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VICUGNA PACOS A$1-CASEIN: IDENTIFICATION OF NEW POLYMORPHISMS AT THE CSN1S1 GENE

Erhardt G.\textsuperscript{1}, Gu M.\textsuperscript{2}, Wagner H.\textsuperscript{3}, Di Stasio L.\textsuperscript{2}, Pauciullo A.\textsuperscript{1,2}

\textsuperscript{1}Institute for Animal Breeding and Genetics, Justus Liebig University, Gießen, Germany, georg.erhardt@agrar.uni-giessen.de
\textsuperscript{2}Department of Agricultural, Forest and Food Sciences, University of Turin, Grugliasco (TO), Italy, alfredo.pauciullo@unito.it
\textsuperscript{3}Department of Obstetrics, Gynaecology and Andrology of Large and Small Animals with ambulance, Justus Liebig University, Gießen, Germany, henrik.w.wagner@vetmed.uni-giessen.de

Genetic polymorphisms in milk proteins is due to gene mutation resulting in either substitution or deletion of amino acids sequence along the peptide chain. South American camelids were genetically poorly investigated so far and little information is available in alpacas (Vicugna pacos) regarding the diversity of the caseins. The aim of this study was to investigate the presence of polymorphisms at the CSN1S1 gene in alpacas at protein and DNA level. The analysis of whole alpaca milk by IEF from 20 samples evidenced polymorphic protein patterns corresponding to αs1-CN migration area. According to the nomenclature in Llamas, the variants were 2 and 3 with 2 being the anodic one. Three variants (2/2, 2/3, 3/3) could be observed. Estimation of frequencies resulted in 0.475 and 0.525 for the variants 2 and 3, respectively. Blood and hair samples were collected from 130 alpacas belonging to different flocks in Germany and Italy. They were used for DNA isolation. The SNP c.366A>G at the exon 12 was successfully genotyped by PCR-RFLP. Two (AA, AG) of the three possible genotypes could be demonstrated resulting in estimated allele frequencies c.366 A 0.91 and c.366G 0.09. Milk samples from IEF showed different genotypes at DNA level for the SNP (c.366A\textsuperscript{Ile}>G\textsuperscript{Val}) as the mutation at the exon 12 does not lead to a relevant change in terms of pI of protein and therefore it was not possible to establish a link with IEF results. The polymorphisms found in alpaca have not been described before. The presence of the adenine (c.366A) at the exon 12 of the alpaca CSN1S1 might represent the ancestral condition of the gene because it has been found also in the other camelids. These data add knowledge to the genetic variability of a species little investigated and they open the opportunity for further investigation in the field of milk protein for South American camelids.