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GENETIC MODELS AND ANALYSIS METHODS FOR SEX-LINKED AND MATERNAL GENE EFFECTS*

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ABSTRACT

Genetic models are proposed for analyzing sex-linked and maternal effects as well as autosomal gene effects. For the model with no genotype × environment interaction, the total genetic effect is partitioned into direct additive (A), direct dominance (D), sexlinked (L), maternal additive (Am) and maternal dominance (Dm) genetic components. For the model including genotype × environment interaction (GE), GE can also be partitioned into components of direct additive by environment interaction (AE), direct dominance by environment interaction (DE), sex-linked by environment interaction (LE), maternal additive by environment interaction (AmE), and maternal dominance by environment interaction (DmE). Linear functions of genetic components are listed for parent, F_1, and F_2. A set of parents, their reciprocal F_1's and F_2's is applicable for efficient analysis. Variance and covariance components can be well esti-

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mated by MINQUE(O/1) with the jackknife procedure. The t-test conducted by the jackknife procedure is applicable for detecting significance of variation. Adjusted Unbiased Prediction (AUP) method is suggested for predicting genetic effects.

Key Words: Diallel analysis, Sex-linked and maternal gene effects Genotype by environment interaction, Variance and covariance components Genetic prediction.

INTRODUCTION

In animal breeding experiments, sex-linked and maternal effects are the primary sources of reciprocal effects. Eisen et al.(*1966*) presented a model containing parameters for sex-linked and maternal effects as well as autosomal genetic effects. Cockerham and Weir (*1977*) suggested a bio-model, including parameters for maternal and paternal effects assuming no sex-linked effects. Carbonell et al. (*1983*) extended the model of Eberhart and Gardner (*1966*) to include sex-linked and maternal effects. In all of these models, variance components of sex-linked effects are not estimable by the standard least squares procedures or ANOVA methods.

Zhu and Weir (*1994*) proposed an animal model which can partition the maternal effects into maternal additive effects and maternal dominance effects. A MINQUE(O/1) method, which is a mixed linear model approach, was also proposed for estimating variance components and covariance components for the animal model (Zhu, *1992*; Zhu and Weir, *1994*).

In this study, genetic models with sex-linked effects, maternal additive and dominance effects are proposed for diallel crosses with a set of parents and their reciprocal F_1's and F_2's. Methods of estimating genetic variance and covariance components, and of predicting genetic effects are presented.

MODELS AND METHODOLOGY

A full diallel crossing system consists of all possible crosses between a set of parents. With method 1 of Griffing's (*1956*) definition for diallel

mating methods, a set of parents, F_1's and reciprocal F_1's as well as their F_2's from random mating among F_1's are suggested in use. The assumptions for our genetic models are: (1) regular diploid segregation; (2) inbred parents randomly sampled from a reference population; (3) no epistatic effects; (4) X (or Z) chromosome dosage compensation; and (5) inert Y (or W) chromosome in XY (ZW) cell. If there is no genotype × environment interaction, the general model for phenotypic mean of sex s in block l from generation k of the cross between maternal line i and paternal line j is

$$Y_{ijksl} = \mu + G_{ijks} + b_l + \varepsilon_{ijksl} \qquad (1)$$

where Y_{ijksl} is the average phenotypic value of genetic entry G_{ijks} in block l; , μ is a fixed population mean; b_l is the random effect of block l, $b_l \sim (O, \sigma^2_b)$; and ε_{ijksl} is a residual effect, $\varepsilon_{ijksl} \sim (O, \sigma^2_e)$.

Partition of the genotypic effects G_{ijks} for heterogametic progeny (XY or ZW, $S=1$) and for homogametic progeny (XX or ZZ, $S=2$) from dam $i \times$ *sire* j depends on specific entry. For parent ($k=O$, $i=j$) or F_1 ($k=1$, $i \neq j$):

$$G^{XY}_{(ijk1)} = A_i + A_j + D_{ij} + L_{jl} + 2A_{mi} + D_{mii}$$

$$or\ G^{ZW}_{(ijk1)} = A_i + A_j + D_{ij} + L_{jl} + 2A_{mi} + D_{mii} \qquad (2)$$

$$G^{XX/ZZ}_{(ijk2)} = A_i + A_j + D_{ij} + \frac{1}{2}L_{i2} + \frac{1}{2}L_{j2} + 2A_{mi} + D_{mji}$$

and for F_2 ($k=2$, $i \neq j$) from random mating among F_1's:

$$G^{XY/ZW}_{(ijk1)} = A_i + A_j + \frac{1}{4}D_{ii} + \frac{1}{4}D_{jj} + \frac{1}{2}D_{ij} + \frac{1}{2}L_{il} + \frac{1}{2}L_{jl} + A_{mi} + A_{mj} + D_{mij}$$

