Comparison of Methods for Prediction of Genetic Gain from Mass Selection on Binary Threshold Traits[1])

By U. V. LOPES[2]), D. A. HUBER[3]) and T. L. WHITE[3])

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Abstract

Techniques of genetic gain prediction are compared through computer simulation in terms of their ability to predict genetic gain close to that realized in a cycle of selection on a binary threshold trait. Genetic gain was predicted by: a) using heritability from variance components estimated directly from 0/1 data (ANOVA method); b) using DEMPSTER and LERNER's method (DL method); c) using an approach similar to that used with DEMPSTER and LERNER's method, but with the true underlying heritability (PT method); and d) using heritability obtained by generalized linear mixed models as described by WOLFINGER and O'CONNELL (WO method) and by SCHALL (SCHALL method). For traits of low heritability ($h^2 < 0.3$) all methods predicted gain close to that realized by selection across all incidences (5% to 95%); except the WO method that overestimated the gain at incidences of the undesirable phenotype above 75%. For traits of moderate to high heritability and at high incidences of the undesirable phenotype, DL and WO methods overestimated, while ANOVA and SCHALL methods underestimated the realized gain. When the goal is prediction of gain on the binary observed scale, there is no extra benefit in using more complex methods than the ANOVA method when the true, underlying heritability is smaller than 0.3. Realized heritability on the binary scale is not symmetric around 50% incidence. Therefore, studies with binary data should cover mean incidence levels across the whole range (0% to 100%).

Key words: binary data, threshold model, heritability, realized gain.

Introduction

Although most traits in breeding are measured on a continuous scale, some important traits are restricted to two classes, the so-called binary traits (e.g., dead/alive, infected/uninfected). Binary phenotypes may have a discontinuous genetic background involving one or a few major genes, or a continuous genetic background involving many minor genes. The analysis of binary traits of the first type is primarily based on Mendelian genetics. The analysis of the second type of trait, called all-or-none or binary threshold traits, is much more complex since quantitative genetics techniques are required and it is known that categorical data violate the major assumptions involved in many such analyses (DEMPSTER and LERNER, 1950; GIANOLA, 1982). First, for binary data, genetic and phenotypic values are not normally distributed, therefore, the linearity between these values required in the prediction of gain does not hold (FOULLEY, 1992). Second, parameters estimated for binary traits are usually dependent on the incidence of the trait in the population (DEMPSTER and LERNER, 1950; GIANOLA, 1982). Third, selection intensity is limited the incidence of the trait (FALCONER, 1989). Fourth, genetic gain for binary traits is given by the difference in incidences (or probabilities of occurrence) of a given phenotype in consecutive generations, rather than differences in means for continuous traits. Therefore, gain is dependent on factors affecting both the mean and the variance of the distribution of the underlying trait.

Most techniques of analysis for threshold traits are based on the threshold, multifactorial model presented by WRIGHT (1934). According to this model, the measured binary trait is underlain by a continuous trait (sometimes called liability) controlled by many minor, additive genes and environmental factors. The discreteness of the binary trait is then produced by a threshold, such that if the underlying continuous trait is larger than some threshold value one phenotype is observed (e.g., infected), otherwise the alternative phenotype is observed (e.g., uninfected).

Prediction of gain for binary threshold traits is complicated by the dependence of genetic parameter estimates on the incidence of the trait in the population (DEMPSTER and LERNER, 1950; GIANOLA, 1982). Several approaches have been suggested to obtain estimates of those parameters which are independent of the incidence. DEMPSTER and LERNER (1950), based on WRIGHT's (1934) threshold model, suggested that heritability on binary scale should be converted to an underlying continuous scale to be independent of the frequency of the trait. This technique has been the most studied and used in animal breeding. Other authors have suggested data transformation (ROCKWOOD and GODDARD, 1973; SOHN and GODDARD, 1979), Bayesian methods (LUSH *et al.,* 1948; HOESCHELE *et al.,* 1987), estimation assuming random effects (the genetic entities) are distributed as beta-binomial (MAGNUSSEN and KREMER, 1995), and more recently some authors have suggested techniques of estimation based on generalized linear mixed models (SCHALL, 1991; WOLFINGER and O'CONNELL, 1993). However, the use of standard linear methods applied directly on 0/1 data as if they were continuous (without any transformation) is still largely used in forest breeding programs (e.g., DIETERS *et al.,* 1996).

The accuracy of some of these methods of estimation has been evaluated (VAN VLECK, 1972; MANTYSAARI *et al.,* 1991; MAGNUSSEN and KREMER, 1995). However, powerful techniques such as those based on generalized linear mixed models (SCHALL, 1991; WOLFINGER and O'CONNELL, 1993) have not been compared with simpler techniques such as those using 0/1 data as continuous and that suggested by DEMPSTER and LERNER (1950). Besides that, usually comparisons of techniques are made on the underlying scale parameters, even knowing that the major interest of the breeder is in genetic gain on the observable binary scale, because this is the scale where progress is made and is the only observable scale. Yet usually no connection with realized gain in this observed scale has been set forth by investigators when comparing methods, and usually the analyses only compare heritability values on the continuous unobservable scale.

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[1]) This is Journal Series of the Institute of Food and Agricultural Sciences, University of Florida, Gainesville.

