er coagulation properties, except for SNP AMGL02030943.1:g.23731G > T which tended to affect A20 ($P < 0.10$). In the *PRLR* gene, SNP rs600947105 genotypes affected protein content throughout lactation ($P < 0.05$) and tended to affect total solids content ($P < 0.10$). It also influenced pH ($P < 0.05$), A20 ($P < 0.01$), and AR ($P < 0.01$). Results suggest that the aforementioned SNPs might be used in gene-assisted selection programs for the improvement of milk quality traits and coagulation properties in Assaf sheep. However, analysis should be extended to a larger number of animals to validate this results. Funding: Project financed by European Fund for Regional Development (ERDF) [ALT20–03–0145-FEDER-000019]

Key Words: sheep, SNPs, milk traits, PRL, PRLR

P53 Genome-wide association studies for somatic cell count in Assaf breed. Y. Öner*[1], M. Serrano[2], M. Ramón[3], M. P. Sarto[4], L. P. Iguacel[4], M. Joy[4], M. Blanco[4], O. Estrada[4], T. Juan[4], and J. H. Calvo[4], *[1]Bursa Uludag University, Bursa, Turkey, [2]INIA (Instituto Nacional de Investigación y Tecnología Agraria y Alimentaria, Madrid, Spain, [3]Centro Regional de Selección y Reproducción Animal (CERSYRA)-Instituto Regional de Investigación y Desarrollo Agroalimentario y Forestal de Castilla-La Mancha (IRIAF-JCCM), Valdepeñas, Spain, [4]Centro De Investigación Y Tecnología Agroalimentaria De Aragón (CITA), Zaragoza, Zaragoza, Spain.*

Alteration in somatic cell count (SCC) is widely used as indicator of mastitis, being one of the most costly production-related infectious disease in dairy industry. The aim of this study therefore was, to identify SNPs and genes associated with somatic cell count in sheep by using the Illumina AgResearch Sheep HD (680K). The animals for association studies were selected from 3 flocks of the Spanish Assaf breed. These flocks belong to the Teruel Association of Dairy and Cheese Producers. Only multiparous ewes with 2 or more lactations and at least 3 test day records during one lactation were considered. In total, we used 6173 records from 1907 ewes with at least 3 test day records between 2 and 7 years old. SCC data were logarithmic transformed. The animal effects phenotype in the whole population (n = 1907) and the SCC phenotype were estimated by fitting a Repeatability Mixed Model that included the herd-test day, the number of lambs born and the days in milk as fixed effects and the ewe herself as a random effect. One hundred and 90 2 animals with extreme values for the animal effects for the SCC phenotype were selected for GWAS analysis (n = 96 for low SCC; and n = 96 for high SCC). The association analysis was performed with the GCTA software. Significance of associations was assessed using a false discovery rate (FDR) multi-test correction. Locations of SNPs and genes were identified based on the sheep genome Ovis aries v 3.1. Genes located within 250 kb on either side of the significant SNP were annotated. After quality control with PLINK 559,762 SNPs were used for association analyses. The MLMA analysis did not reveal any significant SNP at genome level. However, 4 SNP on OAR19 were significant at chromosome level ($P < 0.01$). The first SNP was located in *NUP210* gene (rs419096188), and the other 3 SNPs (rs415580501, rs410336647, and rs424642424) were close to *ARPP21* gene, and mir128–2, expressed in the mammary gland of lactating goats, localized in *ARPP21*. These genes have been involved in viral response, cholesterol homeostasis and stress response.

Key Words: Assaf sheep, GWAS, somatic cell count

P54 Identification of a new mutation responsible for epidermolysis bullosa in Mouton Vendéen sheep. L. Chantepie*, L. Drouilhet, C. Genêt, F. Plisson-Petit, J. Sarry, G. Tosser-Klopp, F. Woloszyn, and S. Fabre, *GenPhySE, Université de Toulouse, INRA, INPT, ENVT, Castanet Tolosan, France.*

Junctional Epidermolysis Bullosa (EB) is a severe congenital disease affecting the skin at the extremities of the limbs and the mucous membranes. In sheep, this recessive disease causes perinatal death of affected lambs. Multiple observations of EB cases were recently reported in the French Mouton Vendéen meat sheep breed. Skin biopsies of 6 affected lambs and when available, blood samples from the parents (n = 7) and unaffected full-sib lambs (n = 4) were collected for genomic DNA extraction. From a bibliography study and analysis of the OMIM database (Online Mendelian of Human Genes and Genetic Disorders), we have focused on 12 candidate genes mainly belonging to the collagen, laminin, integrin and keratin families. Based on the hypothesis of a recessive transmission of a deleterious variant, we have performed whole-genome sequencing of 2 unrelated EB-affected lambs (supposed homozygous carriers) and 1 unaffected full-sib (supposed heterozygous or non-carrier). Using the GATK workflow on a Galaxy platform, we have identified a novel SNP in the exon 23 of the *ITGB4* gene of the integrin family (OAR11_v4.0, g. 54799925 G > A (p.885 R > *)) whose variant allele causes a premature stop codon. By a specific RFLP assay, we have determined that all EB-affected lambs were homozygous for this variant allele, their parents were heterozygous and the full-sibs were either heterozygous or non-carrier, fitting well with the working hypothesis. Following this primary discovery, a larger set of Mouton Vendéen animals was genotyped. We estimated the population allele frequency at 6.8% by genotyping a cohort of renewal ewe lambs (n = 1227). We also found a 5.7% allele frequency among the breeding rams present in 2018 (n = 1007) in artificial insemination center, progeny-testing station, and for natural mating in farms. Moreover, the specific genotyping of producing ewes in the most EB-affected flocks revealed a variant allele frequency up to 13.3%, due to overuse of inbreeding strategy. In conclusion, the discovery of a new mutation in *ITGB4* causing EB in sheep will improve the selection scheme management of the Mouton Vendéen breed to limit the dissemination of this disease.

Key Words: sheep and related species, genetic disorder, genome sequencing, candidate gene

P55 Abstract withdrawn