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Selection on polygenic traits reduce the genetic variance considerably more due to the bulmer effect than typically assumed

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Introduction

Genetic variance is essential for artificial selection (Falconer and Mackay, 1996; Lynch and Walsh, 1998; Walsh and Lynch, 2018), but selection can reduce the genetic variance in a population. Part of this loss is a result of the creation of negative covariances between effects at different loci that affect a trait, known as the Bulmer effect (Bulmer, 1971; Bulmer, 1980). This loss in genetic variance is transient and disappears when selection has stopped.

Generally, the assumptions of the infinitesimal model, being that a large number of independently segregating loci affect a trait, are used to estimate breeding values and variance components. Under the infinitesimal model, a Bulmer equilibrium is reached within 3 to 5 generations in which approximately 5 to 25% of the genetic variance is lost (Bulmer, 1971; Dekkers, 1992; Walsh and Lynch, 2018). However, in actual populations, part of the loci are linked because loci are located on chromosomes. When the number of loci underlying complex traits is relatively limited and the number of chromosomes is high (i.e., above 10), the expected impact of linkage on the Bulmer effect is negligible (Bulmer, 1974). However, nowadays, complex trait are expected to be influenced by a large number of causal loci (Wientjes *et al.*, 2024). This all changes the potential impact of the Bulmer effect on the genetic variance in a selected population. Therefore, the aim of this research was to investigate the impact of the deviation from the infinitesimal model on the genetic variance in livestock populations under selection.

Materials & Methods

Theory. We will assume that loci act additively on a trait, and will indicate the variance in a selected group of individuals with *. Moreover, we will assume that selection is based on the phenotype (i.e., mass selection). In this case, the change in phenotypic variance among the selected parents in generation *t* can be written as:

$$\Delta V_t = \sigma^{2*}_{P,t} - \sigma^2_{P,t} = -\kappa\sigma^2_{P,t} \quad (1)$$

where κ is the proportional reduction in the phenotypic variance due to selection of the parents which depends on the selection intensity, i.e. $\kappa = i(i - x)$, where i is the intensity of truncation selection and x is the standardized truncation point (Falconer and Mackay 1996; Walsh and Lynch 2018). It can then be derived that the additive genetic variance among selected individuals in generation t ($\sigma^{2*}_{A,t}$) can be written as:

$$\sigma^{2*}_{A,t} = \sigma^2_{A,t}(1 - h^2_t\kappa) = \sigma^2_{A,t} - \sigma^2_{P,t}h^2_t h^2_t\kappa = \sigma^2_{A,t} + h^4_t\Delta V_t \quad (2)$$

where h^2_t is the heritability of the trait in generation t.

This indicates that the disequilibrium contribution in generation 1 (d_1), being the change in genetic variance due to negative covariances between loci introduced by selection, is:

$$d_1 = \sigma^{2*}_{A,1} - \sigma^2_{A,0} = h^4_1\Delta V_1 \quad (3)$$

where $\sigma^2_{A,0}$ is the genetic variance in the unselected base population.

Every generation, half of the new disequilibrium contribution buildup in the parents' generation due to selection is preserved. Moreover, recombination breaks down a part of the old

disequilibrium contribution present in the previous generation. In the absence of linkage, 50% is broken down by recombination. In the presence of linkage, this is approximately equal to the harmonic mean of the recombination rate (r_H). Hence, the disequilibrium contribution in generation t+1 can be described as (Bulmer 1974; Walsh and Lynch 2018):

$$d_{t+1} = r_H d_t + \frac{1}{2} h_t^4 \Delta V_t \quad (4)$$

The parameter r_H can be estimated as the harmonic mean of the pairwise recombination rates, which in turn are estimated using Haldane's mapping function (Haldane, 1919). The equilibrium value of the disequilibrium contribution can be estimated as (Walsh and Lynch, 2018):

$$\tilde{d} = (\gamma - h^2)\sigma_{P,0}^2 \quad (5)$$

where γ is calculated as:

$$\gamma = r_H \left(\frac{2h^2 - 1 + \sqrt{1 + 2h^2(1-h^2)\kappa / r_H}}{2r_H + \kappa} \right) \quad (6)$$

Expected impact. We estimated the average value of r_H for different genome structures. We investigated a genome of 30 Morgan with 1, 5, 10, 15 or 30 chromosomes, and genomes of 15 chromosomes with a genome length of 12, 18, 24 or 30 Morgan. The number of causal loci underlying the trait ranged from 30 to 30,000, and the causal loci were either assumed to be equally spread along the genome or randomly spread.

