Simulation Study on Mendelian Sampling Variance Tests

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Abstract

To be able to assess the quality of the data sets and national evaluation models, Interbull and national evaluation centers need a validation test. Thus far, two Mendelian sampling variance test methods have been proposed: Mendelian sampling (IB4) and full model sampling (FMS) variance estimation methods, but neither has been implemented. The aim of this simulation study was to dissect the behaviour of both methods under two different scenarios for bulls and cows. Scenario A served as a control that should pass the test. For Scenario B, a yearly increase of 2% in phenotypic variance was generated. Without heterogeneous variance adjustment, it should fail the test. As an alternative, an analysis of MACE model residuals could be a simple tool to check the data quality. On average, a yearly increase of 1.9% and 1.4% in genetic variance were observed for cows and bulls in Scenario B without HV adjustment. The IB4 test performed well when applied to cows and it was able to detect the simulated heterogeneity in genetic variance. A yearly increase of 1.3% in the variance of MACE residuals was observed in Scenario B without HV adjustment. This was consistent with the genetic variance estimates for bulls, indicating that the analysis of MACE residuals could be utilized to check the data quality for bulls.

Introduction

National evaluation centers and Interbull need a Mendelian sampling variance test to assess the quality of the data sets and national evaluation models. The test should measure within-year estimates of genetic variance and detect all relevant deviations in it. So far, two different methods have been proposed: a method developed by Interbull (Fikse, 2003), hereinafter IB4, and a related method utilizing a full model sampling (Lidauer *et al.*, 2007), hereinafter FMS. They differ in the way they obtain the prediction error variances. However, the results were similar for both approaches (Lidauer *et al.*, 2007). None of them have been implemented yet. Based on the experiences with field data sets, the current tolerance interval in IB4 might be too stringent, whereas no test statistics is available for FMS.

This paper presents the first results of a simulation study designed to dissect the behaviour of the IB4 and FMS under two different scenarios. It also evaluates whether analysis of MACE model residuals could be a quick and simple tool to check the data quality.

Material and Methods

Existing methods

The IB4 method estimates genetic variance within a year by:

$$\sigma^2_{u_i} = \frac{1}{q_i}\sum_{k=1}^{q_i} d_k\left[\hat{m}_k^2 + PEV(\hat{m}_k)\right], \qquad (1)$$

where q_i is the number of animals in year i, d_k is the proportion of the genetic variance not explained by the known parents (i.e. 1/2, 3/4 or 1), $\hat{m}_k^2$ is the squared estimated Mendelian sampling deviation of animal k and $PEV(\hat{m}_k)$ is the prediction error variance of the Mendelian sampling deviation (Fikse, 2003). The 5% lower and 95% upper bounds of the tolerance interval are bootstrapped with a replacement using 1000 samples for each birth year.

In FMS, the genetic variance is estimated within a year i as:

$$\hat{\sigma}^2_{u_i} = \frac{\sum_{k=1}^{q_i} d_k \hat{m}_k^2}{q_i} \frac{1}{r} \sum_{j=1}^{r} \left[\frac{\sum_{k=1}^{q_i} d_k \tilde{m}_{kr}^2}{\sum_{k=1}^{q_i} d_k \hat{\tilde{m}}_{kr}^2} \right], \qquad (2)$$

where $\hat{m}_k$ is the Mendelian sampling deviation estimated from the real data set, $\tilde{m}_{kr}$ is a simulated true Mendelian sampling deviation of replicate *r*, and $\hat{\tilde{m}}_{kr}$ is its estimate solved from the data replicate *r* (Lidauer *et al.*, 2007). Only one replicate was used in this study.

Analysis of MACE model residuals

Because the data in MACE analysis is from deregression, the model residuals should be strongly associated with the Mendelian sampling terms. Based on our hypothesis, provided there exists a trend in within-year estimates of genetic variance for some country, there should be a similar trend in a within-year variance of the MACE model residuals as well. Therefore, relative changes in the within-year variances were compared.

