SMARTER

SMAll RuminanTs breeding for Efficiency and Resilience

Research and Innovation action: H2020 – 772787
Call: H2020-SFS-2017-2
Type of action: Research and Innovation Action (RIA)
Work programme topic: SFS-15-2016-2017
Duration of the project: November, 1 2018 – June, 30 2023

Scientific publications on the genetic characterization, demography and adaptation of local small ruminants breeds

Szilvia Kusza 1, Pablo Peraza2, Johanna Ramirez Diaz3, Alessandra Stella3, Alexandros Tryantafyllidis4,

Valentina Tsartsianidou4, Bertrand Servin 5*

1 Unideb; 2 INIA-UY; 3 CNR; 4 AUTH; 5 INRAE

*** Deliverable leader – Contact: bertrand.servin**@inrae.fr

DELIVERABLE D4.4

Workpackage N°4

Due date: M56

Actual date: 25/03/2024 (previous 25/08/2023)

Dissemination level: Public

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 772787

About the SMARTER research project

SMARTER will develop and deploy innovative strategies to improve Resilience and Efficiency (R&E) related traits in sheep and goats. SMARTER will find these strategies by: i) generating and validating novel R&E related traits at a phenotypic and genetic level ii) improving and developing new genome-based solutions and tools relevant for the data structure and size of small ruminant populations, iii) establishing new breeding and selection strategies for various breeds and environments that consider R&E traits.

SMARTER with help from stakeholders chose several key R&E traits including feed efficiency, health (resistance to disease, survival) and welfare. Experimental populations will be used to identify and dissect new predictors of these R&E traits and the trade-off between animal ability to overcome external challenges. SMARTER will estimate the underlying genetic and genomic variability governing these R&E related traits. This variability will be related to performance in different environments including genotype-by-environment interactions (conventional, agro-ecological and organic systems) in commercial populations. The outcome will be accurate genomic predictions for R&E traits in different environments across different breeds and populations. SMARTER will also create a new cooperative European and international initiative that will use genomic selection across countries. This initiative will make selection for R&E traits faster and more efficient. SMARTER will also characterize the phenotype and genome of traditional and underutilized breeds. Finally, SMARTER will propose new breeding strategies that utilise R&E traits and trade-offs and balance economic, social and environmental challenges.

The overall impact of the multi-actor SMARTER project will be ready-to-use effective and efficient tools to make small ruminant production resilient through improved profitability and efficiency.

Table of contents

1 Summary

The SMARTER Work Package 4 put together a database of genotypes and associated covariates for many breeds of small ruminants (sheep and goats) around the world and in Europe in particular (see Deliverable 4.1). Analyses of these data have taken place within SMARTER to understand the demography and structure of small ruminant species (see report in Deliverable 4.2) and identify genomic regions associated to selection and local adaptation, in particular for hardy and underutilized breeds (see report in Deliverable 4.3). Due to COVID restrictions and data production delays, these analyses could not be included in published scientific papers by the end of the project as initially planned. However, at the end of the project, work is ongoing on 8 manuscripts and one is published. This deliverable lists these manuscripts (published, submitted, under preparation) by the different partners of the project.

2 List of scientific papers under preparation using population genomics approaches on the SMARTER data.

The projected papers are listed by alphabetical order of the first author. All authors lists, titles and abstracts are provisional. Manuscripts are expected to be deposited to preprint servers and/or submitted to peer reviewed journals by December, 31st 2023.

1. Beatriz Carracelas, Pablo Peraza, Brenda Vera, Gabriel Ciappesoni - *Exploring genetic diversity and structure of a Creole sheep population from Uruguay*
2. Manunza, A., Ramirez-Diaz, J., Cozzi, P. *et al.* Genetic diversity and historical demography of underutilised goat breeds in North-Western Europe. *Sci Rep* **13**, 20728 (2023). https://doi.org/10.1038/s41598-023-48005-8

   Abstract. In the last decade, several studies aimed at dissecting the genetic architecture of local small ruminant breeds to discover which variations are involved in the process of adaptation to environmental conditions, a topic that has acquired priority due to climate change. Considering that traditional breeds are a reservoir of such important genetic variation, improving the current knowledge about their genetic diversity and origin is the first step forward in designing sound conservation guidelines. The genetic composition of North-Western European archetypical goat breeds is still poorly exploited. In this study we aimed to fill this gap investigating goat breeds across Ireland and Scandinavia, including also some other potential continental sources of introgression. The PCA and Admixture analyses suggest a well-defined cluster that includes Norwegian and Swedish breeds, while the crossbred Danish landrace is far apart, and there appears to be a close relationship between the Irish and Saanen goats. In addition, both graph representation of historical relationships among populations and f4-ratio statistics suggest a certain degree of gene flow between the Norse and Atlantic landraces. Furthermore, we identify signs of ancient admixture events of Scandinavian origin in the Irish and in the Icelandic goats. The time when these migrations, and consequently the introgression, of Scandinavian-like alleles occurred, can be traced back to the Viking colonisation of these two isles during the Viking Age (793-1066 CE). The demographic analysis indicates a complicated history of these traditional breeds with signatures of bottleneck, inbreeding and crossbreeding with the improved breeds. Despite these recent demographic changes and the historical genetic background shaped by centuries of human-mediated gene flow, most of them maintained their genetic identity, becoming an irreplaceable genetic resource as well as a cultural heritage.

