

Additive and dominance effects of the α_{s1} -casein locus on milk yield and composition traits in dairy goats

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The objective of this study was to evaluate the effects of the CSN1S1 locus polymorphism on 305-d records of milk, fat, protein, lactose and total solids yields, fat, protein, lactose and total solids contents in Mexican dairy goats. A total of 514 lactation records belonging to Alpine ($n=60$), Saanen ($n=105$) and Toggenburg ($n=74$) goats, born from 2003 to 2006 in three herds were used. Discrimination between alleles E, F, N, A* (CSN1S1 A, G, H, I, O1 and O2) and B* (CSN1S1 B1, B2, B3, B4, C and L) were made by amplification of fragments of the gene CSN1S1 and digestion with the restriction endonuclease XmnI. In order to estimate additive and dominance effects, data sets including (1) all genotypes, and (2) only homozygote genotypes, were analysed using linear mixed models. The allele A*, had significant additive effects for protein content ($0.21 \pm 0.07\%$; $P=0.002$) and total solids content ($0.66 \pm 0.23\%$; $P=0.005$) when compared with allele F. An unfavourable additive effect of allele A* on milk yield was found in the Alpine breed (-81.4 ± 40.2 ; $P=0.046$) when compared with allele F. Favourable dominance effects were found for some genotypes ($P<0.05$) for milk yield (A*N and B*N), fat yield (A*N and B*E), protein yield (A*N and B*E), lactose yield (A*N) and total solids yield (A*N). Also, unfavourable dominance effects were found ($P<0.05$) for protein content (A*B* and A*N) and total solids content (A*B*, A*N, and A*F). Allele A* was the only one with a positive effect for protein content. Significant allele-year interaction effects were also observed. The presence of significant dominance effects, estimated between specific pairs of alleles, challenged the purely additive nature of the genetic effect at the CSN1S1 locus. Implications from use of CSN1S1 effects in goat breeding programmes are presented.

Keywords: CSN1S1, alpha-s1 casein, milk trait, dairy goat, dominance effect.

The α_{s1} -casein locus (CSN1S1) is highly polymorphic in goats. According to their individual approximate contribution to the amount of protein in milk observed in previous studies, alleles for this locus have been grouped in 4 levels: strong alleles (A, B1, B2, B3, B4, C, H, L, and M), intermediate alleles (E and I), weak alleles (F, D, and G), and null alleles (O1, O2, and N) with no α_{s1} -casein content (Grosclaude et al. 1987; Martin et al. 2002; Ramunno et al. 2005; Sacchi et al. 2005; Sztankóová et al. 2007).

Previous studies have reported that the CSN1S1 can affect casein, protein, and fat levels, total solids, milk rheology, as well as cheese yield and quality (Pirisi et al. 1994; Clark & Sherbon, 2000; Martin et al. 2002; Roncada et al. 2002; Gómez-Ruiz et al. 2004; Zeng et al. 2007; Pagano et al. 2010).

In addition to additive (individual allelic effects), dominance effects (specific effects for allele combinations), as well as interactions between CSN1S1 alleles and other genes of the studied populations (epistatic effects) are possible. Some evidence of several milk protein genes, acting as haplotypes on dairy traits has also been found (Hayes et al. 2006). Despite those possibilities, dominance effects, in this locus, had not been studied until recently in Norwegian

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Table 1. Genotypic frequencies of the CSN1S1 locus by breed

Genotype†	Alpine		Saanen		Toggenburg		Total	
	n‡	%	n‡	%	n‡	%	n‡	%
A*A*	2	3.3	0	0.0	0	0.0	2	0.8
A*B*	2	3.3	0	0.0	3	4.1	5	2.1
A*N	4	6.7	0	0.0	6	8.1	10	4.2
A*E	3	5.0	1	1.0	1	1.4	5	2.1
A*F	5	8.3	2	1.9	4	5.4	11	4.6
B*B*	2	3.3	1	1.0	1	1.4	4	1.7
B*N	3	5.0	1	1.0	6	8.1	10	4.2
B*E	6	10.0	17	16.2	2	2.7	25	10.5
B*F	4	6.7	2	1.9	19	25.7	25	10.5
NN	5	8.3	1	1.0	2	2.7	8	3.4
NE	3	5.0	5	4.8	5	6.8	13	5.4
NF	1	1.7	1	1.0	4	5.4	6	2.5
EE§	10	16.7	41	39.1	7	9.5	58	24.3
EF	5	8.3	27	25.7	5	6.8	37	15.5
FF	5	8.3	6	5.7	9	12.2	20	8.4
Total	60		105		74		239	

