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Genetic architecture of somatic cell count in Sarda dairy sheep inferred from genomic information

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Introduction

Somatic cell count (SCC) is widely used in dairy species as an indicator of udder health and as a proxy to identify and select animals with reduced susceptibility to intramammary infections (IMI) (Rupp *et al.*, 2009; Banos *et al.*, 2017). In dairy sheep, clinical mastitis is often less evident and less frequently recorded than in dairy cattle, making SCC a useful indirect indicator of udder inflammation. As a result, SCC is commonly included among the traits considered in breeding programs aimed at improving udder health and reducing the economic losses associated with mastitis (Oget *et al.*, 2019). Conventionally, genetic evaluations have focused on the average level of somatic cell score (SCS) across lactation. However, IMI episodes are often transient and irregular, and strongly influenced by environmental challenges. Several studies have shown that traits describing the within-lactation dynamics of SCC can capture complementary aspects of the host immune response that are not fully reflected by average SCS (Green *et al.*, 2004; Urioste *et al.*, 2012). These traits may therefore provide complementary selection criteria for improving resistance to IMI, especially in dairy sheep, in which most intramammary infections occur in a subclinical form. In Sarda dairy sheep, a genomic selection program for economically relevant traits is currently being developed. A key feature of this program is the use of a thoroughly phenotyped female reference population, mainly consisting of nucleus flocks, to estimate genomic breeding values with sufficient accuracy (Carta *et al.*, 2026). In this context, assessing the genetic basis and genomic architecture of SCS-derived traits is essential to determine whether selection for reduced SCC and improved udder health can be effectively implemented in this breed. This study estimated genetic parameters for four lactation-level SCS traits and identified associated genomic regions.

Materials & Methods

A nucleus flock was established in the early 2000s, and subsequent generations were obtained by mating adult ewes with rams supplied by the Sarda breed artificial insemination center. Overall, 5,386 ewes were produced up to 2024. Management practices closely followed those commonly adopted in commercial herds in Sardinia: most adult ewes lambed in autumn, while yearlings lambed between January and March. The feeding system was based on grazing natural pastures and forage crops, with hay and concentrates provided in winter and late spring. The milking period extended from weaning (on average 30 days after lambing) to late June or early July, when high summer temperatures reduced fresh forage availability, and ewes were dried off. Individual milk samples were collected fortnightly between 2000 and 2024. Daily SCC values were calculated as the weighted average of the morning and evening milking, with weight based on the corresponding milk yield, yielding 152,291 test-day SCC records ($\times 10^3$ cells/mL). After data editing, which required at least two test-day records per lactation and the first test-day recorded within 80 days postpartum, 145,765 test-day records from 5,261 ewes were retained. Because SCC showed a highly skewed distribution, it was log-transformed into SCS as $\log_2(\text{SCC}/100{,}000) + 3$ to approach normality. Test-day records were subsequently transformed into lactation-level variables (16,554 records) to reflect different patterns of SCC

variation associated with IMI: lactation average SCS (SCSav), calculated as the mean of all test-day SCS values within each lactation; the standard deviation of test-day SCS (SCSsd), which is expected to capture within-lactation variation in SCS associated with temporary increases due to IMI; the maximum SCS (SCSmx), defined as the highest SCS value recorded for each ewe in each lactation, indicative of pronounced SCC increases potentially associated with IMI; the average deviation of SCS values from the lactation minimum (SCSwd), intended to reflect the animal's physiological baseline SCS and to quantify SCS variability relative to this minimum. All ewes in the nucleus flock, along with their sires and available ancestors (272 rams and 183 ewes), were genotyped using the Illumina OvineSNP50 BeadChip. SNP quality control was performed by applying call-rate and minor allele frequency thresholds of 90% and 1%, respectively, resulting in 43,650 SNPs across 26 autosomes being retained. SNP positions were aligned to the Ovis aries genome assembly v4.0, assuming a conversion of 1 Mb = 1 cM. Variance components were estimated using a multi-trait single-step genomic BLUP model implemented in the BLUPf90 software suite (Lourenco *et al.*, 2022). The four lactation-level traits were analysed using a multitrait animal model with repeated measures within ewes, allowing estimation of genetic correlations among traits. The model included additive genetic and permanent environmental random effects, as well as fixed effects comprising the interactions between year of production and management group, and between age class and month-of-lambing class. The additive genetic effect was modeled using the **H** matrix, that integrates genomic information for genotyped animals with pedigree relationships for the whole population. The pedigree comprised 9,198 animals spanning three generations. The average of lactation records corrected for fixed effects from the previous model, was used as an adjusted phenotypes in a linkage disequilibrium–linkage analysis (LDLA) aimed at QTL detection. This analysis was performed using the method proposed by Usai *et al.* (2019), which summarizes identity-by-descent (IBD) probabilities between gametes through principal component analysis. At each OvineSNP50 SNP position *l*, IBD probabilities (IBD_l) were estimated by combining linkage disequilibrium and linkage information. A genome-wide IBD matrix, obtained by averaging all IBD_l matrices, was included to account for polygenic background effects. QTL were tested using an F-test at each SNP position. The 5% genome-wide significance threshold, adjusted for 43,650 tests using a Bonferroni correction, corresponded to $-\log_{10}P = 6$.