Real data

We used Danish Holstein test-day data for protein yield to study the methods. The sample comprised 2000 herds, 756 537 cows and 13 million test-day records within a 20-year time interval. The pedigree included 1.7 million animals. The data set represents a medium size national population. Breeding values (EBVs) were predicted under the Nordic test-day model for the first three lactations and combined 305-d EBVs were constructed weighting 1st, 2nd, and 3rd lactations by 0.5, 0.3, and 0.2, respectively.

Simulations

Observations of the real data were replaced by simulated observations. Two scenarios, each with 20 replicates (Table 1), were simulated based on variance components and systematic environmental effects from the Nordic test-day model (Lidauer *et al.*, 2006). MiX99 software was used for all analyses in this study (Lidauer *et al.*, 2011).

Scenario A served as a control without variance heterogeneity that should pass the validation test. For Scenario B, a yearly trend of 2% in phenotypic variance was generated. Thus, the ratio between genetic and phenotypic variance remained constant. It was anticipated that a yearly increase of 2% in genetic variance would cause a bias of about 0.1 genetic standard deviation between EBVs of young and proven sires and should therefore be detected by the validation method. Provided no heterogeneous variance (HV) adjustment is carried out, the validation test should fail, whereas when an adequate HV adjustment is applied, the data set should pass the test.

For each scenario and each simulated replicate, IB4 was used as a validation test and FMS to estimate the possible trend in genetic variances over birth years. Analyses were performed for bulls and cows in each testing scheme. Tested bulls had daughters with records at least in 10 herds and were born within years ranging from 1986 to 2006. The average group size was 291. Tested cows were born after 1987. The average size of the birth year groups was 41729, with the smallest group size of 2 334 in 2009.

To analyze MACE model residuals (AMR), EBVs for sires were deregressed, and these were used to fit a classical – although a single trait – MACE model. Residuals were scaled by the square root of the applied weights.

Table 1. Setup for simulations.

EBV Prediction	Scenarios	
	A) Control	B) Yearly trend of 2% in phenotypic variance
BLUP[a]	IB4[c]	IB4
	FMS[c]	FMS
	AMR[c]	AMR
BLUP+HV[b]	-	IB4
	-	FMS
	-	AMR

[a]EBV predictions are carried out without heterogeneous variance (HV) adjustment

[b]EBV predictions are carried out with HV adjustment

[c]IB4: Interbull test IV; FMS: full model sampling; AMR: analysis of MACE model residuals

Results and Discussion

Figures 1-6 show the within-years genetic variance in the simulated data sets for each testing scheme. On average, a yearly increase of 1.9% and 1.4% in genetic variance were observed for cows and bulls in Scenario B without HV adjustment. As expected, the variation between replicates in the cow data was low. However, a rather high variation in the estimates of genetic variance was observed in the last birth year group, where cows had records from the first lactation only, deteriorating the accuracy of the estimates of combined EBVs in that group. Further, in Scenario B in cows, estimates of the last birth year were on average smaller than those of the second last birth year. The lack of observations in later lactations and thus the lack of information on the increase in phenotypic variance may explain the finding. The variation in the bull replicates was relatively high, suggesting that more than one replicate should have been used in the last part of the FMS equation for bulls.

The IB4 test performed well when applied to cows. The cow data replicates passed the validation test when expected and failed when the 2% trend in phenotypic variance was generated, but not accounted for (Table 2). When applied to bulls, the IB4 test performed well for Scenario A and Scenario B with the HV adjustment applied. However, only 7 of 20 replicates failed for Scenario B without HV adjustment.

For each scenario, the variance of MACE residuals was plotted over bulls' birth years (Figures 7-9) and compared to those of genetic variance (Figures 4-6). On average, a yearly increase of 1.3% was observed in Scenario B without HV adjustment, which corresponded to a yearly increase of 1.4% in genetic variance. Thus, relative changes were in a good accordance.