[2]) School of Forest Resources and Conservation at the University of Florida, Gainesville, FL 32611-0410, USA. Currently: Cacao Research Center (CEPEC/CEPLAC), Ilheus, BA, 45600-000, Brazil.

[3]) School of Forest Resources and Conservation, University of Florida, Gainesville, FL 32611-0410, USA.

E-mail: uilsonvl@uol.com.br (UILSON V. LOPES)
dah@harm.sfrc.ufl.edu (DUDLEY A. HUBER)
tlw@harm.sfrc.ufl.edu (TIMOTHY L. WHITE)

The major objectives of this study were to compare the following types of binary genetic gains using a larger number of samples than previous works: a) gain realized from simulations of a complete cycle of mass selection breeding (taken as the standard against which other methods are compared); b) gain predicted using heritability estimated directly from 0/1 data (ANOVA method); c) gain predicted using heritability on the underlying scale converted from that obtained in the ANOVA method by DEMPSTER and LERNER's (1950) formula (DL method); d) gain predicted using an approach similar to that used with DEMPSTER and LERNER's method, but using the true underlying heritability instead that converted by DEMPSTER and LERNER's formula (PT method); e) gain predicted using heritability on the underlying scale obtained using generalized mixed model as described by WOLFINGER and O'CONNELL (1993) and implemented in the SAS® macro called GLIMMIX described in LITTELL *et al.* (1996) (WO method); and f) gain predicted using heritability on the underlying scale obtained using generalized mixed models as described by SCHALL (1991) and implemented in the ASREML package by GILMOUR *et al.* (1997) (SCHALL method).

Material and Methods

Simulations

For each of several different situations, simulation produced a complete generation of mass selection on a binary threshold trait as might be common in a breeding program. The simulation steps for a given generation were as follows: generate a base population (referred to as BP population), make selections, allow random intermating among selections, and generate a progeny population (referred to as PP population). The simulations mimic, for example, populations of plants with different frequencies of infected individuals with a disease; where disease-free individuals are selected, intermated and the resulting progeny evaluated. For convenience, this binary trait will be referred to as infected/uninfected by a hypothetical disease. Then, the objective was to identify methods that accurately predict the reduction in incidence of this disease in the progeny population due to mass selection of uninfected parents.

Two traits were simulated following the threshold model: an underlying continuous trait ($y_{(0)ij}$) and the resultant binary trait ($x_{(0)ij}$). The model used to simulate the continuous trait was

$$y_{(0)ij} = \mu_{(0)} + f_{(0)i} + w_{(0)ij} = \mu_{(0)} + f_{(0)i} + u_{(0)ij} + e_{(0)ij} \qquad [1]$$

where

$y_{(0)ij}$ is the underlying (unobservable) continuous trait in the base population (generation zero or BP population);

$\mu_{(0)}$ is the general mean;

$f_{(0)i}$ is the random effect of the i^{th} female general combining ability,

$i = 1,...,80$, $f_i \sim N(0, 1/4\sigma^2_a)$;

$w_{(0)ij}$ is the random effect of the j^{th} individual within i^{th} family, $j = 1,...,24$, $w_{(0)ij} \sim N(0, 3/4\sigma^2_a + \sigma^2_e = \sigma^2_w)$;

$u_{(0)ij}$ is the random effect associated to the j^{th} male general combining ability and the MENDELian sampling of the ij^{th} individual, $u_{(0)ij} \sim N(0, 3/4\sigma^2_a)$; and

$e_{(0)ij}$ is the error associated to the j^{th} individual of the i^{th} family in the base population, $e_{(0)ij} \sim N(0, \sigma^2_e)$.

In the simulations of the base population, $\mu_{(0)}$ was assumed zero. The effects $f_{(0)i}$, $u_{(0)ij}$ and $e_{(0)ij}$ were drawn from a normal distribution with zero mean and variances $1/4\sigma^2_a$, $3/4\sigma^2_a$, and σ^2_e, respectively. Covariances among all effects in model 1 were assumed to be zero. The true breeding values ($a_{(0)ij}$) of the 1920 individuals in a given sample (80 families * 24 individuals) were obtained by $a_{(0)ij} = f_{(0)i} + u_{(0)ij}$ in the base population. These breeding values were retained and used in the generation of the progenies of the selected individuals, as described in model 3.

The binary trait ($x_{(0)ij}$) was generated for each ij^{th} individual by imposing a threshold (T) in the population of $y_{(0)ij}$'s as

$$x_{(0)ij} = \begin{cases} 0 \text{ if } y_{(0)ij} \leq T & \text{(desirable phenotype)} \\ 1 \text{ if } y_{(0)ij} > T & \text{(undesirable phenotype)} \end{cases} \qquad [2]$$

Different threshold values were imposed in the distribution of the continuous trait to simulate five incidences (5%, 25%, 50%, 75%, and 95%) of the undesirable phenotype (i.e., diseased individuals). For each of the five incidences and five true heritabilities on the underlying scale (h^2 = 0.10, 0.30, 0.50, 0.70, and 0.90), 300 independent BP populations each with 80 half-sib families and 24 individuals per family were simulated, resulting in 7500 heritability-incidence-population combinations (5 heritabilities x 5 incidences x 300 populations).

