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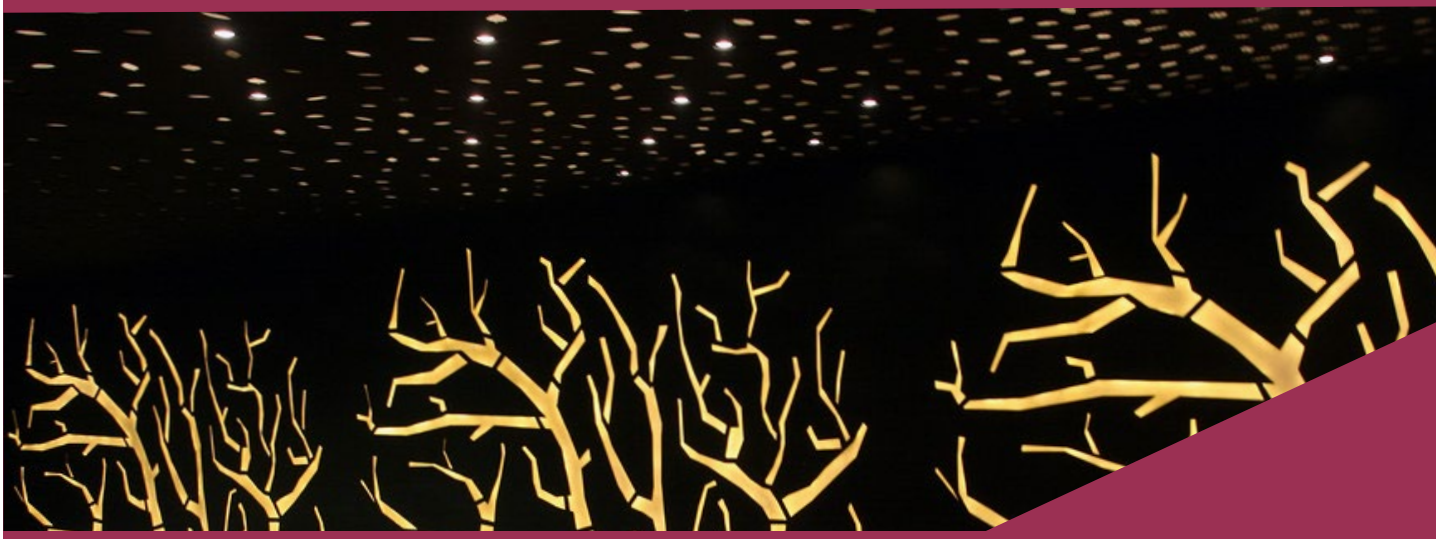

ABSTRACTS

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P49 Abstract withdrawn

P50 Inferring the population structure of six North African sheep breeds using a medium-density SNP chip. S. Ben Jemaa*[1], S. Kdidi[2], A. M. Gdura[3], A. S. Dayhum[4], and M. Boussaha[5], *[1]Institut National de la Recherche Agronomique de Tunisie, Ariana, Ariana, Tunisia, [2]Arid Lands Institute, Médenine, Médenine, Tunisia, [3]Ministry of Agriculture, Tripoli, Libya, [4]Faculty of Veterinary Medicine, Tripoli, Libya, [5]GABI, INRA, AgroParisTech, Université Paris Saclay, Jouy en Josas, Ile de France, France.*

This study aims at providing a detailed assessment of the population structure and the genetic origins of 6 North African sheep populations using the Illumina 50K ovine BeadChip and comparisons with 22 worldwide sheep and mouflon populations. To tackle this question, we genotyped 35 Barbarine (23 from Tunisia and 12 from Libya), 19 Black Thibar (BT), 15 Sicilio Sarde (SS), 17 West Thin Tail (WTT) and 6 D'man individuals on the OvineSNP50 Genotyping BeadChip. A fine-scale assessment of the genetic structure of the populations was provided using principal components Analysis (PCA), ancestry models implemented in ADMIXTURE software and Discriminant Analysis of Principal Components (DAPC). Patterns of splits and mixture of the populations of the study were carried out using TreeMix software. Furthermore, f3-statistics were used to provide further support for a past admixture between populations. Regardless of the analytical method used, patterns of multiple hybridization events were observed within all North African populations leading to a heterogeneous genetic architecture which varies according to the breed. The Barbarine population showed the lowest genetic heterogeneity with a major Southwestern Asian ancestry bringing a further support on the Asian origin of the North African fat-tail sheep. All other breeds presented substantial Merino introgression ranging from 15% for D'man to 31% for Black Thibar. We also identified several signals of ancestral introgression between North African and South European sheep. In addition, model-based clustering analysis identified 2 opposite gradients of ancestry, Southwestern Asian and Central European, occurring between North Africa and Central Europe. Our results bring further evidence for the weak global population structure of sheep resulting from high levels of gene flow between breeds occurring worldwide. At regional level, signals of recent admixture between North African populations were also detected resulting in change of the original genomic architecture of minority breeds.

