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ESTIMATION OF QUANTITATIVE TRAIT LOCI VARIANCE COMPONENTS FOR SOMATIC CELL SCORE IN THE GERMAN HOLSTEIN DAIRY POPULATION

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INTRODUCTION

Somatic cell score (SCS) is an indicator trait for clinical mastitis and is often implemented in routine sire evaluations. Due to its low heritability, SCS is an attractive trait for use in marker assisted selection (MAS) programmes.

The mixed inheritance model, with random quantitative trait locus (QTL) and additive effects, is a suitable tool for statistically describing genetic variation in quantitative traits. Mixed QTL models account for additive polygenic relationships between animals as well as gametic relationships at specific positions on the genome, resulting in more exact parameter estimation. Recursive algorithms are commonly used for the efficient calculation of gametic relationship matrices (FERNANDO AND GROSSMAN 1989, WANG ET AL. 1995, ABDEL-AZIM AND FREEMAN 2001, TUCHSCHERER ET AL. 2004). However, when marker information is missing, direct application of recursive algorithms is impossible (GEORGE ET AL. 2000). The partial pedigree approach described by MAYER ET AL. (2007, submitted) and TUCHSCHERER ET AL. (2007, in preparation) allows for calculation of transmitting probabilities when marker information is missing and fits well into the recursive marker assisted best linear unbiased prediction (MA BLUP) methods currently in use.

The objective of this study was to estimate the proportion of total genetic variance attributed to a QTL on *Bos Taurus* autosome 18 for somatic cell score in the German Holstein dairy population with a data set currently used in the German MAS programme using a mixed inheritance model with transmitting probabilities calculated using a partial pedigree approach.

METHODS

Genotype information on a single highly polymorph marker with 15 alleles on chromosome 18 for 6,520 typed bulls and bull dams (6,050 males, 470 females) was provided by

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the German MAS-program. Allele frequencies ranged from 0.01% to 32.83%. A pedigree containing 12,008 animals (7,174 males and 4,834 females), including non-genotyped ancestors of genotyped animals, was available. There were a total of 201 founder bulls, and the maximum number of offspring for one bull was 834. Phenotype information for 5,021 genotyped offspring in the form of daughter yield deviations (DYDs) for the first lactation was also obtained. DYDs were weighted as described in BENNEWITZ ET AL. (2004) with genetic parameters for covariance matrices taken from REENTS ET AL. (1995).

Pedigree and marker information originated from the second phase of the genome analysis project of the Federation of German Cattle Breeders (Arbeitsgemeinschaft Deutscher Rinderzüchter, ADR) and is currently used for the MA BLUP evaluation of the trait somatic cell score. The pedigree and phenotype information was obtained from the United Information Systems Animal Production (Vereinigte Informationssysteme Tierhaltung w.V., VIT).

Transmitting probabilities were calculated using the partial pedigree strategy described by MAYER ET AL. (2007, submitted) and TUCHSCHERER ET AL. (2007, in preparation). IBD states were found by extracting standard partial pedigrees (individual, parents, grandparents) from original pedigree data and applying SimWalk2 (SOBEL AND LANGE 1996, SOBEL ET AL. 2001) / Merlin (ABECASIS 2002). The condensed gametic relationship matrix $\mathbf{G}^{*}$ - a version of $\mathbf{G}$ where linear dependencies have been removed - and its inverse $\mathbf{G}^{*-1}$ were calculated using COBRA (BAES AND REINSCH 2007, TUCHSCHERER ET AL. 2004).