In each of the 7500 simulated base populations (BP), 80 disease-free individuals were independently mass selected based on the binary trait (individuals with $x_{(0)ij}$= 0) and randomly crossed with 24 different individuals out of the 80 available, generating 24 half-sib offsprings per cross. No action was taken to avoid reciprocal crosses. These 80 simulated half-sib families were hypothetically planted in an experiment (PP population) with the same design as the parental base population described above, that is, 24 individuals from each of the 80 families completely randomized.

The phenotypic values of the progenies of selected parents (PP population) were obtained according to the model

$$y_{(1)ij} = \mu_{(1)} + (\tfrac{1}{2} a_{(0)if} + \tfrac{1}{2} a_{(0)im}) + v_{(1)ij} + e_{(1)ij} \qquad [3]$$

where

$y_{(1)ij}$ is the underlying continuous trait in the progeny population (generation 1);

$a_{(0)if}$ is the breeding value of the *female* parent of the i^{th} individual in generation zero;

$a_{(0)im}$ is the breeding value of the *male* parent of the i^{th} individual in generation zero;

$v_{(1)ij}$ is the random effect associated to the MENDELian sampling of the ij^{th} individual, $v_{(1)ij} \sim N(0, 1/2\sigma^2_a)$; and

$e_{(1)ij}$ is the error associated with the j^{th} individual of the i^{th} family in the progeny population, $e_{(1)ij} \sim N(0, \sigma^2_e)$.

In model 3, $v_{(1)ij}$ and $e_{(1)ij}$ are drawings from a normal distribution with zero mean and variances $1/2\sigma^2_a$ and σ^2_e, respectively. Further, $a_{(0)if}$ and $a_{(0)im}$ are true breeding values, on the underlying scale, of the individuals selected from the parental base population. These were retained for all parents from the simulation of the parental base population.

The same threshold values used in the parental base population (BP) were also applied in the progeny population (PP) to generate the binary variable $x_{(1)ij}$ (equation 2). The difference between the incidences in the progeny of the selected population and the parental base population was used as a measure of the realized genetic gain (reduction in incidence of the binary trait from one generation of mass selection). At the

same time, in each of the 7500 BP populations, genetic parameters were estimated by several analytical methods to predict the gain made by selection. The realized gain was the standard used to compare these analytical methods of gain prediction, since this is the one that is observed after selection, and quantified by the breeder.

Three hundred realized gains were obtained for each method, each level of heritability and each level of incidence of the base population. Therefore, 7500 realized gains were obtained for each method (5 heritabilities x 5 incidences x 300 populations). Results for 0.7 are not shown because the general pattern was the same as that shown for heritability 0.9.

Gains on the binary scale

a) *Realized gain* [$G_{R(x)}$]: The realized gain on the binary scale was obtained by:

$$G_{R(x)} = |p_{(1)} - p_{(0)}| \qquad [4]$$

where $G_{R(x)}$ is the realized gain on the binary scale; $p_{(1)}$ is the mean incidence of the undesirable phenotype in the progeny of the selected parents for the binary trait; and $p_{(0)}$ is the mean incidence of the undesirable phenotype in the parental base population for the binary trait.

b) *ANOVA method* [$G_{ANOVA(x)}$]: Gain was obtained by estimating variance components by the ANOVA method (Type I method of SAS®) on the BP tests using 0/1 data directly and obtaining heritability using these components as:

$$h^2_{ANOVA(x)} = 4 \times \sigma^2_{f(x)} / [\sigma^2_{f(x)} + \sigma^2_{w(x)}] \qquad [5]$$

where $h^2_{ANOVA(x)}$ is the individual heritability on the binary scale (x) as estimated by ANOVA on 0/1 data; $\sigma^2_{f(x)}$ is the general combining ability variance component; and $\sigma^2_{w(x)}$ is the within family variance component.

The heritability obtained by equation 5 was then used to predict gain on the binary scale by:

$$G_{ANOVA(x)} = |h^2_{ANOVA(x)} \times S_{(x)}| = |h^2_{ANOVA(x)} \times (0 - p_{(0)})| = h^2_{ANOVA(x)} \times p_{(0)} \qquad [6]$$

where $G_{ANOVA(x)}$ is the predicted gain on the binary scale using heritability estimated directly on 0/1 data and $S_{(x)}$ is the selection differential on the binary scale.

c) *DL method* [$G_{DL(x)}$]: Gain was predicted using the method described by DEMPSTER and LERNER (1950). The heritability on the binary scale ($h^2_{ANOVA(x)}$, equation 5) was converted to the underlying scale, $h^2_{DL(y)}$, by DEMPSTER and LERNER's (1950) method:

$$h^2_{DL(y)} = [h^2_{ANOVA(x)} \times p_{(0)}(1-p_{(0)})]/c^2 \qquad [7]$$

and gain on the underlying scale ($G_{DL(y)}$) was obtained by:

$$G_{DL(y)} = [i \times h^2_{DL(y)} \times \sigma_{y(0)}]/\sigma_{y(0)} = i \times h^2_{DL(y)} \qquad [8]$$

where $h^2_{DL(y)}$ is the heritability on the underlying scale; c is the ordinate of the normal density function at the threshold; $G_{DL(y)}$ is the predicted gain on the underlying scale, obtained by conversion of heritability from binary to underlying by DEMPSTER and LERNER's method, expressed in units of standard deviations; i is the selection intensity; and $\sigma_{y(0)}$ is the phenotypic standard deviation of the underlying trait.