†A*=A, G, H, I, O1 and O2; B*=B1, B2, B3, B4, C and L

‡Number of genotyped goats

§EE=EE and EO1

goats (Dagnachew et al. 2011) but there is no information published for Alpine and Saanen breeds.

The objective of this study was to evaluate both additive and dominance effects of CSN1S1 locus polymorphism on milk yield and milk composition traits in Alpine, Saanen, and Toggenburg goats.

Materials and Methods

Populations

Analysed data were obtained from 2003 to 2006 from Alpine, Saanen and Toggenburg in three herds located in Apaseo el Grande (at 1780 m above mean sea level), Guanajuato, Mexico. This area has a semi-dry temperate climate with an average temperature of 18 °C, rain during the summer (approx. 70% of the annual rainfall of 605 mm, occurs from May to September) (CONAGUA, 2011). Goats were maintained in an intensive production system, fed with alfalfa hay and commercial concentrate, and were supplemented with vitamins and minerals. The milk produced in these herds is used for making cheese.

The official milk recording system used is the A4 of the International Committee for Animal Recording (ICAR, 2011). Fat, protein, lactose and total solids yields were obtained from milk yield and percentage measures for each monthly test day (based on twice daily milking). Fat, protein, lactose, and total solids contents were monthly obtained using an infrared milk analyser Bentley 150.

Lactation records with less than 3 test-day milk yield or milk components measurements information per goat (or less than 100 d in milk) were discarded. Records with less than 305 d in milk were projected to 305 d using

methods described by Torres-Vázquez et al. (2009). The final data set consisted of 514 lactation 305-d records from 239 goats; 60 Alpine (130 lactations); 105 Saanen (210 lactations); and 74 Toggenburg (174 lactations). Analysed traits were milk (MILY), fat (FATY), protein (PROY), lactose (LACY), and total solids (SOLY) yields, fat (%FAT), protein (%PRO), lactose (%LAC), and total solids (%SOL) contents.

Genotyping

The genomic DNA was extracted from the blood by Phenol-chloroform extraction. Discrimination procedure between CSN1S1 alleles E, F, N, A* (A, G, H, I, O1, O2) and B* (B1, B2, B3, B4, C, L) was developed by amplification of segments of the CSN1S1 gene by PCR and XmnI endonuclease restriction digestion according to Ramunno et al. (2000) as in Torres-Vázquez et al. (2008).

Statistical procedures

Analyses were performed using linear mixed models and PROC MIXED of SAS® program (SAS version 9.0, SAS Institute Inc., Cary NC, USA). Final analyses considered two models; data analysed with model 1 included information from all available genotypes, while model 2 included only information from homozygote genotypes.

Model 1 included the fixed additive covariate effects of the alleles A*, B*, N and E coded representing the number of those alleles present in one genotype (2 for homozygotes; 1 for heterozygotes; and 0 for genotypes without that allele). The effect of the F allele was set to 0 (solution restriction) to avoid the singularity of the design matrix, which is

Table 2. Estimates of additive and dominance effects for milk yield and milk composition in goats of all the studied breeds