Results & Discussion

The average number of test-day records per lactation was 8.8 ± 3.1 (range 2–16), and ewes had on average 3.2 ± 1.0 lactations (range 1–4). The mean ± standard deviation of SCC and SCS were 640 ± 1,858 × 10^3 cells/mL and 4.09 ± 1.77, respectively. Lactation-level SCS traits also showed considerable variability. Mean ± SD values were: SCSav = 4.04 ± 1.33, SCSsd = 1.13 ± 0.61, SCSmx = 5.98 ± 2.01, and SCSwd = 1.38 ± 0.82. All traits were positively correlated, with associations ranging from moderate to high (0.45-0.82). SCSav showed a stronger relationship with SCSmx, whereas traits describing within-lactation variability were more closely related to each other. These raw correlations are partly expected because the traits are mathematically related and derived from the same SCC profile, although they partly reflect different aspects of somatic cell dynamics during lactation. The estimated variance components differed markedly among traits (Table 1). SCSav showed the highest heritability and repeatability, indicating a stronger genetic contribution to differences among animals. These estimates are slightly higher than those commonly reported in dairy sheep, where heritability values for SCS typically range between 0.10 and 0.25 (Oget *et al.*, 2019). In contrast, SCSsd and SCSwd showed the lowest heritabilities, reflecting a substantial contribution of non-genetic individual factors to within-lactation variation.

Table 1. Variance components[1], heritability (h^2), and repeatability (r)

Variable	S^2_a	S^2_{pe}	S^2_e	S^2_{tot}	h^2	r
SCSav	0.404	0.305	0.818	1.526	0.26	0.46
SCSsd	0.020	0.014	0.323	0.356	0.06	0.10
SCSmx	0.483	0.370	2.644	3.497	0.14	0.24
SCSwd	0.040	0.040	0.473	0.553	0.07	0.14

[1] S^2_a: additive genetic variance; S^2_{pe}: permanent environmental variance; S^2_e: error variance; S^2_{tot}: total variance.

SCSmx showed intermediate values, which is consistent with its definition as a trait influenced by both persistent and episodic increases in somatic cells. Overall, SCSav shows the highest genetic component, while indicators conceived to capture SCS dynamics are more strongly affected by non-genetic sources of variation. This finding is in agreement with previous studies, showing that variability-based SCC traits tend to exhibit lower heritability than average SCC (Green *et al.*, 2004; Urioste *et al.*, 2012).

Table 2. QTL regions that include SNP positions exceeding the 0.05 genome-wide significance threshold ($-\log_{10}P) > 6.00$).