This gain on the underlying scale, $G_{DL(y)}$, was converted to a binary scale gain by:

$$G_{DL(x)} = \int_{z_{(1)}}^{+\infty} f(z)dz - \int_{z_{(0)}}^{+\infty} f(z)dz \qquad [9]$$

with, $Z_{(0)} = [T - \mu_{(0)}]/\sigma_{y(0)}$; and

$Z_{(1)} = Z_{(0)} + G_{DL(y)}$

where $G_{DL(x)}$ is the predicted gain on the binary scale transformed from the underlying continuous gain obtained by DEMPSTER and LERNER's method; f(z) is the standard normal probability density function; $Z_{(1)}$ is the normal deviate on the underlying scale corresponding to the mean incidence in the PP population; $Z_{(0)}$ is the normal deviate on the underlying scale corresponding to the mean incidence in the BP population; and T is threshold value. The area under the normal curve defined by the integrals in equation 9 was computed using the PROBNORM function from SAS®.

d) *PT method* [$G_{PT(x)}$]: Gain on the binary scale was obtained using the same steps described for the second type above (DL method); however, the underlying gain in equation 8 was computed using the true underlying heritability instead the underlying heritability converted from a heritability estimated on the binary scale. This mimics a situation in which a method exists that when applied to binary threshold traits would give the true underlying heritability. Some methods approximate to this „perfect" method for some but not for all situations (VAN VLECK, 1972; MANTYSAARI *et al.,* 1991; MAGNUSSEN and KREMER, 1995). The question is: If the true heritability on the underlying scale is known, can it be accurately converted to realized gain on an observable binary scale?

e) *SCHALL method* [$G_{SCHALL(x)}$]: Gain was computed using the method proposed by SCHALL (1991) and implemented in the program ASREML by GILMOUR *et al.* (1997). This is based on a method suggested by STIRATELLI *et al.* (1984) for solution of fixed and random effects and dispersion (variance) parameters in a generalized linear mixed model context. Contrary to the WO method described below, the SCHALL method does not estimate the scale or dispersion parameter (here the error variance component). It automatically sets it to one ($o^2_{w(y)} = 1$). The estimates of variance components on the underlying scale obtained from this method were used to estimate the heritability:

$$h^2_{SCHALL(y)} = 4x\sigma^2_{f(y)SCHALL} / (\sigma^2_{f(y)SCHALL} + \sigma^2_{w(y)SCHALL})$$

where $\sigma^2_{f(y)SCHALL}$ and $\sigma^2_{w(y)SCHALL}$ are the estimates of variance components obtained by the SCHALL method. The genetic gain on the underlying scale was predicted as $G_{SCHALL(y)} = ixh^2_{SCHALL(y)}$. This gain ($G_{SCHALL(y)}$) was then converted to a binary scale as described in equation 9 using $G_{SCHALL(y)}$ in place of $G_{DL(y)}$ and $\sigma^2_{y(0)SCHALL} = (\sigma^2_{f(y)SCHALL} + \sigma^2_{w(y)SCHALL})$ in place of $\sigma^2_{y(0)}$. The resulting gain on the binary scale is $G_{SCHALL(x)}$.

f) *WO method* [$G_{WO(x)}$]: Gain was obtained by the method developed by WOLFINGER and O'CONNELL (1993) and implemented in the GLIMMIX macro written in SAS® (LITTELL *et al.,* 1996). Unlike the SCHALL method, in the WO method both components $\sigma^2_{f(y)WO}$ and $\sigma^2_{w(y)WO}$ are estimated. After these components were estimated using GLIMMIX, the heritability ($h^2_{WO(y)}$) gain on the underlying scale ($G_{WO(y)}$) were computed as described for SCHALL method and then the gain converted to the binary scale (now called $G_{WO(x)}$).

Heritability on the binary scale

Three heritabilities were studied to understand the differences in gains: a) the heritability using variance components estimated using 0/1 data ($h^2_{ANOVA(x)}$); b) the heritability on the binary scale converted from the true heritability by DEMPSTER and LERNER's method ($h^2_{TDL(x)}$); and c) the realized heritability

($h^2_{R(x)}$). The heritability $h^2_{TDL(x)}$ was computed from equation 7 substituting $h^2_{DL(y)}$ with the true heritability and solving for $h^2_{ANOVA(x)}$ (now $h^2_{TDL(x)}$). The realized heritability was computed by:

$$h^2_{R(x)} = G_{R(x)}/p_{(0)} \qquad [10]$$

Results and Discussion

Heritability and gain on the binary scale

Realized gain is a standard method to compare prediction techniques for continuous traits. However, this approach has not been used to compare methods of estimation of heritability for binary threshold traits, even knowing the major use of heritability is the prediction of gain. Moreover, the interest of the breeder is in the realized genetic gain on an observable scale. In this section, heritabilities and gains on a binary scale are presented and compared with those realized by a cycle of mass selection in the binary trait.

