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Residual effects of genotype call rate on genomic breeding values in a commercial Atlantic salmon population

W. Brand[1*], S.A. Boison[1], P. Sae-Lim[1], and M. Baranski[1]

[1]*Mowi Genetics AS, Bergen, Norway;* [*]*Corresponding author: werner.brand@mowi.com*

Introduction

In large-scale commercial SNP array genotyping analysis pipelines, standard operating procedures typically include quality control (QC) filters based on a minimum per-sample SNP call rate (CR) threshold. In practice, some samples may fall slightly below the widely applied standard threshold of ≥97% call rate, as this metric is influenced by variability arising from the instrument, DNA template quality, and other technical factors. To maximize the size of the reference population and preserve these specific candidates, protocols are often employed where samples with lower call rates (e.g., 90% ≤ CR < 97% and reduced heterozygosity thresholds) are retained and subsequently imputed to 100% (Browning and Browning, 2009; Hickey et al., 2012; Cooper et al., 2013; Purfield et al., 2016).

It is generally assumed that after imputation, these rescued samples are fully informative for genomic evaluation (Browning and Browning, 2009). However, the residual impact of original pre-imputation sample quality on the distribution of GEBV is poorly characterized. This study evaluates the sensitivity of GEBV variance to specific QC filtering thresholds, focusing on the potential for persistent bias when thresholds are not fully optimized.

Materials & Methods

Data consisted of 48,645 Atlantic salmon (*Salmo salar*) from the 2023 smolt year class within the Mowi Norway breeding program. The breeding structure followed a partial factorial mating design anchored by 250 high-quality parents used for parentage assignment and genomic evaluation. Animals were genotyped across three distinct Thermo Fisher Axiom SNP arrays. After merging, the final marker set comprised 66,515 SNPs, with approximately 45,000 markers overlapping across all platforms. The QC was performed based on pre-merge CR to isolate sample-specific noise from platform-induced missingness. All genotypes were imputed to 100% using Beagle 5.4 (Browning et al., 2018).

To isolate the driver of the observed variance collapse, three scenarios were investigated. Firstly, the observed set included all animals passing a lenient 90% CR threshold. Secondly, a High-Quality (HQ) control set was established using only candidates with CR ≥99.5% and informants with CR ≥95%. Finally, a masked trial was created by taking HQ genotypes and randomly masking from 0% to 10% additional missing rate. All sets were processed through the same imputation and evaluation pipeline to compare GEBV stability and Mendelian consistency, using Plink 1.9 (Purcell at al., 2007).

Growth, pigment and cardiomyopathy syndrome were analyzed using a multivariate animal mixed model to estimate GEBV. Because the GEBV trends for these traits were similar, only the results for growth were reported. Multivariate GBLUP was used to estimate GEBV for growth using the standard animal model. The genomic relationship matrix (**G**) was constructed using VanRaden method 1 (VanRaden, 2008). To ensure comparability across scenarios, the genomic estimated breeding values (GEBV) for growth among candidates were standardized (Z) by subtracting the mean breeding value of informants and dividing by the square root of the additive genetic variance. Realized group accuracy was quantified as the standard deviation of standardized GEBV (SD_Z) within call rate bins. Biological noise was quantified using Mendelian error rates (MER). MER is calculated by identifying cases where the genotype of

an offspring is biologically impossible given the genotypes of its parents. The MER was correlated with original CR to identify the inflection point where allelic noise deviates from Mendelian inheritance patterns.

Results & Discussion

The genomic evaluation revealed a systematic heteroscedasticity in GEBV distributions linked to pre-imputation call rates. In the observed dataset (Figure 1), individuals with higher missing rates (lower call rates) displayed a pronounced "sideways funnel" effect, where GEBV variance were regressed toward the population mean. Specifically, candidates with >6% missingness were effectively excluded from the 95th percentile (top selection tail), indicating a non-genetic selection bias.