Trait†	Additive allelic effects (b‡)					Dominance genotypic effects (b‡)								
	A*	B*	N	E	A*B*	A*N	A*E	A*F	B*N	B*E	B*F	NE	NF	EF
MILY, kg	-53.6	-63.9	0.60	-7.9	188.1	269.6***	132.1	17.2	163*	94	29.6	73.4	-23.1	-15.3
FATY, kg	-0.56	-1.93	0.50	0.07	3.05	6.75*	3.01	0.99	3.16	3.86*	2.63	1.13	-1.16	0.05
PROY, kg	0.25	-1.32	0.35	0.02	1.89	5.86**	3.13	-0.27	3.74	2.8*	1.64	2.9	0.01	-0.2
LACY, kg	-4.45	-3.25	-0.61	-1.05	9.15	13.41**	6.83	1.34	7.28	3.67	2.03	5.18	1.73	-0.65
SOLY, kg	-0.87	-8.33	2.14	-0.49	16.99	22.3*	19.72	-2.94	15.01	11.32	8.31	7.96	1.29	-0.7
%FAT, %	0.17	0.03	0.00	0.02	-0.37	-0.27	-0.27	-0.07	-0.19	0.02	0.09	-0.14	-0.01	0.06
%PRO, %	0.21**	0.04	0.00	0.02	-0.36**	-0.27*	-0.13	-0.15	-0.05	0.02	0.06	0.08	0.02	0.03
%LAC, %	-0.25*	-0.04	-0.07	-0.08	0.09	0.2	0.13	0.09	-0.04	-0.02	0.00	0.15	0.01	-0.05
%SOL, %	0.66**	-0.17	0.16	0.04	-0.88*	-1.15***	0.05	-0.97**	-0.26	0.17	0.38	-0.06	0.09	0.05

† Where MILY is milk yield, FATY is fat yield, PROY is protein yield, LACY is lactose yield, SOLY is total solids yield; %FAT is fat content; %PRO is protein content; %LAC is lactose content; and %SOL is total solids content
 ‡ b is the additive effect, estimated as a deviation from allele F; *P ≤ 0.05; **P ≤ 0.01; ***P ≤ 0.001
 A* = A, G, H, I, O1 and O2; B* = B1, B2, B3, B4, C and L

associated with the covariates. Hence, the alleles additive effects are defined as deviations with respect to allele F. Besides additive effects, model 1 included fixed dominance effects defined by a group of covariates (1 to indicate the presence of the combination of two alleles in one observation; 0 to indicate its absence), then covariate values for all homozygote genotypes were 0.

Other fixed effects included in model 1 were breed-herd combination, season of kidding (1 from October to March; 2 from April to September), year of kidding, and lactation number (grouped as 1–2, 2–4, and 5 or more lactations). Based on differences observed in preliminary analyses, the interaction effect of additive allele by year of kidding was also included in this model.

Random effects included in model 1 were goat nested in breed-herd subclasses (to model repeated lactation information), and residual effects.

Equation for model 1 was:

$$y_{ijklm} = \mu + \text{breed} - \text{herd}_i + \text{season}_j + \text{lactation number}_k + \text{year}_l + \text{goat}_{im} + b_1A^* + b_2B^* + b_3N + b_4E + b_5A^*B + b_6A^*N + b_7A^*E + b_8A^*F + b_9B^*N + b_{10}B^*E + b_{11}B^*F + b_{12}NE + b_{13}NF + b_{14}EF + e_{ijklm},$$

where y_{ijklm} is one observation of the response variables, b_1, \dots, b_4 are multiple regression coefficients associated to coded additive allelic effects A*, B*, N and E respectively, b_5, \dots, b_{14} are multiple regression coefficients associated to coded dominance effects for combinations of alleles A*, B*, N and E respectively, and e_{ijklm} is the random residual effect.

Model 2 was similar to model 1, but instead of additive and dominance effects, the genotype effect (considered as a categorical variable) was used. Tukey multiple range tests were used for genotype means comparison.

Equation for model 2 was:

$$y_{ijklmo} = \mu + \text{breed} - \text{herd}_i + \text{season}_j + \text{lactation number}_k + \text{year}_l + \text{goat}_{im} + \text{homozygous genotype}_o + e_{ijklmo},$$

where y_{ijklmo} is one observation of the response variables, genotype_o is the effect of the homozygous genotype and e_{ijklmo} is the random residual effect.

