0.299 (0.070)	0.309 (0.105)
h^2(DL)	0.313 (0.075)	0.332 (0.144)	0.313 (0.106)	0.359 (0.192)
h^2(beta)	0.296 (0.061)	0.306 (0.108)	0.295 (0.087)	0.323 (0.143)
$h^2(\rho)$	0.237 (0.032)	0.397 (0.054)	0.239 (0.062)	0.397 (0.054)
h^2(HM)	0.269 (0.074)	0.252 (0.078)	0.279 (0.106)	0.262 (0.119)
h^2(logit)	0.342 (0.059)	0.323 (0.077)	0.306 (0.081)	0.314 (0.112)
$h_f^2(z)$	0.758 (0.039)	0.513 (0.082)	0.753 (0.056)	0.515 (0.111)
h_f^2(beta)	0.872 (0.030)	0.397 (0.030)	0.798 (0.032)	0.339 (0.033)
h_f^2(Δp|beta)	0.800 (0.072)	0.500 (0.113)	0.790 (0.103)	0.502 (0.143)
$h_f^2(\rho)$	0.567 (0.043)	0.634 (0.051)	0.564 (0.062)	0.634 (0.051)
h_f^2(HM)	0.592 (0.074)	0.463 (0.080)	0.585 (0.092)	0.465 (0.110)
h_f^2(logit)	0.775 (0.032)	0.445 (0.032)	0.739 (0.032)	0.431 (0.050)

For probability levels above 0.10, the beta-binomial-derived family mean heritabilities [h_f^2(beta), h_f^2(Δp|beta) and $h_f^2(\rho)$] tracked the "true" values closer than any of the alternative estimators. For low probabilities, the Harville-Mee estimators were better.

A comparison of heritabilities across the four experimental combinations of number of families and number of blocks (replicates) revealed that the beta-binomial algorithm for computing individual narrow-sense heritabilities [h^2(beta)] produced estimates less sensitive to the size of the experiment than found with the alternatives. This method of estimation for the particular model tested here had a bias of less than 5% in any of the four settings. Family mean heritabilities estimated via h_f^2(Δp|beta) were consistently better across the four designs whereas the estimates based on the intraclass correlations were among the poorest.

So far the average bias in the various estimation procedure has been described. Of interest also is a comparison of the accuracy or average root mean square error associated with the various heritability estimates. Table 4 gives a summary of some of the more pertinent relative root mean square errors.

The relative root mean square errors (RMSE) listed in Table 4 indicate a general superiority of the Harville-Mee approach for estimation of narrow-sense heritabilities. This method reduced the average error by ten percentage points over the beta approach. Low heritabilities and low trait probabilities produced the highest RMSEs. A stronger correlation with the true heritabilities and/or lower variances were the main reasons for the lower RMSEs of the Harville-Mee estimates.

Discussion and conclusions

The beta-binomial framework for modelling the distribution of effect-associated (i.e., family) probabilities of trait expression provides a suitable and realistic model for a stochastic binary process with an overdispersion that lends itself to a wide array of analyses (McCullagh and Nelder 1989; Santner and Duffy 1989; Searle et al. 1992). For a binary trait with a simple model (a single random effect of interest, a fixed effect, and a random residual) selection responses ("realized") are also more readily obtained in the beta-binomial framework than

Table 4 Relative root mean square error (RMSE%) of selected estimates. RMSE% = $[E(\text{estimate-“true” value})^2]^{0.5}/\text{“true” value} \times 100^a$ Symbols and legends as in Table 2