Key Words: North African sheep, SNP, genetic structure, admixture

P51 Associations of single nucleotide polymorphisms in the ovine prolactin and prolactin receptor genes with milk traits in Assaf dairy sheep. M. R. Marques*[1,2], D. S. Ribeiro[3], S. Gomes[4], A. T. Belo[1], J. R. Ribeiro[1], A. P. Martins[4,5], and C. C. Belo[1], *[1]UEISPSA, INIAV Instituto Nacional de Investigação Agrária e Veterinária I.P, Vale de Santarém, Portugal, [2]CIISA, Centro de Investigação Interdisciplinar em Sanidade Animal, Lisboa, Portugal, [3]ESAC, Escola Superior Agrária de Coimbra, Coimbra, Portugal, [4]UTI, INIAV, Instituto Nacional de Investigação Agrária e Veterinária I.P, Oeiras, Portugal, [5]LEAF, Linking Landscape, Environment, Agriculture and Food, ISA, Lisboa, Portugal.*

Milk production is a process coordinated by a complex endocrine and nutritional signaling cascade in which genes from the somatotropic axis play a key role. Among them, the prolactin (*PRL*) gene is known to regulate mammary gland growth, lactogenesis, and galactopoiesis. The actions of PRL, and also the ones of growth hormone and placental lactogen, are mediated by PRL receptor (PRLR), which activate several intracellular signaling cascades. The aim of this work is to identify single nucleotide polymorphisms (SNPs) in *PRL* and in *PRLR* genes associated with milk traits in Assaf ewes. Nine SNPs were genotyped by SNapShot® analysis in 450 Assaf ewes. Data regarding 150 d adjusted milk yield (P150d), total milk yield (PTotal) and lactation length from 2007 to 2017, and milk quality traits (fat, protein, lactose, total solids and fat free total solids contents) evaluated in 184 ewes (one lactation), were analyzed by mixed model procedure to disclose associations between genotypes and milk traits. The 9 genotyped SNPs were found to be polymorphic (MAF ranging from 0.004 to 0.493). In the *PRL* gene, AA ewes at rs412263261 had higher P150d (372.8 ± 35.56 *vs* 229.1 ± 25.71 L/150d; $P < 0.01$), and PTotal (503.0 ± 58.10 *vs* 318.5 ± 42.02 L; $P < 0.05$) than CA ewes. TT ewes at rs406266481 had higher PTotal than GG ewes (412.5 ± 16.64 *vs* 346.8 ± 11.04 L; $P < 0.01$), but GG ewes presented shorter lactations (211.0 ± 3.50 *vs* 230.1 ± 5.23 d; $P < 0.01$). No associations were found between *PRL* genotypes and milk quality traits. In the *PRLR* gene, CT ewes at rs604784916 had higher PTotal (446.7 ± 31.17 *vs* 340.5 ± 32.22 L; $P < 0.05$), and longer lactations (241.1 ± 9.69 *vs* 208.4 ± 10.70 d; $P < 0.05$) than TT ewes. Regarding milk quality traits, CT ewes at rs600947105 produced milk with a significantly higher protein content than homozygous ewes ($P < 0.05$). The results suggest that SNPS rs412263261 and rs406266481 at the PRL gene, and rs604784916 at the PRLR gene may be useful as early selection markers for milk production traits in Assaf sheep. Funding: Project financed by European Fund for Regional Development (ERDF) [ALT20–03–0145-FEDER-000019]

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

P52 Effects of prolactin and prolactin receptor polymorphism upon milk composition and milk coagulation properties in Assaf ewes. M. R. Marques*[1,2], S. Gomes[3], D. S. Ribeiro[4], J. R. Ribeiro[1], A. T. Belo[1], A. P. Martins[3,5], and C. C. Belo[1], *[1]UEISPSA, INIAV, Instituto Nacional de Investigação Agrária e Veterinária I.P, Vale de Santarém, Portugal, [2]CIISA, Centro de Investigação Interdisciplinar em Sanidade Animal, Lisboa, Portugal, [3]UTI, INIAV, Instituto Nacional de Investigação Agrária e Veterinária I.P, Oeiras, Portugal, [4]ESAC, Escola Superior Agrária de Coimbra, Coimbra, Portugal, Coimbra, Portugal, [5]LEAF, Linking Landscape, Environment, Agriculture and Food, ISA, Lisboa, Portugal.*

Associations of 5 SNPs of the prolactin (*PRL*) and 4 SNPs of the PRL receptor (*PRLR*) genes with milk production, composition (fat, protein, lactose, total solids and fat free total solids content), pH, and coagulation properties assessed by Optigraph [clotting time (R), gel firmness after 20 min (A20), and after a 2R (AR) period and rate of firming (OK20)] were investigated in Assaf ewes. Milk production and composition were evaluated monthly until the sixth month of lactation in 184 ewes, and pH and coagulation properties were evaluated at the first and third month of lactation in 92 ewes. Data were analyzed using a mixed-model procedure with fixed effects of SNP and month of lactation, considering the linear and quadratic effect of ewe' lambing age covariate. In the *PRL* gene, SNP AMGL02030943.1:g.23731G > T genotypes affected milk production throughout lactation ($P < 0.01$). Regarding milk composition, rs406266481 genotypes influenced fat free total solids content ($P < 0.05$). Moreover, *PRL* gene SNPs tended to affect protein (rs406266481), and lactose (rs422713690) and fat free total solids (rs422713690, rs412263261) contents throughout lactation ($P < 0.10$). pH tended to be affected by rs406266481 ($P < 0.10$). Considering clotting time, R was affected by AMGL02030943.1:g.23731G > T ($P < 0.05$), rs412263261 ($P < 0.10$), and by rs408430940 genotypes throughout lactation ($P < 0.05$), and. *PRL* gene SNPs had no effect upon the oth-

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