Results

Allelic and genotypic frequencies

In the Alpine breed, the frequencies of the alleles A*, B*, N, E, and F at the CSN1S1 locus were 0.150, 0.158, 0.175, 0.308 and 0.208, respectively. The same allelic frequencies for Saanen were 0.014, 0.105, 0.043, 0.629 and 0.210, respectively. And for Toggenburg they were 0.095, 0.216, 0.169, 0.182 and 0.338, respectively. Genotypes at CSN1S1 locus by breed are shown in Table 1.

In Alpine and Saanen breeds, alleles E (0.31 and 0.63, respectively) and F (0.21 in both breeds) were the most

Table 3. Least-squares means in homozygote genotypes† for milk yield and milk composition traits in the studied goats

Trait‡	A*A*	B*B*	NN	EE§	FF
MILY, kg	865.6±96.7	819.8±77.2	927.8±51.6	935.6±34.2	969.8±39.2
FATY, kg	28.8±3.0	26.6±2.4	30.2±1.6	29.16±1.0	30.24±1.2
PROY, kg	26.8±2.4	23.1±1.9	25.7±1.3	25.1±0.84	25.91±1.0
LACY, kg	31.7±4.3	34.9±3.4	39.0±2.2	38.12±1.5	41.76±1.7
SOLY, kg	98.8±10.3	86.5±8.3	102.6±5.7	98.22±3.7	103.31±4.2
%FAT, %	3.5±0.3	3.4±0.2	3.3±0.1	3.18±0.1	3.19±0.1
%PRO, %	3.2±0.1 ^a ¶	2.9±0.1 ^b	2.8±0.1 ^b	2.72±0.1 ^b	2.72±0.1 ^b
%LAC, %	3.6±0.2 ^c	4.3±0.2 ^{abc}	4.3±0.1 ^{ab}	4.11±0.1 ^{bc}	4.35±0.1 ^a
%SOL, %	12.1±0.4 ^a	10.7±0.4 ^{ab}	11.33±0.3 ^{ab}	10.79±0.2 ^b	10.89±0.2 ^{ab}

†A*=A, G, H, I, O1 and O2; B*=B1, B2, B3, B4, C and L

‡Where MILY is milk yield; FATY is fat yield; PROY is protein yield; LACY is lactose yield; SOLY is total solids yield; %FAT is fat content; %PRO is protein content; %LAC is lactose content; and %SOL is total solids content

§EE=EE and EO1

¶ Genotypes without a common superscript letter are different ($P<0.05$)

frequent. For Toggenburg breed, the most frequent alleles were F and B* (0.34 and 0.22, respectively). In general terms, the most frequent genotypes (Table 1) were EE (24.3%), EF (15.5%), B*E, and B*F (10.5%). The rest of the genotypic frequencies were lower than 10%. The frequency of the alleles A* and B* was low.

Additive allelic effects

Allele additive effects estimated using model 1 are presented in Table 2, while genotype least-square means obtained with model 2 are presented in Table 3. As allele F effect was set to 0 because its frequency was relatively high with homozygous FF goats present in all breeds (Table 1), and because it was also expected to be a weak allele. All additive effects are expressed as deviations from the mean effect of allele F.

Allele A* had a positive additive effect on %PRO (0.21%; $P=0.002$) and %SOL (0.66%; $P=0.005$), and a negative additive effect on %LAC (−0.25%; $P=0.023$). The allele E had a negative additive effect on %LAC (−0.08%; $P=0.058$). The additive affects for the other alleles were not significant ($P>0.10$).

Similar estimates from the model 1 were obtained with model 2 using only homozygous genotypes (Table 3). The additive effect of A* on %PRO, compared with F, was 0.23% ($P<0.05$) and −0.37% in the case of %LAC ($P<0.05$). Regarding %PRO, the additive effect of A* compared with E was 0.23% ($P<0.05$), and the difference for %SOL comparing A* and E was 0.65% ($P>0.05$). No statistically significant differences were observed between homozygote genotypes for other traits.