Simulation. We validated the theory using simulations. We simulated a population using QMSim (Sargolzaei and Schenkel, 2009) and our own developed Fortran program, which represented a livestock population in terms of linkage disequilibrium and allele frequency pattern. The population was under 50 generations of either mass or GBLUP selection with a selected proportion of 20%, followed by 500 generations of random selection. The set-up of the simulations was based on our previous studies (Wientjes *et al.*, 2022; Wientjes *et al.*, 2023), but we now focussed only on additive traits with a heritability of 0.4. In short, the simulated trait was influenced by 2000 causal loci, 20,000 markers were used for genomic selection, and new mutations were added following a mutational variance of ~$0.001\sigma_e^2$. We used two genome compositions; 1) No Linkage, assuming all causal loci and markers were placed on a separate chromosome, and 2) Linkage, assuming 10 chromosomes of 1 Morgan length. We tracked the true genetic variance, being the variance in true breeding values, and the genic variance calculated as $\sum 2p_i(1-p_i)\alpha_i^2$, where p_i is the allele frequency and α_i is the allele substitution effect at causal locus i. All simulated scenarios were replicated 20 times, and results show the mean values across replicates.

Results

Harmonic mean of recombination rate. The value for r_H was to a very limited extent influenced by the number of chromosomes, and heavily influenced by the genome size (Table 1). When a relatively limited number of loci (≤300) was equally spread on a rather large genome (≥18 Morgan), r_H was in the range of 0.45-0.5. When loci were randomly spread along the genome, r_H was considerably lower. For a relatively limited number of loci (≤300) that were randomly spread along a large genome (30M), r_H dropped to 0.37. The r_H dropped further when the number of causal loci increased or the genome size decreased, resulting in an r_H of 0.22 when the number of causal loci was 30,000 and the genome size 12M. For livestock populations, the genome is ~20 (pigs) to ~30 (cattle and chicken) Morgan and thousands of loci are expected to underlie complex traits, resulting in an expected r_H around 0.3.

Table 1. Impact of number of chromosomes, genome size, number of causal loci and their distribution along the genome on the harmonic mean of the recombination rate (r_H).

	Number of causal loci							
	Loci equally spread				Loci randomly spread[1]			
	30	**300**	**3,000**	**30,000**	**30**	**300**	**3,000**	**30,000**
Genome size of 30M; Number of chromosomes:								
1	0.49	0.47	0.44	0.41	0.42	0.38	0.35	0.35
5	0.50	0.47	0.44	0.41	0.42	0.37	0.34	0.34
10	0.50	0.47	0.44	0.41	0.42	0.36	0.34	0.34
15	0.50	0.47	0.44	0.41	0.41	0.37	0.34	0.33
30	0.50	0.48	0.44	0.42	0.44	0.37	0.34	0.33
15 chromosomes; Genome size (M):								
12	0.49	0.42	0.36	0.32	0.34	0.27	0.22	0.22
18	0.49	0.45	0.40	0.37	0.38	0.32	0.28	0.27
24	0.50	0.46	0.43	0.39	0.41	0.35	0.31	0.31
30	0.50	0.47	0.44	0.41	0.41	0.37	0.34	0.33

[1] 100 replicates where run in which loci were randomly placed on the genome when 30, 300, and 3000 causal loci were underlying the trait. 10 replicates were run with 30,000 causal loci due to computational reasons. Standard error across replicates were always <0.01.

Equilibrium variance. The expected variance when Bulmer equilibrium is reached is influenced by r_H. Fig. 1 shows the predicted equilibrium variance for mass selection, which depends on the selection intensity and selection accuracy determined by the heritability. A recombination fraction of 0.5 indicates that loci are segregating independently. For an r_H of 0.3, the additional loss in genetic variance due to linkage between loci is slightly less than 10% of the starting genetic variance compared to an r_H of 0.5 when the heritability was 0.4, and slightly more than 10% when the heritability was 0.8.

