Validating Genomic Reliabilities and Gains from Phenotypic Updates

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Summary

Reliability can be validated from the variance of the difference of earlier and later estimated breeding values as a fraction of the genetic variance. This new method avoids using squared correlations that can be biased downward by selection. Published genomic reliabilities of U.S. young bulls agreed closely with validation formulas. Similar formulas based on reliability differences can estimate potential gains from more frequent phenotypic updates such as monthly or weekly updates, but those gains are small when young animal reliabilities are high.

Key words: reliability, genomic prediction, validation, phenotypic update

Introduction

Genomic validation currently tests if estimated breeding values (EBVs) are unbiased and accurate but does not test if published reliabilities (RELs) are accurate. The standard Interbull test (Mäntysaari *et al.*, 2010) computes regressions of later daughter deviations or deregressed proofs on earlier genomic EBVs to test if observed regressions are close to the expected value, which is 1.0 if all bulls in the validation group were genotyped or <1.0 if bulls were selected for genotyping after progeny test. Squared correlations of EBVs and parent averages with later data are also compared to test if genomic predictions are more accurate than conventional predictions.

Because breeders see EBVs, tests using later EBVs instead of deregressed proofs could be more convincing and easier to understand in genomic validation. A similar test could compare how well differences between earlier and later published RELs match the observed variance of EBV changes. The math is very simple, and the observed REL is easy to compute for any trait using a few assumptions. In April 2017, Interbull provided snp_blup_rel as a standard program to compute genomic RELs. The following math may provide a standard way to validate if later actual EBV changes agree with expected variances or if theoretical RELs need to be discounted as discussed by Calus *et al*. (2010) and Liu *et al.* (2017). Thus, snp_blup_rel software can provide individual RELs, and the math that follows can help with general scaling of those RELs.

Methods

Selection can have large effects on variance such that Var(EBV) no longer equals REL × Var(BV), where BV is true breeding value, but prediction error variances (PEVs) should still match the expected formula (Henderson, 1982; Gorjanc *et al.*, 2015):

$$\begin{aligned} PEV &= Var(EBV - BV) \\ &= (1 - REL) \times Var(BV). \end{aligned}$$

Similarly, differences between later and earlier EBVs should be independent of the earlier EBV regardless of the genomic pre-selection practiced so that expected regression of later on earlier EBV is 1.0 and variance of the EBV difference is proportional to the difference in RELs. If EBV_1 and EBV_2 are earlier and later genomic evaluations with reliabilities REL_1 and REL_2, respectively, then

$$\begin{aligned} &Var(EBV_2 - EBV_1) \\ &\quad = (REL_2 - REL_1) \times Var(BV). \end{aligned} \quad (1)$$

Reliability Validation

Fairly accurate and high estimates of REL_2 are available for most traits and countries because most validation bulls were marketed extensively as young bulls and now have many daughters. Thus, we can assume REL_2 is known and simply solve for REL_1 by plugging REL_2 into Equation 1:

$$REL_1 = REL_2 - \frac{Var(EBV_2 - EBV_1)}{Var(BV)}.$$

This math implies that observed prediction REL equals final REL minus variance of observed genomic EBV changes as a fraction of genetic variance. Similar math is used in the Interbull Verify program to list bulls that changed more than expected based on REL gain, but the direction of math is now reversed to check if REL is correct given the overall standard deviation (SD) of bull changes.

Published REL_1 from April 2014 were compared with corresponding observed REL_1 for U.S. validation bulls born before 2013 that had no daughters in April 2014 but had >100 daughters in April 2017. Interbull estimates of genetic SD were used because those are easily available and re-estimated twice per year; the U.S. SD(TA) from file proddoc1704r.itb was 17.5 pounds for Holstein protein, for example.

Calculation Frequency

Math very similar to that for REL validation can estimate REL gains or losses and genetic gain if phenotypic updates are more or less frequent. In many countries, genomic predictions for newly genotyped animals are computed using previous allele effect estimates without updating phenotypic data. For example, genomic evaluations may be computed weekly, but phenotypes are updated only every 4 months (Wiggans *et al.*, 2015). If REL increases steadily from REL_1 to REL_2 across a year, REL gain from n updates per year (REL_n) instead of 1 annual update is demonstrated in Figure 1 and should average

$$REL_n = 0.5(REL_2 - REL_1)(n - 1)/n.$$

Results and Discussion

For validation bulls, average published REL_1 was 0.76, REL_2 was 0.95, and SD of change was 8.4 for Holstein protein yield. Observed REL_1 was calculated as

$$REL_1 = 0.95 - (8.4)^2/(17.5)^2 = 0.72,$$

which is a little less than the 0.76 published officially in 2014. Results for other traits show an average overestimation of 2 percentage points for Holstein REL_1 (Table 1) and an average underestimation of 3 percentage points for Jersey REL_1 (Table 2). The REL can be adjusted using methods of Liu *et al.* (2017).

