PREDICTION OF ELBOW DYSPLASIA IN DOGS BY BREEDING VALUES OF RELATIVES

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SUMMARY

In the first part of a data set on elbow dysplasia in dogs (n=188) variance components and breeding values of animals were computed. Observations in the second part (n=65) were subsequently regressed on estimated breeding values. A direct effects model was found inappropriate: although a high heritability was found (0.53), regression of new observations on the pedigree index was -0.25. In a maternal effects model heritability was 0.34, and the relevant regression of new observations on the breeding value of the dam was 1.1, around its expected value of 1. Regressions on breeding values of grandparents revealed a possibly more complex inheritance and offers interesting starting points for further investigations. This case showed that an incorrect choice of model, here the direct effects model, could work counterproductive for prediction of new observations.

Keywords: dogs, elbow dysplasia, prediction of observations, maternal effects

INTRODUCTION

Several dog-breeders are faced with breed-specific defects. A well known defect in dogs is for instance hip dysplasia, but also various physiological defects are known. Due to the breed specificity a genetic basis for these defects is generally assumed. In this study, elbow dysplasia was analysed in a Labrador retriever population. For this population, Ubbink *et al.* (1997) showed that the disease was present in some related clusters within the population, which suggest a genetic bases for this defect as well.

In this study, two genetic models for elbow dysplasia were validated by studying the regressions of new observations on breeding values. In this manner, the employed genetic model(s) can be "externally" validated, which is important for setting up genetic counselling programmes. The models investigated in this study were two polygenic models, one assuming a direct effect on elbow dysplasia, and the other assuming a maternal effect.

MATERIAL AND METHODS

Records were available on a Labrador retriever populations owned (bred or bought) by the Royal Dutch Guide Dog for the Blind Association. All dogs present from 1988 to 1992, which were born between 1986 and 1992, were screened for signs of elbow dysplasia. Suspected dogs were further examined at the Clinic for Companion Animals of Utrecht University. The majority of cases of elbow dysplasia in this breed was found caused by a particular defect, a fragmented coronoid process, or LPC (Hazewinkel *et al.* 1988). In this study, this particular

LPC defect was analysed, using the dichotomy absent/present. In total, 243 records were available with a mean LPC incidence of 15.6%. Genetic inferences (variance components and breeding values) were based on 188 records of animals born before 1991. The remaining 65 records of animals born in 1991 and 1992 were then regressed on estimated breeding values of their parents or grandparents. Pedigrees of each animal were traced back for six generations, and pedigree records were extended to include all parents of animals with records to be predicted, resulting in a total of 3307 animals in the analyses.

Genetic models. A linear polygenic model with a single mean μ was used, denoted $\mathbf{y} = \mu + \mathbf{Zu} + \mathbf{e}$, where $\mathbf{Z}$ is a design matrix relating breeding values $\mathbf{u}$ to records $\mathbf{y}$ (0/1 for absence or presence of LPC), and $\mathbf{e}$ are errors. Distributional assumptions are $\mathbf{u} \sim N(0, \mathbf{A}\sigma_u^2)$, with $\mathbf{A}$ the numerator relationship matrix between animals, $\mathbf{e} \sim N(0, \mathbf{I}\sigma_e^2)$ and flat priors are assumed for μ, σ_u^2, and σ_e^2. For a direct genetic effects model, $\mathbf{Z}$ links observations to the polygenic effects of the observed individuals and σ_u^2 is the variance of direct genetic effects. For a maternal genetic effects model, $\mathbf{Z}$ links observations to the polygenic effect of the mothers of the observed individuals and σ_u^2 is the variance of (heritable) maternal effects. Estimates for variance components were the marginal posterior means of these parameters. Genetic evaluations were computed as BLUP, conditional on these estimates of variance components. Computations were performed by a Markov Chain Monte Carlo (MCMC) algorithm using the maGGic package (Janss 1997). Multiple MCMC chains of 27000 cycles were run, discarding the first 2000 cycles to allow for burn-in, until for each variance component a minimum of effectively 250 independent samples was obtained. Convergence of the multiple chains and the effective numbers of independent samples were determined by an Analysis of Variance procedure for dependent samples (Janss, 1997), similar to the procedure for independent samples used by Janss *et al.* (1997)