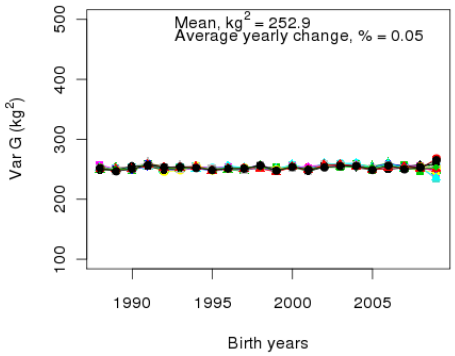

Figure 1. Genetic variance of cows over birth years in 20 replicates for Scenario A. Estimated with the FMS method.

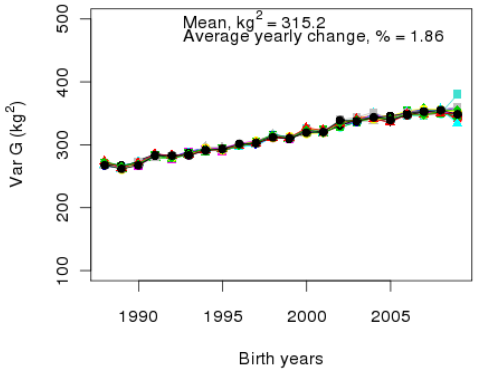

Figure 2. Genetic variance of cows over birth years in 20 replicates for Scenario B, no HV adjustment. Estimated with the FMS method.

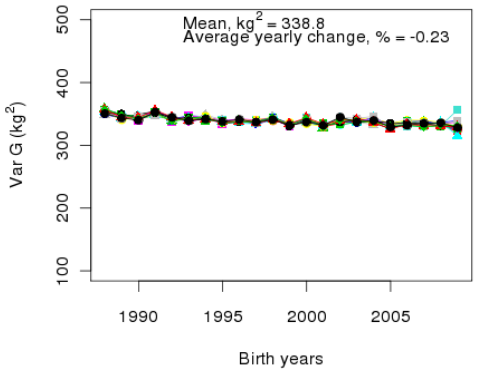

Figure 3. Genetic variance of cows over birth years in 20 replicates for Scenario B, HV adjustment carried out. Estimated with the FMS method.

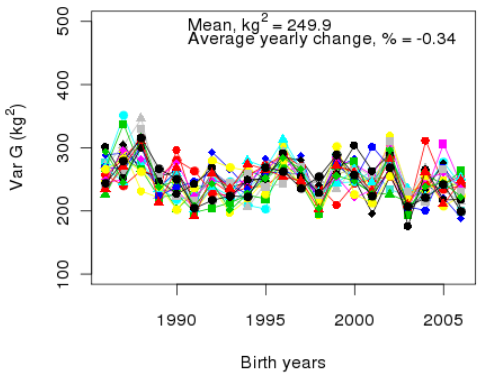

Figure 4. Genetic variance of bulls over birth years in 20 replicates for Scenario A. Estimated with the FMS method.

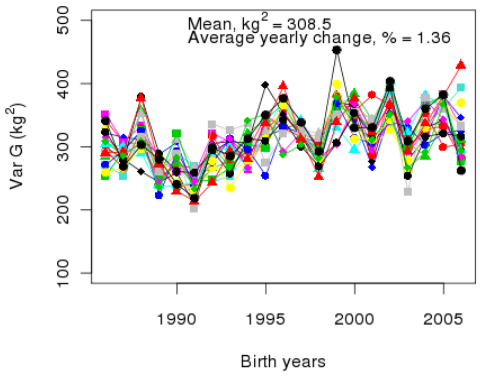

Figure 5. Genetic variance of bulls over birth years in 20 replicates for Scenario B, no HV adjustment. Estimated with the FMS method.