Heritabilities on the binary scale are presented on *figure 1,* each point being an average of 300 samples. For most situations the heritability using variance components estimated from 0/1 data directly ($h^2_{ANOVA(x)}$, equation 5) tracks that obtained on the binary scale converted from the true heritability by DEMPSTER and LERNER's (1950) method ($h^2_{TDL(x)}$) (*Figure 1*). These two heritabilities ($h^2_{ANOVA(x)}$, $h^2_{TDL(x)}$) provide good estimates of the realized heritability ($h^2_{R(x)}$, equation 10) at low true values of heritability. However, they underestimate the realized heritability at high true heritabilities, especially when the incidence of the undesirable phenotype is high. These results agree with those from YANG *et al.* (1998). Further, the heritabilities obtained by DEMPSTER and LERNER's (1950) method and those obtained from variance components estimated using 0/1 data are symmetric around 50% incidence, but the realized heritability is not symmetric (*Figure 1*). Therefore, when comparing methods of estimation of binary threshold traits, it is not a good strategy to examine only incidences smaller than 50% or only incidences larger than 50%. The results obtained from one side of 50%, may not be valid for the other side as is assumed for symmetry.

Realized and predicted gains on the binary scale are presented in *figure 2*. For traits of low heritability (h^2<0.3) all methods, including the ANOVA method based on 0/1 data, result in predicted gains close to that realized by selection across all incidences (5% to 95%). However, the WO method from WOLFINGER and O'CONNELL (1993) overestimated the gain at incidences of the undesirable phenotype larger than 75%, even for low true heritabilities (h^2 = 0.1). For traits of moderate to high heritabilities (h^2 > 0.5) and at high incidences (p > 60%), both DL and WO methods overestimate, while ANOVA and SCHALL underestimate the realized gain. The SCHALL method is the least biased among those tested.

For true heritabilities smaller than 0.5, the maximum realized gain and gains predicted by ANOVA and SCHALL methods occur near 75% incidence of the undesirable phenotype. SOHN

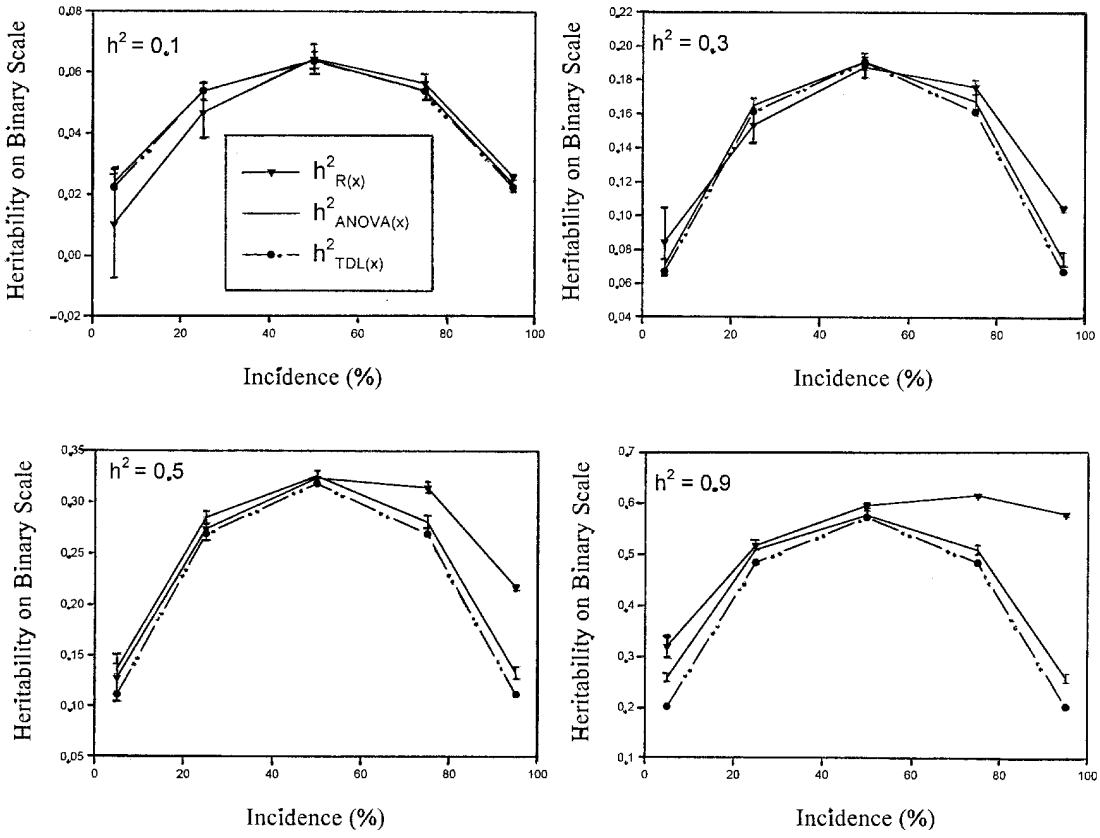

Figure 1. – Realized heritability on the binary scale ($h^2_{R(x)}$) and heritabilities obtained from variance components estimated directly on 0/1 data ($h^2_{ANOVA(x)}$) and obtained by transforming the true underlying heritability to binary scale by DEMPSTER and LERNER's method ($h^2_{TDL(x)}$), for several levels of incidence of the undesirable phenotype and true heritability levels on the underlying scale (h^2). Each point is an average of 300 samples of simulated experiments and the vertical bars are 95% confidence intervals.