The following mixed linear model was applied in ASReml Version 2.0 (GILMOUR ET AL. 2006) to incorporate marker information into parameter estimation (gametic effects model):

$$\begin{bmatrix} \mathbf{X'D^{-1}X} & \mathbf{X'D^{-1}Z} & \mathbf{X'D^{-1}W} \\ \mathbf{Z'D^{-1}X} & \mathbf{Z'D^{-1}Z + A_u^{-1}\alpha_1} & \mathbf{Z'D^{-1}W} \\ \mathbf{W'D^{-1}X} & \mathbf{W'D^{-1}Z} & \mathbf{W'D^{-1}W + G_v^{*-1}\alpha_2} \end{bmatrix} \begin{bmatrix} \hat{\boldsymbol{\mu}} \\ \hat{\mathbf{u}} \\ \hat{\mathbf{v}} \end{bmatrix} = \begin{bmatrix} \mathbf{X'D^{-1}y} \\ \mathbf{Z'D^{-1}y} \\ \mathbf{W'D^{-1}y} \end{bmatrix}$$

where $\mathbf{y}_{(m\times 1)}$ is a vector of *m* DYDs for *n* animals, μ is a fixed effect common to all observations, $\mathbf{u}_{(n\times 1)}$ is a vector of random polygenic effects and $\mathbf{v}_{(gam\times 1)}$ is a vector of random gametic effects of a marked QTL that is linked to a single polymorphic marker locus with *gam* equal to the number of unique gametes. Subscripts in parenthesis of the vectors and matrices denote their dimensions. $\mathbf{X}_{(m\times n)}$, $\mathbf{Z}_{(m\times n)}$ and $\mathbf{W}_{(m\times n)}$ are known incidence matrices, $\mathbf{A}^{-1}_{u\,(n\times n)}$ is the inverse of the relationship matrix and $\mathbf{G}^{*-1}_{v\,(gam\times gam)}$ is the inverse of the condensed gametic QTL relationship matrix. $\mathbf{D}^{-1}_{(m\times n)}$

is the inverse of a diagonal matrix containing weights for each observation. Expectations of $\mathbf{u}$, $\mathbf{v}$ and $\mathbf{e}$ and covariances between them are assumed to be zero, and $\boldsymbol{\alpha}_1 = \frac{\sigma_e^2}{\sigma_u^2}$ and $\boldsymbol{\alpha}_2 = \frac{\sigma_e^2}{\sigma_v^2}$.

RESULTS

The partial pedigrees were grouped by available genotype information. The individual and its male ancestors (sire and dam's sire) were genotyped in 33.6% of all partial pedigrees, followed closely by partial pedigrees in which the individual and its sire were genotyped (32.8%). Full marker information for all individuals was available in 1.2% of the partial pedigrees.

There were 19,368 unique gametes in the pedigree using a condensing factor of 0.02 (all gametes with transmitting probabilities >0.98 and those <0.02 were considered identical to their ancestral gametes). At the marker, 4,584 typed males and 373 typed females had inbreeding coefficients of zero, while the remaining 1,466 typed males had an average inbreeding coefficient of 0.0332 and the 97 remaining females one of 0.0342.

Random polygenic effects were estimated at 0.0741 and random gametic effects were estimated at 0.00231 directly at the marker position. The QTL variance was 0.00462; the ratio of QTL to polygenic variance was estimated at 5.87%, with a likelihood ratio test statistic of 14.62. Variance component estimation further away from the marker resulted in a QTL variance of 0.00477 at 5 cM from the marker and 0.00507 at 10 cM from the marker.

CONCLUSION

A mixed inheritance model was used to estimate the proportion of total genetic variance attributed to a QTL on *Bos Taurus* autosome 18 for somatic cell score in the German Holstein dairy population. The partial pedigree approach used to calculate QTL transmitting probabilities proved efficient. Our results indicate that the highly significant QTL in the chromosomal area studied is responsible for approximately 6% of the genetic variance in somatic cell score in the German Holstein population.

ACKNOWLEDGEMENTS

The authors would like to thank the Federation of German Cattle Breeders (Arbeitsgemeinschaft Deutscher Rinderzüchter, ADR) and United Information Systems Animal Production (Vereinigte Informationssysteme Tierhaltung w.V., VIT) for their supply of data. We thank Dörte Wittenburg for helpful comments on ASReml. Financial support from the FUGATO.M.A.S.-Net project is gratefully acknowledged.

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