Estimate	h^2(beta)	h^2(DL)	h^2(HM)	$h^2(\rho)$	h_f^2(beta)	$h_f^2(\Delta p\|\text{beta})$	$h_f^2(\rho)$	h_f^2(HM)
$h^2 = 0.1$	67	80	45	107	54	44	71	36
$h^2 = 0.2$	43	54	36	68	35	36	38	24
$h^2 = 0.3$	33	44	31	53	30	31	27	20
$h^2 = 0.4$	29	39	25	46	26	26	21	14
$h^2 = 0.5$	27	35	21	42	24	23	17	13
$\bar{p} = 0.05$	50	89	50	95	46	49	52	24
$\bar{p} = 0.10$	44	60	33	66	37	39	34	19
$\bar{p} = 0.15$	41	49	28	58	37	34	32	17
$\bar{p} = 0.20$	39	43	27	54	32	30	32	14
$\bar{p} = 0.30$	37	38	23	53	28	25	32	15
$\bar{p} = 0.40$	35	36	24	54	28	24	32	21
$\bar{p} = 0.50$	34	36	28	62	24	23	33	38
Grand mean	40	50	30	63	33	32	35	21

[a] E(•) denotes the expected value of •

from more complex analyses of mixed linear threshold models (Breslow and Clayton 1993; Harville and Mee 1984; Stiratelli et al. 1984; Foulley et al. 1983, 1987). Heritability estimates (“realized”) obtained with the beta-binomial framework for a single binary trait were often superior to those computed with the most common alternatives (Van Vleck 1971; Gianola 1979; Harville and Mee 1984; Stiratelli et al. 1984; Foulley et al. 1987) especially in the domain of intermediate heritabilities ($0.3 \leq h^2 \leq 0.5$) and low trait threshold probabilities ($\bar{p} < 0.3$), where the alternatives tended to overestimate true heritability. Furthermore, reliable estimates of the standard deviation of heritabilities and family mean heritability obtained via the beta-binomial model strengthen the arguments for this framework for the analysis of binary data in a simple model of one random effect nested within a fixed effect. When minimum root mean square error is the leading criterion for model selection the Harville and Mee estimators emerged as clearly superior.

Analyses of polychotomous data, unbalanced categorical data, and models with several fixed and random effects, become intractable with the beta-binomial framework. Analytical procedures for these cases have been developed by others (Van Vleck 1971; Gianola 1979; Harville and Mee 1984; Stiratelli et al. 1984; Foulley et al. 1987). Despite the greater versatility and formally more correct theoretical underpinnings of the mixed linear threshold models these computational, rather complex, models have deficiencies when applied to a single binary trait in a simple mixed model of one random effect and a single fixed effect. The shortcomings are all rooted in the non-linear transformation of trait values and the variances that occur between the presumed intrinsic variable and the observable categorical expression. Ingenious weighting schemes and empirical correction factors (see Breslow and Clayton 1993 for a discussion) are, for these reasons, common in analyses of categorical data.

When a continuous quantitative covariate is measured along with the binary trait it is possible to use the correlation between the traits, in conjunction with a prior probability function for the traits, to obtain unbiased estimates of variance components and thus heritabilities (Fouelley et al. 1983; Simianer and Schaeffer 1989). Computations are intensive, and the degree of bias will depend on the reliability of the prior information.

Data imbalance, and the effects of plot size and incomplete blocks, were not assessed in this study. Provided that unbiased variance components can be estimated from the entertained linear models (Searle et al. 1992) the beta-binomial approach should work equally well with data that are unbalanced (designed or accidental). Testing how well the fitted beta distribution of phenotypic family mean probabilities matches the observed values would be a necessary verification process. Plot sizes will affect precision in well known ways (confirmed in a preliminary pilot project), but quantification of this effect was deemed outside the scope of this paper. Large sample sizes are clearly needed when the binary trait is rare. The root mean square errors confirmed that precise heritability estimates are notoriously difficult to obtain (Hallauer and Miranda 1981), which stresses the importance of choosing the ‘best’ estimation procedure to avoid basically meaningless results.

Although the intrinsic (unobservable) process variable (z) was assumed to be normally distributed in this study, other distributional models could have been chosen without changing the (beta) distributions of family, phenotypic family mean, and additive genetic effects. Only the link function (Φ) used to translate the selection response from probabilities to the underlying process variable would have to be changed. However, given the presumed polygenic character of this underlying intrinsic process, justification of a non-normal distribution may be difficult (Falconer 1981). Note that the normal distribution was a necessary assumption for the heritability algorithms derived by, among others, Gianola (1979).