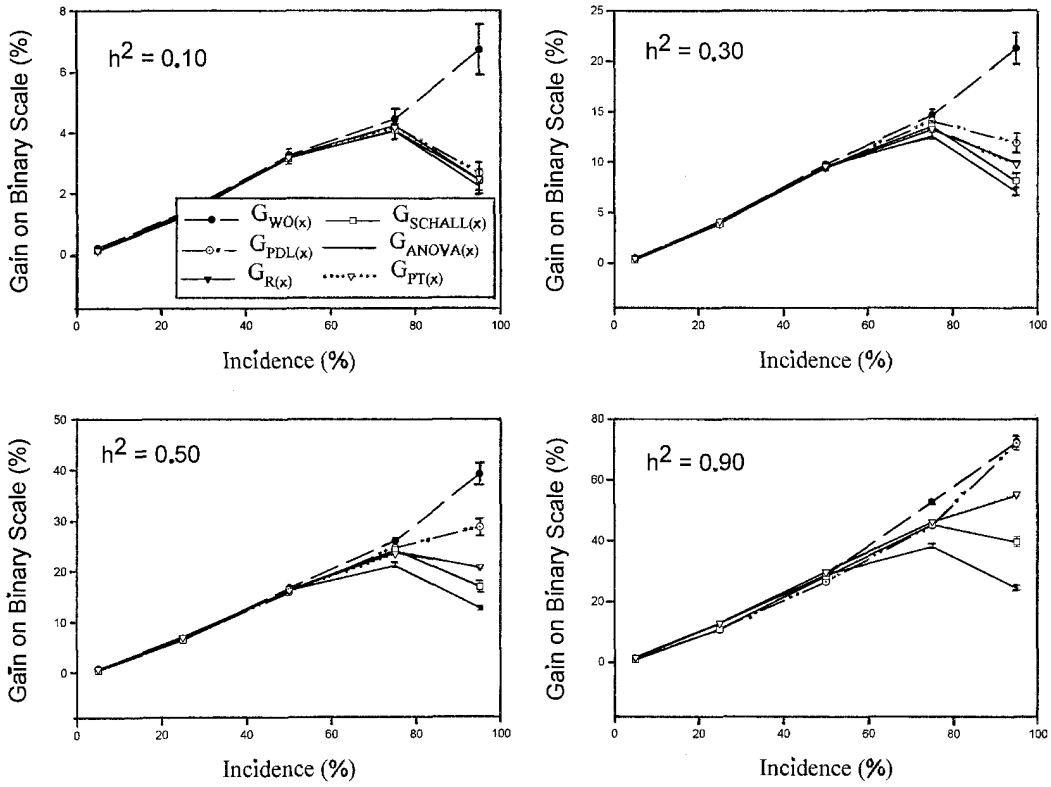

Figure 2. – Realized gain on the binary scale ($G_{R(x)}$) and gains predicted by WO ($G_{WO(x)}$), DL ($G_{DL(x)}$), SCHALL ($G_{SCHALL(x)}$), AVOVA ($G_{ANOVA(x)}$), and PT methods ($G_{PT(x)}$), expressed in percent of incidence, for several levels of incidence of the undesirable phenotype and true heritability levels on the underlying scale (h^2). Note that the graphs for $G_{R(x)}$ and $G_{PT(x)}$ are superimposed and that each point is an average of 300 simulated experiments and the vertical bars are 95% confidence intervals.

and GODDARD (1979), using real data of fusiform rust incidence in pines (a fungal disease of pines), observed that the gain predicted using ANOVA method on 0/1 data tracks the realized gain, with a slight underestimation. Those authors suggested that maximum gain from mass selection would occur at disease incidences of 70%. The predicted gain peaks near 75% because it is a combination of two factors (see equation 6): the heritability on the binary scale (which peaks at 50%, *Figure 1*) and selection differential (which is the incidence in the base population and peaks at 100%). These two factors are jointly maximized in the region between 50% and 100%.

Since the predictions are very close to the realized gain at low incidences of the undesirable phenotype (infected) and not so close at high incidences, we might be attempted to analyze the frequency of the desirable phenotype (uninfected) for high incidences of the undesirable phenotype. However this is not true. For example, for the ANOVA method, the heritability at 5% and 95% of incidence is 0.26, for a true heritability equal to 0.90 (*Figure 1*). However, the selection differential is an increasing function (equation 6). Therefore, below 50% the selection differential, $S_{(x)}$, is very small and then the underprediction of the gain very small. But at higher incidences, the selection differential is large and then the underprediction is also large.

Although many complex methods have been suggested to analyze binary threshold traits (DEMPSTER and LERNER, 1950; SCHALL, 1991; WOLFINGER and O'CONNELL, 1993), it is clear from the results presented here that heritability estimated from 0/1 data directly results in predicted gain close to the realized gain for traits of low heritability ($h^2 \leq 0.3$) and for traits of high heritability with incidences smaller than 75%. Many estimates of heritability obtained for binary threshold traits in animals and plants, when transformed to the underlying scale by DEMPSTER and LERNER's (1950) method, will be smaller than 0.3 (e.g., LUSH *et al.,* 1948; ROBERTSON and LERNER, 1949; ROCKWOOD and GODDARD, 1973; TOMAR and KUMAR, 1991; DIETERS *et al.,* 1996). Therefore, at least for these published results, the estimation of heritability directly from 0/1 data is appropriate if the goal is to use this heritability to predict gain from mass selection on the observable binary scale in a single environment.