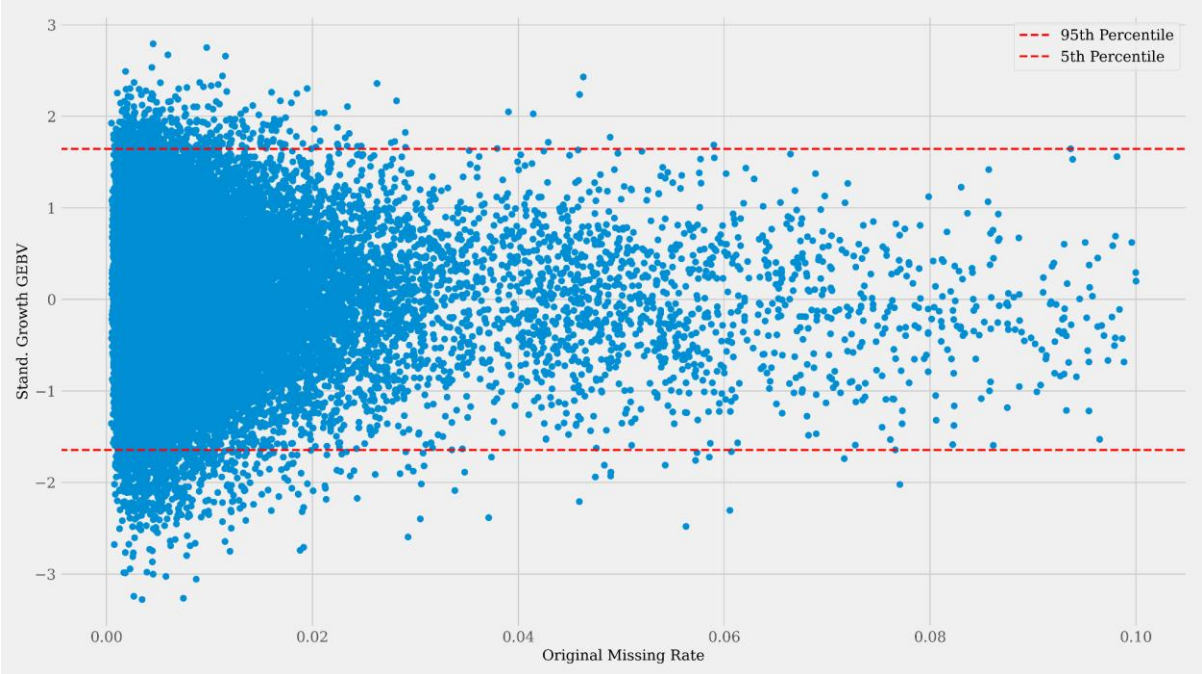

Figure 1. Relationship between pre-imputation missing rate and the distribution of standardized growth GEBV. Dashed lines represent the 5th and 95th percentiles of the high-quality population.

To determine if this variance shrinkage was a mathematical artifact of imputing missing markers, we compared the HQ control set against a masked trial where up to 10% of markers were randomly removed and subsequently imputed. The Pearson correlation between HQ and masked GEBV was nearly unity (r=0.9996). Unlike the observed set, the masked trial maintained stable, homoscedastic GEBV variance across the entire range of missingness (Figure 2), definitively ruling out the imputation of missing data as the source of bias.

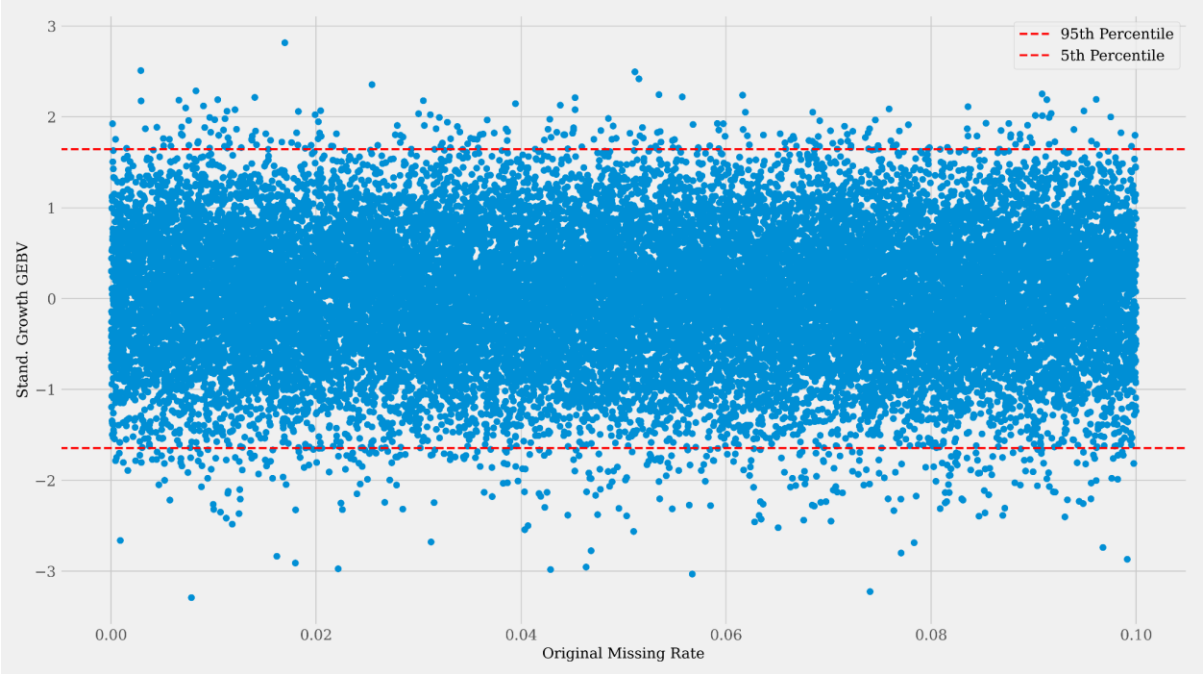

Figure 2. GEBV distribution for the 'masked' sensitivity trial. High-quality genotypes (≥99.5% CR) were stochastically masked to match the missingness distribution of the Observed set. The stability of the 5th and 95th percentiles confirms that imputation successfully preserves GEBV variance in the absence of allelic noise.