$$G^{XX}_{(ijk2)} = A_i + A_j + \frac{1}{4}D_{ii} + \frac{1}{4}D_{jj} + \frac{1}{2}D_{ij} + \frac{3}{4}L_{il} + \frac{1}{4}L_{jl} + A_{mi} + A_{mj} + D_{mij} \qquad (3)$$

$$or\ G^{ZZ}_{(ijk2)} = A_i + A_j + \frac{1}{4}D_{ii} + \frac{1}{4}D_{jj} + \frac{1}{2}D_{ij} + \frac{1}{4}L_{il} + \frac{3}{4}L_{jl} + A_{mi} + A_{mj} + D_{mij}$$

where A_i (or A_j) is the cumulative additive effect of the autosomal genes from dam i (or sire j), A_i (or A_j) $\sim (O, \sigma^2_A)$; D_{ij} is the cumulative dominance effect of the autosomal genes from the cross of dam $i \times$

sire j, $D_{ij} \sim (O, \sigma_D^2)$; L_i (or L_j) is the cumulative additive effect of the sex-linked genes in heterogametic offspring from parent i (or j) L_{i2} (or L_{j2}) is the cumulative additive effect of the sex-linked genes in homogametic offspring from parent i (or j), L_{i1}, L_{j1} (or L_{j1}, L_{j2}) $\sim$ (O σ_L^2); A_{mi} (or A_{mj}) is the maternal additive effect of dam i (or sire j), A_{mi} (or A_{nj}) $\sim$ (O, σ^2_{Am}); D_{mij} is the maternal dominance effect of the cross (dam i $\times$ sire j), $D_{mij} \sim (O, \sigma^2_{Dm})$. There are covariances between direct and maternal gene effects, $Cov(A_i, A_{mi}) = \sigma_{A.Am}$ and $Cov(D_i, D_m) = \sigma_{D.Dm}$

This genetic models can be written in the matrix form of a mixed linear model for all the entries in the mating design,

$$\mathbf{Y} = \mathbf{l}\mu + \mathbf{U}_A\mathbf{e}_A + \mathbf{U}_D\mathbf{e}_D + \mathbf{U}_L\mathbf{e}_L + \mathbf{U}_{Am}\mathbf{e}_{Am} + \mathbf{U}_{Dm}\mathbf{e}_{Dm} + \mathbf{U}_e\mathbf{e}_e$$

with variance-covariance matrix

$$Var(Y) = \sigma_A^2\mathbf{U}_A\mathbf{U}_A' + \sigma_D^2\mathbf{U}_D\mathbf{U}_D' + \sigma_L^2\mathbf{U}_L\mathbf{U}_L' + \sigma^2_{Am}\mathbf{U}_{Am}\mathbf{U}_{Am}' + \sigma^2_{Dm}\mathbf{U}_{Dm}\mathbf{U}_{Dm}'$$
$$+\sigma^2\mathbf{U}\mathbf{U}' + \sigma_{A.Am}(\mathbf{U}_A\mathbf{U}_{Am}' + \mathbf{U}_{Am}\mathbf{U}_A') + \sigma_{D.Dm}(\mathbf{U}_D\mathbf{U}_{Dm}' + \mathbf{U}_{Dm}\mathbf{U}_D')$$
$$+\sigma^2\mathbf{I} \qquad (4)$$

where $\mathbf{U}$ is the known incidence matrix relating to the random vector $\mathbf{e} \sim (\mathbf{O}, \sigma^2\mathbf{I})$; $\mathbf{U}'$ is the transpose of $\mathbf{U}$, and $\mathbf{I}$ is an identity matrix;

when genotype $\times$ environment interactions do exist, genetic experiments should pe conducted in different environments, The genetic model including genotype $\times$ environment interactions is an extension of Equation (1). Phenotypic mean of sex s in block l within environment h from generation k of the cross $(i \times j)$ is

$$Y_{hijks} = \mu + E_h + G_{ijks} + GE_{hijks} + b_{l(h)} + \varepsilon_{hijksl} \qquad (5)$$

where Y_{hijksl} is the average phenotypic value of genetic entry G_{ijks}; μ is a fixed population mean; E_h is the fixed effect of the h^{th} environment; $\mathbf{b}_{l(h)}$ is the random effect of block l within environment h, $\mathbf{b}_{l(h)} \sim$

(O, σ_0^2); and ε_{hijksl} is a residual effect, $\varepsilon_{hijksl} \sim (O, \sigma_e^2)$.

