Assessment of 29 candidate genes for milk traits in Italian dairy cattle

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ABSTRACT

Several investigations have recently searched for significant association between gene polymorphisms and milk traits in livestock and model species. In several cases, it remains rather difficult to assess if the observed effects are caused by the mutation tested, by a nearby mutation in the same gene or by a mutation in a different gene or DNA region in linkage disequilibrium with the former. As a consequence, only in a few cases (e.g., κ-casein, SCD, DGAT1) the causative mutation seems to have been identified and, even when evidence is rather clear, genetic heterogeneity and genetic background may influence the size of allele substitution effects. Therefore, the significance of gene-trait associations and the estimate of their effect have to be verified in any new population in which this information is planned to be used, to estimate its actual utility in gene assisted breeding. In the SelMol project, we selected 29 candidate genes on the basis of known relationships between physiological or biochemical processes and evidence of significant association with milk traits in cattle, in related (e.g., sheep and goats) and model (e.g., mouse) species. A total of 106 SNPs were selected, using either information available in literature, or *in silico*, searching the NCBI dbSNP database. SNPs found significantly associated in other investigations were preferentially targeted. Otherwise non-synonymous SNPs and those in putative control regions (e.g., in promoter binding sites) were selected from dbSNP. If within a gene no SNP having one of these characteristics was available in dbSNP, synonymous SNPs, occurring in introns and untranslated non-control regions were chosen. DNA was extracted from semen of elite sires. SNPs polymorphism was confirmed by screening a panel of 32 individuals each of Pezzata Rossa (PR), Bruna Italiana (BI), and Frisona Italiana (FI) dairy cattle breeds. A total of 73 SNPs were confirmed as polymorphic in at least one breed: 63 in PR, 61 in BI, and 68 in FI. Polymorphic SNPs were genotyped on 400 individuals of PR and 600 of BI. Statistical tests were applied to detect selection sweeps, significant association to EBVs and phenotypic traits related to milk production and quality (milk yield, protein and fat yield and percentage), together with a number of functional traits (fertility, SCS as indicator of mastitis resistance, conformational traits, and milkability). *This work has been supported by the SelMol project financed by MIPAAF.*