QTLR	TRAIT	OAR	Significant	Highest peak		Significant	Pvalue
			SNP (n)	SNP name	Pos. (Mb)	region (Mb)	
1	SCSav	1	8	rs399216686	19.7	19.7-20.3	7.28
2		1	1	rs401825520	140.3	140.3-140.3	6.17
3		1	5	rs430323970	260.1	258.1-260.3	7.44
4		2	5	rs417412611	21.8	12.0-21.8	7.19
5		2	2	rs420332966	94.4	86.3-94.4	7.13
6		2	12	rs407860173	157.3	157.0-158.2	7.00
7		3	73	rs406548594	18.5	17.0-24.4	11.70
8		3	6	rs399754751	148.8	141.4-160.5	7.47
9		4	1	rs418980674	21.2	21.2-21.2	6.17
10		4	17	rs413636816	62.5	52.7-62.8	7.54
11		8	19	rs406888748	75.1	71.7-75.1	7.82
12		10	2	rs404084643	30.6	30.6-30.6	6.53
13		10	2	rs410535459	74.9	74.9-74.9	6.61
14		12	9	rs425016615	14.3	11.9-14.3	7.74
15		14	1	rs411120068	58.5	58.5-58.5	7.43
16		17	4	rs430601155	7.5	7.5-7.6	6.57
17		18	7	rs423599441	21.9	17.3-21.9	7.54
18		21	2	rs408421719	47.9	46.5-47.9	7.57
19		26	4	rs416027623	37.7	34.9-37.7	7.38
20	SCSmx	21	11	rs429302639	13.1	11.9-13.2	6.12
21	SCSwd	21	11	rs429302639	13.1	11.9-13.2	6.09

QTLR, QTL region identifier; TRAIT, SCSav: average SCS per lactation, SCSmx: max SCS per lactation, SCSwd: average deviation from minimum SCS per lactation; OAR, *Ovis aries* autosomes; Significant SNP (n), number of SNP positions exceeding the 0.05 genome-wide significance threshold; Highest peak, the most significant SNP; SNP name, Pos. (Mb), name and position in Mb (from the ovine genome assembly v4.0) of the most significant SNP; Significant region (Mb), position in Mb of the first and last significant SNP of the QTL region; Pvalue, highest significance among the SNPs within a QTL region exceeding the 0.05 genome-wide significance threshold.

Genetic correlations were consistently positive (0.44–0.96), with SCSav showing strong associations with the other traits, particularly SCSmx (0.96) and SCSwd (0.72). Considering these correlations, the mathematical links among derived measures, and the generally higher heritability of SCSav, selection on SCSav is expected to produce favourable correlated responses, potentially larger than the direct response achievable when selecting on the other traits. The LDLA scan revealed multiple genomic regions associated with the lactation-level SCS traits (Table 2). A total of 21 QTL regions above the genome-wide threshold were identified across 12 chromosomes. Most significant associations were detected for SCSav, which showed the strongest signals, including a prominent peak on chromosome 3 at ~18.5 Mb ($-\log_{10}P = 11.70$). Additional QTL for SCSav were identified on chromosomes 1, 2, 4, 8, 10, 12, 14, 17, 18, 21, and 26, indicating that multiple genomic regions contribute to the genetic variation of this trait, with some showing major contributions. Significant QTL for the remaining traits were less common. A noteworthy signal was detected on chromosome 21, where SNP rs429302639 reached genome-wide significance for SCSwd and SCSmx and showed a suggestive association with SCSsd ($-\log_{10}P = 5.48$). This was the only region apparently specific to traits aimed at capturing within-lactation dynamics, and may therefore help target the genetic component of these traits that is not shared with SCSav.

Conclusions

This study provides new insights into the genetic architecture of somatic cell count traits in Sarda dairy sheep, using a female reference population developed for genomic selection. Among the traits analysed, the average somatic cell score showed the strongest genetic determinism and the most consistent genomic signals, supporting its role as a suitable criterion for selection aimed at improving udder health. Traits describing within-lactation variability displayed lower heritability, indicating a stronger influence of non-genetic factors. Further research is needed to assess whether these traits capture aspects of SCC dynamics related to episodic IMI events that are not fully explained by average SCS. Several genomic regions were associated with the analysed traits, with loci on chromosomes 3 and 21 emerging as particularly promising targets for further investigation. Overall, the ability to estimate reliable genetic parameters and to identify significant QTL in this nucleus flock supports its suitability as reference population for the development of genomic tools aimed at improving udder health and reducing susceptibility to intramammary infections in Sarda dairy sheep.

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