The genotypic effect G_{hijks} is defined the same way as in Equations (2) and (3). The effect of genotype × environment interaction GE_{hijks} is delined for parent ($k=O$, $i=j$) or F_1 ($k=1$, $i \neq j$) as:

$$\begin{aligned} GE^{XY}_{hijkl} &= AE_{hi} + AE_{hj} + DE_{hij} + LE_{hil} + 2A_mE_{hi} + D_mE_{hii} \\ or\ GE^{ZW}_{hijkl} &= AE_{hi} + AE_{hj} + DE_{hij} + LE_{hjl} + 2A_mE_{hi} + D_mE_{hii} \\ GE^{XY/ZZ}_{hijk2} &= AE_{hi} + AE_{hj} + DE_{hij} + \frac{1}{2}LE_{hi2} + \frac{1}{2}LE_{hj2} + 2A_mE_{hi} + D_mE_{hii} \end{aligned} \quad (6)$$

and for F_2 ($k=2$, $i \neq j$) from random mating among F_1's:

$$\begin{aligned} GE^{XY/ZW}_{hijkl} &= AE_{hi} + AE_{hj} + \frac{1}{4}DE_{hii} + \frac{1}{4}E_{hjj} + \frac{1}{2}DE_{hij} + \frac{1}{2}LE_{hil} + \frac{1}{2}LE_{hjl} + A_mE_{hi} \\ &\quad + A_mE_{hj} + D_mE_{hij} \\ GE^{XX}_{hijk2} &= AE_{hi} + AE_{hj} + \frac{1}{4}DE_{hii} + \frac{1}{4}E_{hjj} + \frac{1}{2}DE_{hij} + \frac{3}{4}LE_{hi2} + \frac{1}{4}LE_{hj2} + A_mE_{hi} \\ &\quad + A_mE_{hj} + D_mE_{hij} \\ or\ GE^{ZZ}_{hijk2} &= AE_{hi} + AE_{hj} + \frac{1}{4}DE_{hii} + \frac{1}{4}E_{hjj} + \frac{1}{2}DE_{hij} + \frac{1}{4}LE_{hi2} + \frac{3}{4}LE_{hj2} + A_mE_{hi} \\ &\quad + A_mE_{hj} + D_mE_{hij} \end{aligned} \quad (7)$$

where AE_{hi} (or AE_{hj}) is the cumulative additive × environment effect, AE_{hi} (or AE_{hj}) $\sim (O, \sigma^2_{AE})$; DE_{hij} is the cumulative dominance × environment effect, $DE_{hij} \sim (O, \sigma^2_{DE})$; LE_{hil} (or LE_{hjl}) is the cumulative additive × environment effect of the sex-linked genes in heterogametic offspring from parent i (or j), LE_{hi2} (or LE_{hj2}) is the cumulative additive × environment effect of the sex-lined genes in homogametic offspring from parent i (or j), LE_{hii}, LE_{hij} (or LE_{hjj}, LE_{hj2}) $\sim (O, \sigma^2_{LE})$; A_mE_{hi} (or A_mE_{hj}) is the maternal additive × environment effect, A_mE_{hi} (or A_mE_{hj}) $\sim (O, \sigma^2_{AmE})$; D_mE_{hij} is the maternal dominance × environment effect, $D_mE_{hij} \sim (O, \sigma^2_{DmE})$. Covari

ances between direct and maternal gene by environment interaction include $Cov(AE_i\ A_nE_i) = \sigma_{AE.AmE}$ and $Cov(DE_i\ D_mE_j) = \sigma_{DE.DmE}$.

The matrix form for all the entries in the mating design is

$$Y = \mathbf{X}\beta + \mathbf{U}_A e_A + \mathbf{U}_D e_D + \mathbf{U}_L e_L + \mathbf{U}_{Am} e_{Am} + \mathbf{U}_{Dm} e_{Dm} + \mathbf{U}_{AE} e_{AE} + \mathbf{U}_{DE} e_{DE} + \mathbf{U}_{LE} e_{LE} + \mathbf{U}_{AmE} e_{AmE} + \mathbf{U}_{DmE} e_{DmE} + \mathbf{U}_\mu e_\mu + \mathbf{U}_e e_e$$