Dominance genotypic effects

Some traits were affected by significant dominance effects (Table 2). Favourable dominance interaction effects were found for several genotypes ($P<0.05$) for MILY (A*N and B*N), FATY (A*N and B*E), PROY (A*N and B*E), LACY (A*N) and SOLY (A*N). Also, unfavourable dominance

effects were observed ($P<0.05$) for %PRO (A*B* and A*N) and %SOL (A*B*, A*N and A*F).

Within breed additive effects

In order to verify whether the effects found occur across all breeds, within-breed analyses using model 1, were done. Results for Saanen and Toggenburg revealed that additive and dominance effects might be confounded due to the information structure characterized by the absence of homozygote individuals for the allele A*. Within-breed analysis showed that only Alpine breed results agreed, in general, with those from the complete data analysis (Table 4). However, Alpine breed analysis allowed us to detect a significant additive negative effect of the allele A* on MILY (−81.4; $P=0.046$) and of the allele E on LACY (−4.2; $P=0.039$); additionally, allele N (null for α_{s1} -casein production) was found to be significantly related to higher MILY (114.5; $P=0.027$) and PROY (3.64; $P=0.016$) (Table 4). These results for Alpine were very different with respect to the results from the general analysis (Table 1), in which N allele effect was not different from F allele for any trait, and therefore should be regarded with caution since they might be the product of some confounded effects. Dominance effects in Alpine breed agreed with complete data analysis. However, positive dominance effects were found ($P<0.05$) for MILY (A*E), FATY (B*N), PROY (A*E and NE), SOLY (A*E and B*N) and %SOL (NF); and a negative dominance effect was found ($P<0.05$) for MILY (NF).

Additive alleles by year interaction effects

Additive alleles by year interaction effects trends were found on several yield and composition traits ($P<0.10$). For milk yield, significant interaction additive by year effects were found for alleles A*, N, and E; and for composition traits a significant interaction of additive by year effects were found for alleles N and E. This suggests that environmental circumstances may modify additive allele effects.

Table 4. Estimates of additive and dominance effects for milk yield and milk composition traits† in Alpine goats

Trait	Additive allelic effects (b‡)										Dominance genotypic effects (b‡)											
	A*\$	B*	N	E	A*B*	A*N	A*E	A*F	B*N	B*E	B*F	NE	NF	EF	A*E	A*F	B*E	B*F	NE	NF	EF	
MILY, kg	-81.4*	-22	114.5*	-37	70.9	212.4**	184.6*	45.7	273.3	-5.1	-9	162	-309.4*	-29								
FATY, kg	-2.21	-1.6	2.02	-1.56	1.82	5.5*	4.37	0.35	12.68*	-0.8	1.52	4.92	-9.3	-0.7								
PROY, kg	-0.27	-0.5	3.64*	-0.36	1.5	5.22**	6.01**	1.54	7.52	0.71	0.75	6.53*	-7.58	-0.1								
LACY, kg	-6.52	-3	1.74	-4.2*	0.99	5.91	7.34	-2.44	13.07	-0.6	-4.1	10.7	-	-3.1								
SOLY, kg	-5.89	-5	7.16	-4.51	3.06	14.77	19.59*	-0.99	39.67*	0.92	1.73	19.5	-25.91	-1.9								
%FAT, %	0.12	-0.1	-0.25	-0.04	-0.2	-0.25	-0.34	-0.19	0.2	-0.1	0.14	-0.13	0.42	0.03								
%PRO, %	0.26***	0.03	0.02	0.07	-0.1	-0.15	0	-0.01	-0.1	0.08	0.08	0.17	0.24	0.08								
%LAC, %	-0.4*	-0.2	-0.31	-0.3*	-0.5	-0.17	0.12	-0.39	-0.01	-0.1	-0.4	0.37	-	-0.4								
%SOL, %	0.62*	-0.2	-0.51	0.03	-0.9	-1.24**	-0.48	-0.94*	0.73	0.18	0.28	0.06	2.16*	0.2								