ELSTON (1977) has defended the use of the heritability directly on the binary scale. According to him, although heritability estimated on the underlying is independent of the incidence of the binary trait, it is dependent on the distribution of the unobservable, underlying continuous trait; which is dependent on the mode of inheritance of the trait (monogenic or polygenic). However, the use of heritability estimated directly on 0/1 data has also received some criticisms, the major one being the dependence of the estimate on the incidence; not allowing comparison of estimates obtained from populations with different incidences (DEMPSTER and LERNER, 1950; GIANOLA, 1982). Another criticism is the dependence among effects in the linear model (GIANOLA, 1982). However, as pointed out by GUIARD *et al.* (1985), to estimate variance components there is no requirement of independence of the effects in the model; what is in fact required is the absence of correlation among those effects. These authors showed this correlation is zero for binary traits, providing support for estimation of variance components directly from 0/1 data.

Heritability on the underlying scale

Although the major focus of this investigation was the estimation of heritability and prediction of gain on the binary observable scale, heritabilities on the underlying scale were obtained to evaluate the methods on the same basis as other authors (VAN VLECK, 1972; MANTYSAARI *et al.,* 1991), but with a larger number of samples. These heritabilities are presented in *figure 3.*

The method suggested by DEMPSTER and LERNER (1950) closely estimates the true underlying heritability at incidences between 25% and 75%. However, this method overestimates the true heritability of the underlying trait for extreme incidences of the undesirable phenotype (incidences either smaller than 25% or larger than 75%) (*Figure 3*). The overestimation is larger for larger values of heritability. In general terms, these results agree with most studies evaluating DEMPSTER and LERNER's method (DEMPSTER and LERNER, 1950). Overestimation at extreme incidences and higher heritabilities has been shown by VAN VLECK (1972) and by MCGUIRK (1989), referring to unpublished results of MCGUIRK and THOMPSON. However, VAN VLECK and GREGORY (1992) observed the overestimation associated with DL method occurs at lower heritability and MANTYSAARI *et al.* (1991) observed no overestimation at all. MANTYSAARI *et al.* (1991) pointed out as a possible reason for the absence of overestimation in their study, the fact that the transformation was done sample by sample and using the incidence of that sample, not the presumed level of incidence. However, we used the same approach in our study and still we observed overestimation. A possible reason for lack of overestimation in MANTYSAARI and coworkers' study is the fact they used a low true heritability ($h^2 = 0.20$). At low heritabilities, we observed the DL method only slightly overestimates the true underlying heritability (*Figure 3*).

Other methods of estimation used here were those based on generalized linear mixed models (SCHALL, 1991; WOLFINGER and O'CONNELL, 1993), a powerful technique particularly appropriate for estimation of fixed and random effects for non-normal data, including binary threshold data. The algorithms by SCHALL (1991) and by WOLFINGER and O'CONNELL (1993), despite their incorporation into two major software packages for data analysis (SAS® and ASREML), have not been studied in the context of estimation of heritability or gain prediction for binary traits. The method using the algorithm from WOLFINGER and O'CONNELL (1993) (WO method, $h^2_{WO(y)}$), tracks the true heritability only for intermediate incidences (*Figure 3*). At extreme incidences, the heritabilities estimated by this method overestimate the true heritability, as occurred with DL method. However, the bias associated with WO method is larger than that associated with DL method. On the other hand, the method using the algorithm suggested by SCHALL (1991) (SCHALL method, $h^2_{SCHALL(y)}$) underestimates the true heritability (*Figure 3*). The underestimation is accentuated at extreme incidences and at higher heritabilities. The bias associated with the SCHALL method is close to that assaciated to DL method, but in the opposite direction.

The SCHALL method produced estimates of variance components for the random effects (here family variance component) smaller than those produced by WO method, particularly at extreme incidences (results not presented; see LOPES, 1998).