with variancn-covariance matrix

$$\begin{aligned} Var(Y) = {} & \sigma_A^2 \mathbf{U}_A \mathbf{U}_A' + \sigma_D^2 \mathbf{U}_D \mathbf{U}_D' + \sigma_L^2 \mathbf{U}_L \mathbf{U}_L' + \sigma_{Am}^2 \mathbf{U}_{Am} \mathbf{U}_{Am}' + \sigma_{Dm}^2 \mathbf{U}_{Dm} \mathbf{U}_{Dm}' \\ & + \sigma_{AE}^2 \mathbf{U}_{AE} \mathbf{U}_{AE}' + \sigma_{DE}^2 \mathbf{U}_{DE} \mathbf{U}_{DE}' + \sigma_{LE}^2 \mathbf{U}_{LE} \mathbf{U}_{LE}' + \sigma_{AmE}^2 \mathbf{U}_{AmE} \mathbf{U}_{AmE}' \\ & + \sigma_{DmE}^2 \mathbf{U}_{DmE} \mathbf{U}_{DmE}' + \sigma_\mu^2 \mathbf{U}_\mu \mathbf{U}_\mu' + \sigma_{A.Am}(\mathbf{U}_A \mathbf{U}_{Am}' + \mathbf{U}_{Am} \mathbf{U}_A') \\ & + \sigma_{D.Dm}(\mathbf{U}_D \mathbf{U}_{Dm}' + \mathbf{U}_{Dm} \mathbf{U}_D') + \sigma_{AE.AmE}(\mathbf{U}_{AE} \mathbf{U}_{AmE}' + \mathbf{U}_{AmE} \mathbf{U}_{AE}') \\ & + \sigma_{DE.DmE}(\mathbf{U}_{DE} \mathbf{U}_{DmE}' + \mathbf{U}_{DmE} \mathbf{U}_{DE}') + \sigma_e^2 \mathbf{I} \end{aligned} \quad (8)$$

The MINQUE(O/1) method (Zhu, *1992*; Zhu and Weir, *1974*) can be used to estimate variance and covariance components in Equations (*4*) and (*8*). MINQUE(O/1) is a MINQUE method (Rao, *1971*) with prior values of zero for covariances and one for variances.

After obtaining unbiased estimates of variance and covariance components; phenotype variance can then be estimated for genetic model without genotype × environment interaction

$$V_P = V_A + V_D + V_L + V_{Am} + V_{Dm} + 2C_{A.Am} + 2C_{D.Dm} + V_e$$

or for genetic model with genotype × environment interaction

$$\begin{aligned} V_P = {} & V_A + V_D + V_L + V_{Am} + V_{Dm} + V_{AE} + V_{DE} + V_{LE} + V_{AmE} + V_{DmE} \\ & + 2C_{A.Am} + 2C_{D.Dm} + 2C_{AE.AmE} + 2C_{DE.DmE} + V_e \end{aligned}$$

Components of phenotype variance should be calculated according to each generation. For genetic model without genotype × environment interaction,

$$\begin{aligned} V_P(F_1) &= 2\sigma_A^2 + \sigma_D^2 + \sigma_L^2 + 4\sigma_{Am}^2 + \sigma_{Dm}^2 + 4\sigma_{A.Am}^2 + \sigma_e^2 \\ V_P(F_2) &= 2\sigma_A^2 + \frac{3}{8}\sigma_D^2 + \sigma_L^2 + 2\sigma_{Am}^2 + \sigma_{Dm}^2 + 4\sigma_{A.Am}^2 + \sigma_{D.Dm}^2 + \sigma_e^2 \end{aligned} \quad (9)$$

or for genetic model with genotype × environment interaction

methods which can be applied for estimating variance and covariance components in the models. Since these two methods need iterations, there are enormous computations involved with jackknife procedure. We suggest the use of the MINQUE(O/1) method for its unbiased estimation and non-iterative computation.

In breeding practice, parent and hybrid genetic merits are sometimes of more concern to the breeders. The random genetic effects in the genetic models are predictable by the BLUP procedure (Henderson, *1963*). Since BLUP needs all the parameter values of variance and covariance components, true BLUP can not be obtained in real experiments. When estimates of variance and covariance components are used in BLUP procedure, it will result in a so-called "BLUP" prediction which is then no longer a linear unbiased prediction. Monte Carlo simulations showed that AUP is superior to BLUP for its unbiasedness in both mean and variance of predicting genetic effects (Zhu, submitted).

Due to the random inactivation of one X-chromosome in the primitive ectoderm lineage of female mammals, the genetic activity of only one X-chromosome is expressed in somatic cells (Lyon, *1988*). By the assumption of X-chromosome dosage compensation, heterozygous females are mosaic with half chance inactivation of maternal or paternal X-chromosome. For some domestic animals, there may be no dosage compensation of sex-linked genes (Cox and Morton, *1963*). In those situations, the coefficients before L_{i2} and L_{j2} in Equations (2) and (3) or before LE_{i2} and LE_{h2} in Equations (6) and (7) should be dropped

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