† MILY: milk yield; FATY: fat yield; PROY: protein yield; LACY: lactose yield; SOLY: total solids yield; %FAT: fat content; %PRO: protein content; %LAC: lactose content; and %SOL: total solids content
 ‡ b is the additive effect, estimated as a deviation from allele F; * $P \leq 0.05$; ** $P \leq 0.01$; *** $P \leq 0.001$
 \$A* = A, G, H, I, O1 and O2; B* = B1, B2, B3, B4, C and L

Discussion

Our results showed that alleles E and F were the most frequent in Alpine (E: 0.308 and F: 0.208) and Saanen (E: 0.629 and F: 0.210). In these breeds, the observed frequencies agree with those reported in France (Saanen, E: 0.41 and F: 0.43; Alpine, E: 0.34 and F: 0.41), Italy (Saanen, E: 0.49 and F: 0.46; Alpine, E: 0.35 and F: 0.59), Mexico (Saanen, E: 0.42 and F: 0.37; Alpine, E: 0.24 and F: 0.28) and USA (Saanen, E: 0.71 and F: 0.30; Alpine, E: 0.36 and F: 0.46) (Grosclaude et al. 1987; Martin & Leroux, 2000; Torres-Vázquez et al. 2008; Maga et al. 2009). In general, alleles E and F were the most frequent. This may help to explain the low content of protein in goat milk in high-yielding breeds, which limits cheese yield.

In this study, some confounding effects are possible since A* allele may contain O1 and O2 null alleles which are related to low protein percentages. However, previous research (Martin & Leroux, 2000; Maga et al. 2009) indicates that frequencies of these alleles in Alpine, Saanen and Toggenburg breeds are low, most probably below 0.05. Soares et al. (2009) found O1 allele frequencies of 0.01 and 0.02 in Alpine and Saanen goats, respectively. Therefore, probably these null alleles have a small effect in the estimated additive or dominance effects involving allele group A* in our study. Most previous studies considered that main allelic groups for this locus, in these breeds, are A, B, E, F and O (Martin & Leroux, 2000), but with different allele aggregation, and more recently A, B, E, F and N (Maga et al. 2009). Low frequencies for several alleles make it impossible to obtain accurate estimates of their individual effects.

Results showed that %PRO, %LAC and %SOL were significantly influenced by allele A* when compared with F. Our findings regarding %PRO (additive effects varying from 0.21 to 0.26%) (Tables 2 & 4) agree with previous studies, which ranged from 0.9 to 3.2 g/l, corresponding approximately to a difference varying from 0.09 to 0.32% (Remeuf, 1993; Analla et al. 2000; Chilliard et al. 2006) and are essentially the same to those observed by Manfredi et al. (1993). In general terms, our results are very similar to most French studies for Alpine and Saanen breeds (Mahé et al. 1993; Manfredi et al. 1993; Remeuf, 1993; Barbieri et al. 1995).

No significant effects of allele A* on PROY were found. Alleles N and E on this trait had similar effects to allele F, contrary to other studies where allele E effect was shown to be intermediate. However, superiority of allele A over alleles E and F for PROY had been observed also in some studies (Mahé et al. 1993; Remeuf, 1993). It is important to mention that the contrast of the effects of allele E and F on %PRO was not significant, while it was smaller for %LAC, similar to the case of allele A* (Table 2).

We found no evidence that allele B* effect was larger for %PRO (Tables 2 & 4), because allele B* behaved as a weak allele. This may be caused by the existence of several subtypes of alleles B (B1, B2, B3 and B4) whose effects on milk protein have been shown to be different in a Poitevine

Table 5. Predicted averages for different genotypes† compared with actual population averages for yields of milk (MILY), fat (FATY), protein (PROY), and total solids (SOLY); and contents of protein (%PRO), fat (%FAT), and total solids (%SOL)‡