Expected young bull RELs have also been estimated using nonlinear formulas based on size of domestic and foreign reference populations (Sullivan and Jakobsen, 2014). For example, their table 2 listed U.S. Holstein REL_1 of 0.76 published versus 0.75 expected for protein and 0.72 published versus 0.73 expected for somatic cell score. Thus, REL_1 published by the United States was very close to that expected from reference population size, and most other countries had similar published REL_1 that should match well to their observed REL_1. To increase the power of the REL_1 test, countries with small populations might pool EBV changes across different year groups. For example, EBVs of bulls that were young 2, 4, 6, or 8 years ago could be compared with their current EBVs instead of using only one truncation time.

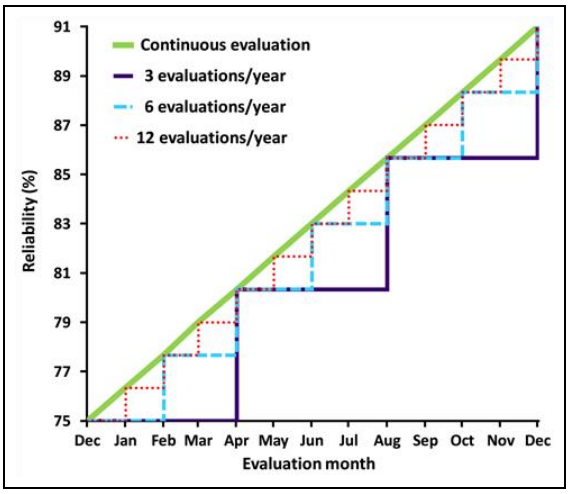

Figure 1. Reliability gains from more frequent evaluation.

Table 1. Observed and published RELs from April 2014 for 2,202 U.S. Holstein validation bulls[1]

Trait	SD(TA)	SD(PTA_2 – PTA_1)	REL_2 (%) published	REL_1 (%)		
				Published	Observed	Difference
Net merit (US$)	193	89.5	90	73	68	–5
Milk (pounds)	670	324	95	76	72	–4
Fat (pounds)	24.9	11.4	95	76	74	–2
Protein (pounds)	17.5	8.4	95	76	72	–4
Productive life (months)	2.30	1.05	86	70	65	–5
Somatic cell sore	0.21	0.08	91	73	77	+4
Daughter pregnancy rate (%)	2.32	0.89	84	68	69	+1
Average	…	…	91	73	71	–2

[1]REL = reliability, SD = standard deviation, TA = transmitting ability; PTA = predicted transmitting ability; REL_1 = REL from earlier genomic evaluation; REL_2 = REL from later genomic evaluation

Table 2. Observed and published RELs from April 2014 for 253 U.S. Jersey validation bulls[1]

Trait	SD(TA)	SD(PTA_2 – PTA_1)	REL_2 (%) published	REL_1 (%)		
				Published	Observed	Difference
Net merit (US$)	193	90.8	90	64	68	+4
Milk (pounds)	617	293	96	68	73	+5
Fat (pounds)	24.5	11.9	96	68	72	+4
Protein (pounds)	18.4	9.2	96	68	71	+3
Productive life (months)	2.45	1.47	83	55	47	–8
Somatic cell sore	0.19	0.10	92	62	64	+2
Daughter pregnancy rate (%)	2.61	1.18	83	52	63	+11
Average	…	…	91	62	65	+3

[1]REL = reliability, SD = standard deviation, TA = transmitting ability; PTA = predicted transmitting ability; REL_1 = REL from earlier genomic evaluation; REL_2 = REL from later genomic evaluation

Table 3. Average RELs (%) by frequency of phenotypic updates (number of updates per year in parentheses) for young bulls (1 year old) or transitioning to progeny-tested (4 years old)

	Phenotypic update frequency								
Bull status	Annual (1)	6 months (2)	4 months (3)	3 months (4)	2 months (6)	Monthly (12)	Weekly (52)	Daily (365)	Instant (∞)
Progeny-tested	75.0	79.0	80.3	81.0	81.6	82.3	82.80	82.97	83.0
Young	73.0	73.5	73.7	73.8	73.83	73.92	73.98	73.99	74.0

This method of validating genomic REL requires little work and no daughter yield deviations or deregressed proofs and can also be applied to single-step models where only genomic EBVs are available and not conventional EBVs. Some further theory may be needed on what happens if assumptions are violated (such as regressions that differ from 1.0). The formula could also be applied to the data simulated with and without selection to verify theoretical reliability (Calus *et al.*, 2015), and differences in REL can also be tested through simulation by removing some SNPs from actual data and treating those as QTLs (Calus *et al.*, 2009).

