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To cite this article: L. Fontanesi, E. Scotti, F. Schiavini, V. La Mattina, R. Davoli, V. Russo & A. Bagnato (2007) Investigation of the DGAT1 K232A and VNTR mutations in dairy and dual purpose cattle breeds, Italian Journal of Animal Science, 6:sup1, 137-137, DOI: [10.4081/ijas.2007.1s.137](https://doi.org/10.4081/ijas.2007.1s.137)

To link to this article: <https://doi.org/10.4081/ijas.2007.1s.137>



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Published online: 15 Mar 2016.



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# Investigation of the *DGAT1* K232A and VNTR mutations in dairy and dual purpose cattle breeds

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## ABSTRACT

Several studies have reported that the centromeric end of bovine chromosome 14 harbours QTL for milk production and composition traits. The acyl-Coenzyme A:diacylglycerol acyltransferase 1 (*DGAT1*) gene was indicated to be the quantitative trait gene affecting these traits with a major effect on milk fat content. A two bp mutation in exon 8 causing a nonconservative lysine to alanine amino acid substitution at codon 232 (*K232A*) showed a confirmed effect across breeds with allele *K* resulting associated with an increase on fat yield, fat percentage and protein percentage while allele *A* was associated with higher milk yield. Another mutation in the 5' regulatory region of this gene, a variable number of tandem repeat (VNTR) of 18 bp, was suggested to affect fat percentage. The objective of the present work was to investigate the occurrence of the *DGAT1* K232A and VNTR polymorphisms in several Italian dairy and dual purpose cattle breeds as a first step to evaluate their effects on milk production traits. A PCR-RFLP method was used to analyse the K232A polymorphic site in 114 Italian Holstein, 105 Italian Brown, 95 Italian Simmental, 95 Valdostana Red Pied, 50 Rendena, 120 Reggiana and 48 Modenese animals (sires and/or cows). Sequencing of animals with different genotypes was carried out to verify the PCR-RFLP genotyping protocol. Italian Brown and Modenese breeds resulted fixed for allele *A*. In either Italian Simmental, Valdostana and Rendena breeds all individuals were with genotype *AA* except for one animal that showed genotype *AK*. In Italian Holstein and Reggiana breeds allele *K* had a frequency of 0.25 and 0.17, respectively. The VNTR was genotyped using a capillary sequencer to detect fluorescent labelled PCR products in the same Italian Holstein, Rendena, Reggiana and Modenese animals indicated above showing 5, 2, 4 and 3 alleles, respectively. In Italian Holstein and Reggiana breeds allele "3" was the most frequent (0.59 and 0.51, respectively) while in Rendena and Modenese breeds the most frequent was allele "2" (0.81 and 0.56, respectively). Sequencing of the VNTR region in several animals confirmed the genotypes obtained in fragment analysis.