Prediction of new records. To evaluate predictive performance of the genetic models, regressions of the new observations on estimated breeding values were computed. Breeding values used were the breeding values of the sire and dam (u_s, u_d) of the observed animals, the pedigree index for the observed animal ($\frac{1}{2}u_s+\frac{1}{2}u_d$) and the breeding values of paternal grandsire and granddam and maternal grandsire and granddam (u_{Pgs}, u_{Pgd}, u_{Mgs}, u_{Mgd}). For the direct effects model, expected values for these regressions are ½ for the regressions on breeding values of parents, 1 for the regression on the pedigree index, and ¼ for the regressions on breeding values of grandparents. For the maternal effects model, regression on the breeding value of the mother should be 1, on the maternal grandparents ½, and on sire and paternal grandparents zero.

RESULTS AND DISCUSSION

Estimated variances and resulting heritabilities for the models with direct genetic effects and for the model with maternal genetic effects are in Table 1. For the maternal effects model, variance components were also computed for a model with maternal effects and additional direct effects

and for a model with maternal effects and a common environment effect for full sibs. In these additional models, variance components for direct effects and for common environmental effects were <1% of the total variance. Hence, in a maternal effects model, records appeared to be influenced solely by the (heritable) maternal effects.

Table 1. Estimated marginal posterior means of residual variance (σ_e^2), genetic variance (σ_u^2) and heritability (h^2), and posterior standard deviation (pSD) of h^2 in a direct effects model and in a maternal effects model

	Direct	Maternal
σ_e^2	0.069	0.099
σ_u^2	0.078	0.052
h^2	0.518	0.336
pSD h^2	0.194	0.092

Table 2. Regressions of new observations on breeding values of sire and dam (u_s, u_d) in a direct effects model and in a maternal effects model

	Direct	Maternal
$\frac{1}{2}u_s+\frac{1}{2}u_d$	-0.25	
u_d		1.10
u_s [a]	-0.20	-0.03
u_d [a]	0.43	1.10

[a] Regressions on these breeding values were simultaneously fitted.

Regressions of new observations on breeding values of sire and dam and on the pedigree index are in Table 2, for the direct effects model and for the maternal effects model. For the direct effects model, the regression on the pedigree index is negative. This indicates that use of the direct effects model would consistently predict wrongly. Separating the pedigree index in the breeding values of sire and dam, showed that this is due to the breeding value of the sire; the regression on the breeding value of the mother is around its expected value of 0.5. These results led to the hypothesis that records were more affected by the genotype of the mother and led to the investigations with a maternal effects model, as described above. Regression on the breeding values of the dam in a maternal effects model (Table 2) was positive and was around its expected value of 1, showing that the maternal effects model was indeed more appropriate. Regressions on both sire and dam breeding value in a maternal effects model showed that regression on the sire breeding value also was around its expected value of zero.

New observations were also fitted by regressions on the breeding values of grandparents (Table 3). For the direct effects model, this showed that the negative regression on the sire breeding appeared to be related to the breeding value of the paternal granddam.
On the maternal side, the positive regression on the breeding value of the dam appeared to be related to the breeding value of the maternal grandsire. Similar results, but more clearly, also showed from the maternal effects model. Striking in these results are the non-zero regressions on the breeding values of paternal grandsires and the too high absolute values of these regressions. These regressions on grandparents indicate that the inheritance of the trait investigated could also involve sex-linked or imprinted effects.

Table 3. Regressions of new observations on breeding values of paternal grandsire (u_{Pgs}) and granddam (u_{Pgd}) and maternal grandsire (u_{Mgs}) and granddam (u_{Mgd}) in a direct effects model and in a maternal effects model (all regressions were simultaneously fitted)

	Direct	Maternal
u_{Pgs}	0.46	1.17
u_{Pgd}	-0.30	-0.44
u_{Mgs}	1.00	1.07
u_{Mgd}	0.12	-0.06

CONCLUSIONS

The technique of studying regressions of new observations on breeding values proved to be very valuable to validate models and to formulate alternative models. In this study, a negative regression on the "direct effect" breeding values of sires led to the hypothesis of a maternal influence on the records. Regressions on breeding values of grandparents indicated a possibly more complicated mechanism and appear to be valuable information for further research. To set up genetic counselling programs for defects, this study showed that a preliminary critical investigation of various models will be required: here the direct effects model would have worked counterproductive for such a genetic counselling. Unfortunately this implies that a genetic counselling program can not easily *ad hoc* be implemented.

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