Calculation Frequency

Suppose bulls increase REL from 75% (REL_1) to 91% (REL_2) during the year that they transition from no daughters to many daughters with records. Corresponding values of REL_n for differing frequencies of update are in Table 3. Minimum gain is 0 percentage points with an annual update because the bulls would stay at 75% for the whole year, and maximum gain is 8 percentage points with instant updating because bulls would average (75 + 91)/2 = 83% during that year. Average REL is 80.3% with the current frequency of 3 updates per year but could increase to 82.3% with monthly updates

Table 4. Average RELs (%) by age group for males in artificial-insemination service and for females

	Age group (years)							
Sex	<1	1	2	3	4	5	6	7
Males	72.5	73.1	75.5	76.0	90.6	91.6	92.0	92.0
Females	70.4	71.4	73.0	73.5	73.8	73.6	73.7	74.6

or decrease to 79.0% with 2 updates per year. Young bull RELs are also provided in Table 3, but their gains are small for any update frequency because their RELs increase only about 1-2 percentage points per year for the first 3 years.

Current average REL by age is shown in Table 4. Bulls averaged 72.5% when <1 year old, 73.1% for yearlings, and 75.5% for bulls 2 years old when maternal sibs are phenotyped and the bulls' sires are progeny tested. Average REL for females is a little lower because some have incomplete pedigrees and because REL increases little when a cow's own phenotypes are added. Phenotypic update frequency was much more important with conventional selection on progeny-tested bulls (Misztal, 1993) because REL_1 was much lower at about 35%. With genomics, the update frequency is less important because young bull REL is higher and does not change much during the selection period.

Breeding companies and researchers may be more concerned about accurately estimating the average REL of a trait for investment decisions than farmers are about individual bull RELs. For example, decisions on funding data collection for new traits, frequency of phenotypic updates, and major changes to evaluation models all may depend on the expected RELs of evaluations.

Conclusions

Validation of genomic RELs can be fairly simple given earlier initial RELs that need to be tested and later higher RELs that can be assumed to be correct. The RELs should be calculated using prediction error variances and differences between EBVs because EBV variances are reduced by selection. Published RELs were slightly too high (2 percentage points) for U.S. Holsteins but slightly too low (3 percentage points) for U.S. Jerseys. The value of more frequent phenotypic updates can be estimated using similar math. Updates more frequent than 3 times per year have small benefits when initial genomic REL is high.

References

Calus, M., VanRaden, P., Lidauer, M., Ducrocq, V., Liu, Z. & Harris, B. 2015. Working group on genomic reliability. *Interbull Technical Workshop* presentation, Walsrode, Germany. http://www.interbull.org/static/web/2_1_Harris.pdf.

Calus, M.P.L., Mulder, H.A., Verbyla, K. & Veerkamp, R.F. 2009. Estimating reliabilities of genomic breeding values. *Interbull Bulletin 40*, 198–201.

Calus, M.P.L., Mulder, H.A. & Veerkamp, R.F. 2010. Comparison of reliabilities of direct genomic values. *Interbull Bulletin 41*, 25–28.

Gorjanc, G., Bijma, P. & Hickey, J.M. 2015. Reliability of pedigree-based and genomic evaluations in selected populations. *Genetics Selection Evolution 47*, 65.

Henderson, C.R. 1982. Best linear unbiased prediction in populations that have undergone selection, in: Barton, R.A., Smith, W.C. (Eds.). *Proceedings of the World Congress on Sheep and Beef Cattle Breeding*, Dunmore Press, Palmerston North, N.Z., vol. 1, pp. 191–200.

Liu, Z., VanRaden, P.M., Lidauer, M., Calus, M.P., Benhajali, H., Jorjani, H. & Ducrocq, V. 2017. Approximating genomic reliabilities for national genomic evaluation. *Interbull Bulletin*, in press.

Mäntysaari, E., Liu, Z. & VanRaden, P. 2010. Interbull validation test for genomic evaluations. *Interbull Bulletin 41*, 17–22.

Misztal, I. 1993. Technical considerations in implementation of continuous genetic evaluation, in: Misztal, I. (Ed.), *Proceedings of the Symposium on Continuous Genetic Evaluation in Dairy Cattle*, College Park, MD, pp. 29–39.

Sullivan, P.G. & Jakobsen, J.H. 2014. GMACE pilot #4: Adjusting the national reliability input data. *Interbull Bulletin 48*, 40–45.

Wiggans, G.R., VanRaden, P.M. & Cooper, T.A. 2015. *Technical note:* Rapid calculation of genomic evaluations for new animals. *Journal of Dairy Science 98*, 2039–2042.