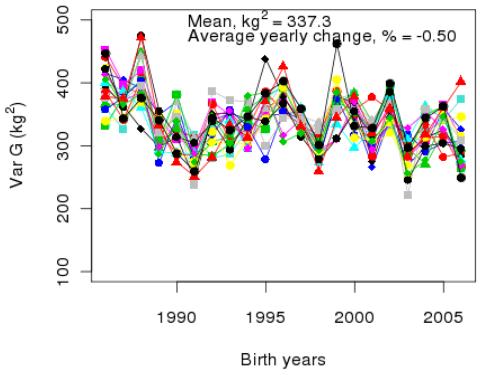

Figure 6. Genetic variance of bulls over birth years in 20 replicates for Scenario B, HV adjustment carried out. Estimated with the FMS method.

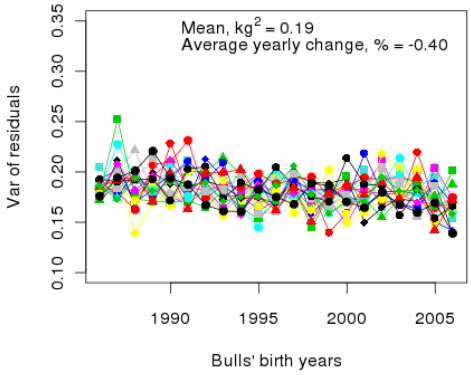

Figure 7. Variance of MACE residuals over birth years in 20 replicates for Scenario A. Estimated with the FMS method.

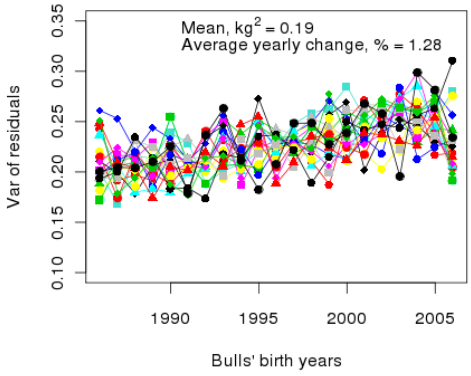

Figure 8. Variance of MACE residuals over birth years in 20 replicates for Scenario B, no HV adjustment. Estimated with the FMS method.

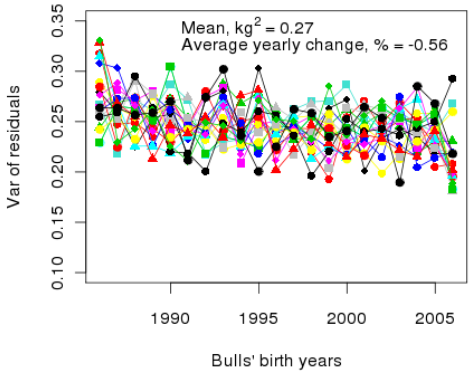

Figure 9. Variance of MACE residuals over birth years in 20 replicates for Scenario B, HV adjustment carried out. Estimated with the FMS method.

Conclusions

The IB4 test was most reliable when applied on cows and it was able to detect the simulated heterogeneity in genetic variance. The most recent year groups in cows should be excluded from the test since they have no records on later lactations. Analysis of MACE model residuals could serve as a quick and simple preliminary tool for Interbull to verify the quality of the input data.

Acknowledgements

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Table 2. Test results for the IB4 validation method.

A scenario				**B scenario, no HV**				**B scenario, HV**			
Cows		**Bulls**		**Cows**		**Bulls**		**Cows**		**Bulls**	
Years outside tolerance interval											
Out[a]	Prop, %[b]	Out[a]	Prop, %[b]	Out[a]	Prop, %[b]	Out[a]	Prop, %[b]	Out[a]	Prop, %[b]	Out[a]	Prop, %[b]
0	100	0	30	12	15	1	15	0	100	0	30
		1	35	13	45	2	50			1	50
		2	30	14	35	4	20			2	15
		3	5	15	5	5	10			3	5
Number of failed replicates (> 2 years outside tolerance interval)											
0/20		1/20		20/20		7/20		0/20		1/20	

[a]Number of birth years outside the tolerance interval in a replicate
[b]Proportion of the replicates in a class Out