	MILY, kg	PROY, kg	FATY, kg	SOLY, kg	%PRO	%FAT	%SOL
Actual population average	984	27.3	32.4	108.9	2.77	3.29	11.07
Average for optimum genotype for MILY (B*F)	1308	33.9	43.3	146.1	2.59	3.31	11.17
Average for optimum genotype for %PRO (A*A*)	779	24.6	28.5	96.8	3.16	3.66	12.43
Average for optimum genotype for %SOL (NF)	747	21.7	26.8	95.9	2.9	3.59	12.84

†A*=A, G, H, I, O1 and O2; B*=B1, B2, B3, B4, C and L

‡Values for PROY, FATY and SOLY, were calculated from MILY and the appropriate content average

goat population (Ricoordeau et al. 2000). It is worth mentioning that inconsistencies regarding the allelic effects on milk traits have been already observed by other authors (Hayes et al. 2006).

Although there was not a significant effect of the allele A* on %FAT, the estimated effect was positive and similar to that found for %PRO. Also, the effect of the allele A* on %SOL was important with an increment ranging from 0.62 ($P=0.031$) to 0.66% ($P=0.005$) when compared with allele F (Tables 3 & 5). While the effect of the allele A* on %LAC was negative (-0.25 ; $P=0.023$), results suggest that there is a favourable effect of the allele A* on %FAT that could not be statistically detected due to its higher variability (Table 2).

The effect of the allele A* on PROY and FATY was close to zero and not significant ($P>0.05$); this could be related to the possible negative effect of the allele A* on MILY, which was detected in Alpine goats (Table 4). Other authors have previously detected negative effects of the allele A on MILY (Barbieri et al. 1995; Ricoordeau et al. 2000). This agrees with the negative genetic correlations between milk yields and milk contents estimated in dairy goat populations (Torres-Vázquez et al. 2009). In our study, the genetic effects of the alleles at the CSN1S1 locus on the production of fat and protein by lactation were not significant, in agreement with Ricoordeau et al. (2000).

Dagnachew et al. (2011) found significant dominance effects of single SNP within CSN1S1 and CSN3 on contents of protein, fat, lactose and milk yield in Norwegian goats. Those findings are in agreement with our results. Dominance effects may compromise the efficiency of breeding programmes aimed simply to increase the frequency of strong alleles and to reduce the frequencies of weak and null alleles. With dominance, the mean for the %PRO could depend not only on the allelic frequencies at the CSN1S1 locus but also on the number and proportion of heterozygous genotypes in the population.

The effect of the alleles can be different, not only because the different level of the allele disaggregation used in a particular study, but also because the genes of the different dairy proteins could act as a whole. Studies are needed to examine the protein haplotypes through several dairy gene proteins to identify their effects accurately. Hayes et al. (2006) detected significant effects of haplotypes involving several casein loci (CSN2, CSN1S2, and CSN3), in addition to CSN1S1 locus.

Based on our results, we conclude that there are favourable effects of the allele A* on %FAT, %PRO, PROY and %SOL compared with the effects of the F allele, but the rest of the studied alleles behaved rather as weak alleles regarding these traits. Increasing the frequency of the allele A* may increase mainly strong allele frequencies and therefore lead to larger cheese yield per unit of processed milk (Zeng et al. 2007), but an important increment of the quantity of total solids, fat and protein per lactation on the population is not expected. Owing to the negative effects of the allele A* on %LAC, increasing its frequency may have a detrimental effect in the yield of products that contain or require lactose.

Despite the fact that some of the allelic effects are statistically significant, they do not allow the prioritizing of Genetic Marker-Assisted Selection (GAS) over conventional genetic evaluation methods. So far, the milk production and milk composition traits can be increased in a profitable way based only on phenotypic and pedigree information (BLUP evaluations), because they affect all loci involved on these traits (Montaldo & Manfredi, 2002). Simulations have shown that using this polymorphism in the context of breeding programmes for dairy goats may increase the rates of genetic gain for %PROT, over conventional progeny testing programmes based on phenotypic information only, but not for PROY (Sánchez et al. 2005).