The biological driver of the variance shrinkage was identified through Mendelian inconsistency analysis. In the observed set, the number of Mendelian errors increased sharply with increasing missing rate, with a notable point of change at approximately 4%–6% missing rate (Figure 3).

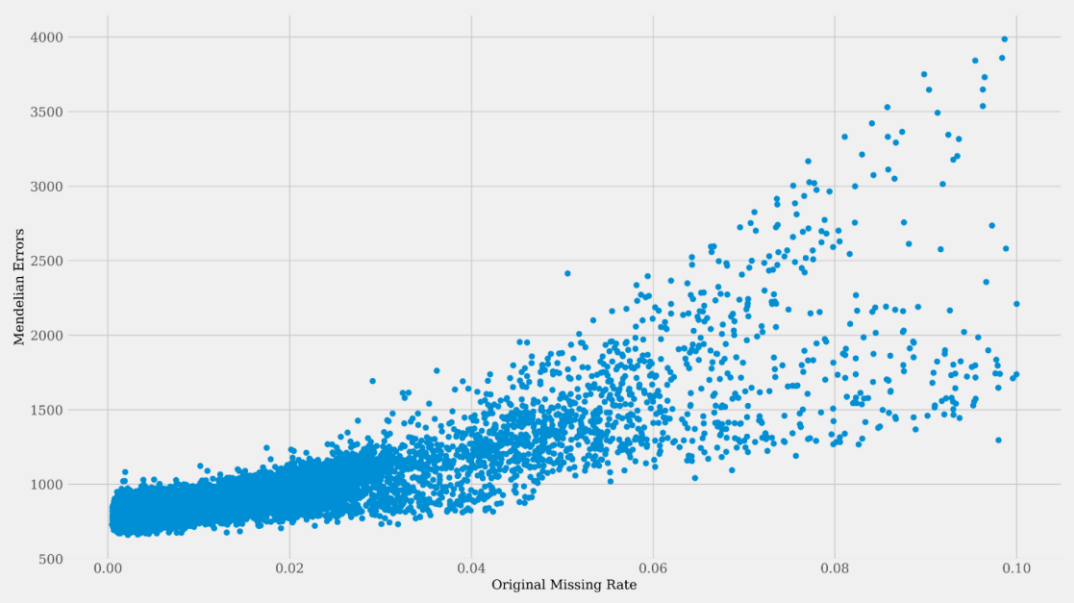

Figure 3. Relationship between pre-imputation missing rate (x-axis) and Mendelian error rate (MER) in the observed dataset (y-axis).

The masked trial showed no increase in errors despite high missingness. The spike in the observed set confirms the presence of significant allelic noise in low-call-rate samples. The

GBLUP model detects this noise as uncertainty and regresses the individual's GEBV toward the mean to minimize prediction error variance. Consequently, elite candidates with poor genotype quality are systematically "hidden" in the middle of the distribution, leading to a direct loss of potential genetic gain.

Table 1. Comparison of GEBV distribution and Mendelian consistency across quality scenarios.

Scenario	Sample Subset (CR)	Mean MER	SD of Std. GEBV	Correlation to HQ (r)
HQ control	≥99.5%	~672	0.73	1.00
Masked		~639	0.73	0.9967
Observed	>97%	~825	0.74	-
	94% - 97%	~1278	0.72	-
	90% - 94%	~1924	0.68	-

As shown in Table 1, the realized accuracy - represented by the standard deviation of standardized GEBV (SD_Z) - dropped from 0.73 in the HQ/masked sets to 0.68 in the lowest-quality observed bin. This 6.8% reduction in SD_Z correlates with a nearly three-fold increase in Mendelian errors, indicating that the GBLUP model regresses these individuals toward the mean to minimize prediction error variance caused by genotype noise. The most significant finding for commercial breeding is the effective exclusion of low-CR individuals from the top selection percentiles. Superior genetic candidates (the "top tail") who happen to have poor genotype quality are systematically hidden within the population average. Because these candidates are never selected, the program suffers a direct, unrecoverable loss in achievable genetic gain.

Conclusions

Imputation is an effective tool for handling missing marker data but is an ineffective remedy for low genotyping call rate. Our analyses show that low call rate samples carry a high burden of Mendelian inconsistencies, indicating incorrectly called alleles rather than simply more missing data. Under this scenario, alternative imputation algorithms are unlikely to recover the correct genotypes, because they operate on noisy input rather than on truly missing markers. To safeguard the integrity of genomic evaluations and maximize selection response, we therefore recommend a stricter QC threshold (e.g., ≥97% CR); retaining samples below this threshold under "rescue" protocols primarily introduces allelic noise and can undermine the identification of elite candidates.

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