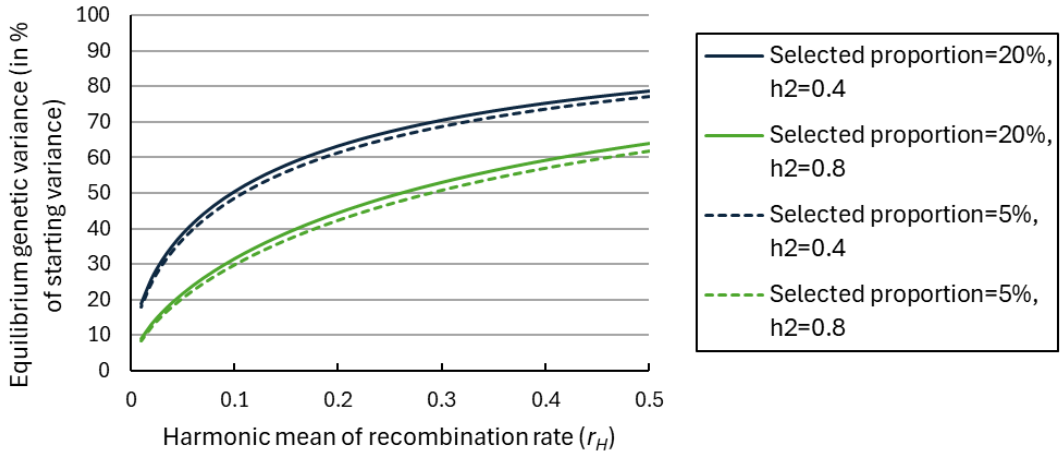

Figure 1. Expected equilibrium genetic variance (in % of the starting variance) as a function of r_H for heritabilities of 0.4 and 0.8; and selected proportions of 20% and 5% under mass selection. The infinitesimal model values are given by the y-values for an r_H of 0.5.

Simulations. After 50 generations of mass selection, the genetic variance was 17% lower than the genic variance in the No Linkage scenario, and 34% lower in the Linkage scenario. For GBLUP selection, the loss in genetic variance was 18% in the No Linkage scenario and 44% in

the Linkage scenario. This indicates that the loss in genetic variance is substantially higher when causal loci are linked and when selection is more accurate. When selection was stopped, the genetic variance became equal to the genic variance within a couple of generations for the No Linkage scenario, while it took around 150 to 250 generations for the Linkage scenario.
The r_H in the No Linkage scenario was 0.5, while it was on average 0.2 in the Linkage scenario. Based on those values, the predicted loss in genetic variance for mass selection was 21% for the No Linkage scenario and 36% for the Linkage scenario (Figure 1), which are in good agreement with the results of mass selection. Under mass selection, an equilibrium variance is predicted to be obtained within 4 generations for the No Linkage scenario, while it was predicted to take 7 generations for the Linkage scenarios.

Discussion

The usual assumptions about the transient loss in genetic variance are based on the infinitesimal model, assuming that all loci are segregating independently (Bulmer, 1974; Walsh and Lynch, 2018). However, selection can also create negative covariances between loci that are linked. The total loss in genetic variance due to the Bulmer effect is then dependent on the harmonic mean of the recombination rate (r_H) between the causal loci. We show that r_H can be considerably lower than 0.5 as is expected under the infinitesimal model, and depends on the number of causal loci and the genome size. For complex traits in livestock populations, we expect that the r_H is around 0.3. Therefore, depending on the selection intensity and accuracy, around 10% more genetic variance is expected to be lost due to the Bulmer effect. Moreover, the break-up of negative covariance on a chromosome happens at a much slower rate than the break-up of negative covariances between chromosomes, therefore, much more generations are needed to restore the genetic variance after selection has stopped. Those results indicate that using infinitesimal model assumptions, as for example done in the Animal Model, can create bias when estimating breeding values and genetic variance components.

Conclusions

Altogether, our results show that the impact of the Bulmer effect can be much greater than predicted by the infinitesimal model due to linkage between causal loci.

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References

Bulmer M. (1974) Genet. Res. 23:281-289.
Bulmer M.G. (1971) Am. Nat. 105:201-211.
Bulmer M.G. (1980) The mathematical theory of quantitative genetics. Clarendon Press., Oxford.
Dekkers J.C.M. (1992) Anim. Prod. 54:351-360. https://doi.org/10.1017/S0003356100020808
Falconer D.S., and Mackay T.F.C. (1996) Introduction to quantitative genetics. Fourth ed. Pearson Education Limited, Harlow, United Kingdom.
Haldane J. (1919) J. Genet. 8:299-309.
Lynch M., and Walsh B., (1998) Genetics and analysis of quantitative traits. Sunderland: Sinauer
Sargolzaei M., and Schenkel F.S. (2009) Bioinform. 25:680-681.https://doi.org/10.1093 /bioinformatics/btp045
Walsh B., and Lynch M. (2018) Evolution and selection of quantitative traits. Oxford University Press, Oxford.
Wientjes Y.C.J., Bijma P., Calus M.P.L., *et al*. (2022) Genet. Sel. Evol. 54:19.
https://doi.org/10.1186/s12711-022-00709-7
Wientjes Y.C.J., Bijma P., van den Heuvel J., *et al*. (2023) Genetics 225:iyad141. https://doi.org/10.1093/genetics/iyad141
Wientjes Y.C.J., Peeters K., Bijma P., *et al*. (2024) Genet. Sel. Evol. 56:76. https://doi.org/10.1186/s12711-024-00941-3