



## Genomic BLUP with additive mutational effects

J. Casellas, C. Esquivelzeta, Andres Legarra

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**Genomic BLUP with additive mutational effects**

Casellas, J.<sup>1</sup>, Esquivelzeta, C.<sup>1</sup> and Legarra, A.<sup>2</sup>, <sup>1</sup>Universitat Autònoma de Barcelona, G2R, Dep. Ciència Animal i dels Aliments, Campus UAB, 08193 Bellaterra, Spain, <sup>2</sup>Institut National de la Recherche Agronomique, UR631 SAGA, BP 52627, 31320 Castanet-Tolosan, France; joaquim.casellas@uab.cat

Genomic BLUP can accommodate both additive genetic (a) and mutational (m) effects by implementing  $y = \mu + Za + Zm + e$ , where y stores phenotypic data,  $\mu$  accounts for the population mean, Z is an incidence matrix and e is the residual term. Vectors a and m distribute under  $N(0, G\sigma_g^2)$  and  $N(0, M\sigma_m^2)$ , respectively, G being the genomic relationship matrix. Following the original development by N. R. Wray for mutational relationships, G can be generalized to accommodate the occurrence of new mutations, i.e. the mutational genomic relationship matrix (M). Previous model was exemplified on simulated data. Simulations relied on a genome composed by 5 chromosomes (1 cM each) with 2,500 SNP and 200 QTL per chromosome. Each replicate evolved during 1000 non-overlapping generations with effective size 100. After that, generations expanded to 200 individuals and analyses were performed on three different scenarios with 3 (Scen3), 5 (Scen5) or 10 (Scen10) generations with genomic (i.e. genotypes from polymorphic SNP) and phenotypic data ( $h^2=0.5$ ), and a last generation only contributing genomic data. A total of 20 replicates were analyzed for each scenario under the genomic BLUP described above ( $GB_m$ ) and under a standard genomic BLUP without mutational effects ( $GB_0$ ). Precision (i.e. correlation coefficient between simulated and predicted breeding values) for individuals with phenotypic data did not show differences between models  $GB_0$  and  $GB_m$ ; on the contrary, model  $GB_m$  increased precision for individuals from the last generation with advantages of  $0.15 \pm 0.15\%$  (Scen3),  $0.42 \pm 0.13\%$  (Scen5) and  $1.15 \pm 0.15\%$  (Scen10). These results evidenced that genomic BLUP can be efficiently adapted to accommodate genetic variability from new mutations, increasing the precision of the predicted breeding values for selection candidates.

**Design of the reference population affects the reliability of genomic selection**

Pszczola, M.<sup>1,2,3</sup>, Strabel, T.<sup>2</sup>, Mulder, H.A.<sup>3</sup> and Calus, M.P.L.<sup>3</sup>, <sup>1</sup>Wageningen University, Animal Breeding and Genomics Centre, Marijkeweg 40, 6700 AH Wageningen, Netherlands, <sup>2</sup>Poznan University of Life Sciences, Department of Genetics and Animal Breeding, Wolynska 33, 60-637 Poznan, Poland, <sup>3</sup>Wageningen UR Livestock Research, Animal Breeding and Genomics Centre, P.O. Box 65, 8200 AB Lelystad, Netherlands; mbee@jay.up.poznan.pl

The objective of this study was to investigate the effect of design of the reference population in terms of relationship levels within the reference population and relationship level of evaluated animals to the reference population on reliability of direct genomic values (DGV). Data reflecting a dairy cattle population structure was simulated for a trait with heritability of 0.3. Reference populations were small and consisted of cows only, to reflect a trait that is difficult or expensive to measure, e.g. methane emission. Reference populations with different family structures were chosen: highly, moderately, lowly, and randomly related animals. Evaluated animals were chosen from one generation after the reference populations. Reliabilities of DGV were predicted deterministically using selection index theory. Randomly chosen reference populations had the lowest values of average relationship within the reference population. Family structure of the reference population strongly influenced its average relationship and had an impact on reliabilities. Reliabilities increased when the average relationship within the reference population decreased, being 0.44 for highly and 0.53 for randomly related reference populations. Average squared relationship was chosen to measure the relationship to the reference population as it was closely related to calculated reliabilities. Individual reliability strongly increased with the relationship to the reference population. Composing the most suitable reference population is an optimization problem due to a trade-off between obtaining low average relationship within the reference population and high average squared relationship to the animals in the evaluated population.

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