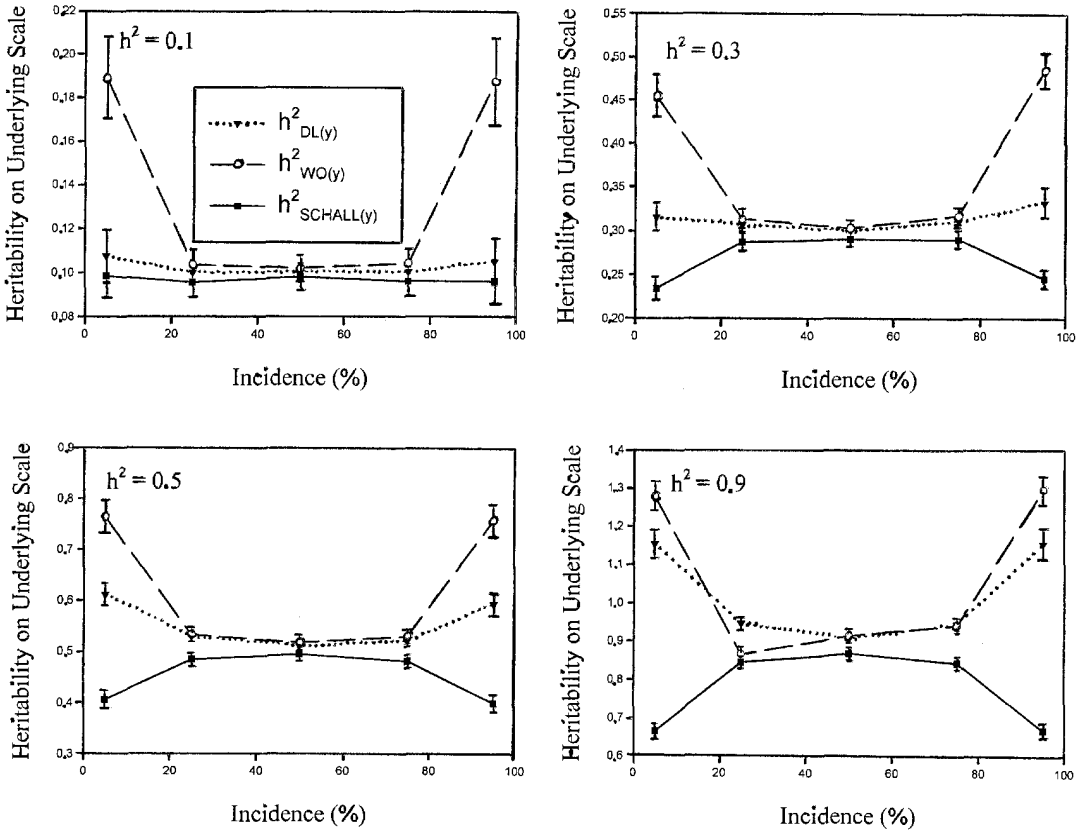

Figure 3. – Heritabilites on the underlying scale obtained from binary data by WO ($h^2_{WO(y)}$), SCHALL ($h^2_{SCHALL(y)}$), and DL methods (h^2 $DL_{(y)}$), for several incidences of the undesirable phenotype and true heritability levels on the underlying scale (h^2). Each point is an average of 300 samples of simulated experiments and the vertical bars are 95% confidence intervals.

These results confirm those obtained by WOLFINGER and O'CONNELL (1993) with salamander mating data. The SCHALL method constrains the error variance component to one (when this is smaller than one if not constrained), resulting in smaller estimates for the other components. The combination of smaller family variance component estimates ($\sigma^2_{f(y)SCHALL}$) and larger error variance estimates ($\sigma^2_{w(y)SCHALL}$) in the SCHALL method, resulted in underprediction of heritability (*Figure 3*).

Currently, the approach used to compare estimates of heritability from different estimation methods is to compare them with the true heritability of the underlying trait. However, another attractive strategy is the use of the realized gain on the binary observable scale as standard of comparison. In this study the true heritability on the underlying continuous scale was used, first, to predict gain on the underlying scale and then this gain was transformed to binary scale ($G_{PT(x)}$, equation 8 substituting $h^2_{DL(Y)}$ by the true heritability). This mimics the situation in which a perfect method of estimation of heritability on the underlying scale for threshold traits can be developed. The gain obtained in this way matches perfectly the gain realized from selection ($G_{R(x)}$, *Figure 2*). Therefore, the two strategies used to compare methods of estimation of heritability or gain prediction are equivalent. This supports the strategy of most authors in comparing their methods of estimation of heritability for binary threshold traits with the true underlying heritability (VAN VLECK, 1972; OLAUSSON and RONNINGEN, 1975; MCGUIRK, 1989).

Conclusions

Although many complex methods have been suggested to analyze binary threshold traits (DEMPSTER and LERNER, 1950; SCHALL, 1991; WOLFINGER and O'CONNELL, 1993), it is clear from the results presented here that heritability obtained from 0/1 data directly (without transformation of data or parameter estimates) results in predicted gain close to the realized gain for traits of low heritability ($h^2 \leq 0.3$) and for traits of high heritability with incidences smaller than 75%. Many traits in animal and plant breeding fall into this category. One of the limitations of this study is that it is restricted to mass selection in a single test location (single mean incidence). The results found here may not apply to situations in which tests with different incidences are combined together for prediction of gain or for more complex types of selection indices.

The method by DEMPSTER and LERNER (1950), which is the most extensively evaluated and used methodology for estimating heritability of threshold traits, and the methods by WOLFINGER and O'CONNELL (1993) and SCHALL (1991), based on generalized linear mixed models, do not result in better prediction of the gain on the observable binary scale than methods using the heritability estimated directly from binary data.

The results of this paper support the strategy of most authors in comparing their methods of estimation of heritability of binary threshold traits with the true or estimated underlying heritability (VAN VLECK, 1972; OLAUSSON and RONNINGEN, 1975; MCGUIRK, 1989; MANTYSAARI *et al.,* 1991). In this paper it was shown that methods of analysis of binary data that produce estimated heritabilities on the underlying scale close to the true heritability, also result in accurate predictions of gain on the binary observable scale.

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