A simple alternative to exploit the additive effect of the allele A* is to detect young males with high predicted breeding value based on best linear unbiased predictors (BLUP) for an economic index, which have at least one copy of the allele A*, or that have no weak alleles. These young males could have priority in breeding programmes with AI and progeny tests. However, the implications of this procedure, including the consequences on all the economically important traits and in the genetic variation of the population must be considered.

The existence of dominance effects complicates the use of this polymorphism in the genetic improvement of dairy goats, because as different genotypes are favourable/unfavourable for different traits, optimizing mating to obtain the desired genotypes in the population may be very difficult. Moreover, in practical goat breeding, the need of mating planning yield the ideal gene and genotype frequencies for this locus, may easily interfere with the breeding programme

mating plan for other traits depending on polygenic effects (Montaldo et al. 2010).

Genotypes for maximum MILY, %PRO and %SOL estimated from model 1 analyses are shown in Table 5. Means for all important milk traits are also displayed. The best genotype for MILY was BF*, the best for %PRO was A*A* and the best for %SOL was NF. Potential changes on the mean by substituting all population alleles by A*, are shown in Table 5. Changes for %PRO were 14% (from 2.77 to 3.16%), 11% for %FAT (from 3.29 to 3.66%), and 12% for %SOL (from 11.07 to 12.43%). As the change for MILY was – 21% of the mean (from 984 to 779 kg), the results on PROY and SOLY for substituting all the alleles by A* were negative (– 10 and – 11%, respectively).

Consequences of maximizing the average for some traits by manipulating the genetic make up of the population for this locus is shown in Table 5. The higher values for PROY and SOLY were observed for genotype B*F, which gave the highest average for MILY. Therefore, even if the use of this polymorphism for breeding might allow for the increase of %PRO (genotype A*A*) and %SOL (genotype NF), that might be at the expense of total cheese production by lactation per goat. The genotype with the highest average for %SOL has also reduced PROY, and SOLY averages if compared with the current population averages (Table 5).

Whether increasing %PRO and %SOL is a priority on a particular population will depend on many factors, particularly the relative economic value of increasing one unit of %PRO compared with increasing one unit of MILY, particularly when there is a high economic value for increasing the cheese-making properties of milk (Montaldo & Manfredi, 2002; Sánchez et al. 2005). Moreover, because allele A* is in such small frequency in all studied breeds, increasing its frequency may be impossible without compromising the effective population size (Table 1), creating potential problems regarding conservation of genetic diversity and avoiding an increase of inbreeding rate.

Therefore, using this polymorphism in goat breeding may be only justified when breeding goals are clearly defined and when there is a breeding organization able to carry out the programme and transfer the benefits to farmers (Gama & Bressan, 2011). Significant interaction effects between additive allelic and environment level (year) effects in our study were also observed by Pagano et al. (2010). These interactions constitute a further complication in the use of this major gene in breeding programmes addressed to increasing productivity in dairy goat populations.

Low frequency of A* allele in these high milk yield populations may be the result of an unfavourable pleiotropic effect for a selected trait (i.e. milk yield) as found in Alpine breed in our study. Another possibility is a negative effect of the A* allele on some important fitness traits (i.e. natural selection effects) yet to be identified.

Further research on this locus with larger samples and more precise techniques for allele discrimination are needed to confirm the estimates of additive and dominance effects found in this study.

Conclusions

Significant additive effects for allele A* v. allele F were found on protein and total solids milk contents, by using a model that included additive and dominance effects. However, no significant effects of the allele B, compared with allele F, were observed.

The presence of significant dominance effects at the CSN1S1 locus, modifies the prediction of GAS results by using just a model of allelic replacement.

Potential benefits from increasing the frequency of the allele A* in the goat population to increase cheese yield, needs to be balanced with its potential disadvantages, such as the deterioration of the mean or the reduction of genetic progress in other economically important traits, such as milk, protein and total solids yields, and a potential reduction of genetic variability of the population.

The presence of year by allele interaction effects may imply complications to accurately predicting the long-term effects of these alleles across production environments.

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