

Testing candidate gene effects under various parameterizations and inheritance modes

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Running head: modeling candidate gene effects

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ABSTRACT

The objectives of this study was to assess the statistical properties of models commonly used for the estimation of single nucleotide polymorphisms (SNP) effects under the assumption of various inheritance modes and various parameterizations of SNP genotypes using simulated data and to compare effects of polymorphisms located within *BTN1A1*, *DGAT1*, *LEP* and *LEPR* candidate genes on milk production traits using real data from two dairy cattle breeds - 190 Jersey cows and 475 Polish Holstein-Friesian cows. Simulation results show that type I error and power are not dependent on the assumed parameterization, but differences are observed regarding confidence intervals of estimated SNP effects. Correct confidence intervals for all inheritance modes are provided only by the parameterizations independent of SNP allele frequency, while confidence intervals of parameterizations using SNP allele frequencies are only correct unless there is dominance in the model. Results based on real data show that for both breeds the additive effects of polymorphisms were generally similar, except *LEPR*, which for fat content has different increasing allele in Holstein-Friesian than in Jersey. In both breeds, *DGAT1* had the largest additive effect of the polymorphisms considered, but its effect on most milk traits was more pronounced in Jerseys than in Holstein-Friesians. The evidence of epistasis was found between *LEPR* and *DGAT1*, as well as between *LEPR* and *BTN1A1*, but only for milk content traits and only in the Holstein-

Friesian breed. There was also more evidence for dominance in Holstein-Friesian than in Jersey cows.