Predicting an individual’s genetic merit for a binary trait was beyond the scope of this paper. In a beta-binomial context the estimates would consist of conditional probabilities (conditioned on the outcome of the binary trait) of the *posterior* beta-binomial for each family in the trial. Searle et al. 1992 provides estimation procedures for the individual family parameters (pp. 344–345). However, research is still needed to find a uniformly 'best' estimation procedure. Harville and Mee (1984), Quaas and Van Vleck (1980) and Foulley et al. (1987) provide alternative estimation procedures of genetic merit based on either standardized threshold models, a multivariate (conjugate) approach, or a Poisson-binomial prior.

Acknowledgements Valuable assistance with the implementation of the Harville-Mee algorithm was kindly provided by Dr. S. Kachman, University of Nebraska. Numerous helpful suggestions and questions from the review panel are greatly appreciated and have modified earlier versions of this manuscript in substantial ways.

References

Breslow NE, Clayton DG (1993) Approximate inference in generalized linear mixed models. J Am Stat Assoc 88:9–25

Dempster ER, Lerner IM (1950) Heritability of threshold characters. Genetics 35:212–236

Falconer DS (1981) Introduction to quantitative genetics, 2nd edn. Longman, London

Foulley JL, Gianola D, Thompson R (1983) Prediction of genetic merit from data on binary and quantitative variates with an application to calving difficulty, birth weight, and pelvic opening. Genet Sel Evol 15:401–424

Foulley JL, Gianola D, Im S (1987) Genetic evaluation of traits distributed as Poisson-binomial with reference to reproductive characters. Theor Appl Genet 73:870–877

Gianola D (1979) Heritability of polychotomous characters. Genetics 93:1051–1055

Gianola D (1980) Genetic evaluation of animals for traits with categorical responses. J Anim Sci 51:1272–1276

Hallauer AR, Miranda JB (1981) Quantitative genetics in maize breeding. Iowa State University Press

Harville DA, Mee RW (1984) A mixed-model procedure for analyzing ordered categorical data. Biometrics 40:393–408

Henderson CR (1984) Applications of linear models in animal breeding. University of Guelph, Canada

Johnson NL, Kotz S (1970) Continuous univariate distributions, 2. John Wiley and Sons, New York

Landis JR, Koch GG (1977) A one-way components of variance model for categorical data. Biometrics 33:671–679

Magnussen S (1990) Alternative analyses of parent-offspring correlations for stem straightness in jack pine. Silvae Genet 3:237–244

McCullagh P, Nelder A (1989) Generalized linear models. Chapman and Hall, London

Prentice RL (1986) Binary regression using an extended beta-binomial distribution, with discussion of correlation induced by covariate measurement errors. J Am Stat Assoc 81:321–327

Quaas RL, Van Vleck LD (1980) Categorical trait sire evaluation by best linear unbiased prediction of future progeny category frequencies. Biometrics 36:117–122

Ripley BD (1987) Stochastic simulation. John Wiley and Sons, New York

Santner TJ, Duffy DE (1989) The statistical analysis of discrete data. Springer-Verlag, New York

Searle SR (1982) Matrix algebra useful for statistics. John Wiley and Sons, New York

Searle SR, Casella G, McCulloch CE (1992) Variance components. John Wiley and Sons, New York

Siegel S (1956) Non-parametric statistics for the behavioral sciences. McGraw-Hill, New York

Simianer H, Schaeffer LR (1989) Estimation of covariance components between one continuous and one binary trait. Genet Sel Evol 21:303–315

Stiratelli R, Laird N, Ware JH (1984) Random-effects models for serial observations with binary response. Biometrics 40:961–971

Van Vleck LD (1971) Estimation of heritability of threshold characters. J Dairy Sci 55:218–222

Wright S (1978) Evolution and the genetics of populations. The University of Chicago Press, Chicago