3. Antonis Otapasidis, Valentina Tsartsianidou, Giorgios Arsenos, Alexandros Triantafyllidis - *Genome-wide diversity and homozygosity/heterozygosity patterns of Greek goats. To be submitted by the end of February 2024*

   Goats are of major importance for the livestock sector in Greece. The national herd consists of two indigenous breeds, the Eghoria and Skopelos. We studied the genome-wide structure, the inbreeding and genetic diversity levels of these two Greek breeds using the GoatSNP50 BeadChip (70 and 289 genotyped samples respectively). We characterized the homozygosity and heterozygosity patterns (ROH/ROHet) of the Greek goat genome, and we detected ROH/ROHet hotspots potentially representing candidate signatures of selection. Principal component analysis of Greek goats in comparison to 21 global goat breeds from the AdaptMap project showed breed and farm level genetic differentiation and revealed Greek goat genetic relatedness mostly with the Italian goats (Garganica, Maltesse), other Mediterranean and the African Toggenburg breed. High levels of average expected (He= 0.395-0.423) and observed (Ho= 0.391-0.4203) heterozygosity were estimated for Greek national goat herd. We identified a common ROH region (~0.7 Mb) in Greek goats on chromosome 18 associated with immune response while higher number of ROH estimated in total for Skopelos compared to Eghoria goat genome. We also found 4 ROHet islands for Skopelos and 6 for Eghoria goat, where 80 and 103 genes were located respectively. An annotation analysis detected biological processes related to metabolic and immune responses. This study presents a genome-wide extensive analysis of the Greek goat and reveals their genetic relatedness and differences with previously studied worldwide breeds. Inbreeding estimation and characterization of homozygosity/heterozygosity status can contribute to future genetic improvement schemes and conservation practices.

4. Lucy Peters, Bertrand Servin – *A new method to characterize the genome-wide distribution of adaptive effects from multiple population data.*

   Detecting signatures of selection across the genome is a central objective in population genetics and of great importance in the field of evolutionary biology, but also in commercial settings, such as animal breeding. Deciphering the evolutionary history of a species not only contributes to our understanding of evolution in general, but also informs animal breeding programs. To distinguish patterns of selection from neutral processes such as genetic drift, an appropriate null model that can account for complex demographic histories is needed. The FLK statistic uses a phylogenetic estimation of the populations' kinship matrix, which integrates historical branching and heterogeneity of genetic drift. Here we extend the FLK method and use a Bayesian approach to obtain a new test statistic for selection that specifies an explicit alternative model and also enables us to estimate adaptive effect sizes. By computer simulations, we show the ability of the method to estimate the distribution of selective effects and compare it to a previously proposed approach. Finally, we illustrate the properties of the method on a high-density dataset of French Sheep populations.

5. Johanna Ramirez Diaz, Arianna Manunza, Paolo Cozzi, Filippo Biscarini, Gabriel Ciappesoni, Pablo Peraza, Juan Carlos Rincon Florez, Luz Angela Alvarez, Gerson Barreto Mourao, Juan Jose Arranz, Fernando Freire, Valentina Tsartsianidou, Alexandros Triantafyllidis, Bertrand

Servin , Alessandra Stella - *Patterns of genetic variation along environmental gradients in underutilised sheep and goat worldwide breeds.*

6. Valentina Tsartsianidou, Kostas Gkagkavouzis, Karaiskou N., Vouraki S., Chatziplis D., Banos G., Girogios Arsenos , Alexandros Triantafyllidis, *The genomic makeup of native Greek sheep breeds reveals population and admixture patterns linking European with Asian domestic breeds*

   Greece hosts one of the largest and highly diverse pools of animal genetic resources in the Mediterranean region, where numerous sheep breeds have been regionally selected for centuries. The present study focuses on the genome-wide characterization of population structure, diversity, and admixture patterns of local Greek domestic sheep collectively analyzed with 38 global breeds from the Sheep HapMap project. A total of 1,008 animals (Chios=173, Frizarta=367, Boutsko=300, Mytilini=108, Pelagonia=60) from 23 locations in mainland and island Greece were sampled and genotyped utilizing the OvineSNP50 Genotyping BeadChip featuring 52,152 SNP markers. Principal component analysis and model-based clustering depicted a central position for Greek sheep among worldwide sheep as an intermediate genetic link between Asia and Europe, indicating cross-continent and within Europe genetic clines. Genetic clusters of Asian fat-tailed and Balkan Zackel sheep were identified as major contributors in Greek breed formation. Additionally, genetic relatedness between Greek sheep revealed several admixture and crossbreeding events for human-mediated selection purposes, while the studied sheep were well differentiated at breed level. Average genetic diversity levels of Greek breeds, explained by heterozygosity indices (Ho = 0.358 and He = 0.363), were estimated similar to Mediterranean sheep. Long-term directional selection was characteristically depicted for Greek Chios sheep with higher genetic differentiation (FST=0.09-0.15). These results suggest a diverse and composite genetic makeup of Greek sheep indicating multiple migration routes during sheep domestication and expansion from Asia to Europe, while inferred admixture patterns are related to productivity and adaptation enhancement.

7. Valentina Tsartsianidou, Vouraki S., Girogios Arsenos, Alexandros Triantafyllidis - *Genomic signatures of adaptive response driven by vertical transhumance practices and seasonal precipitation in mountainous Boutsko sheep*

   Current and future climate fluctuations combined with selected livestock populations for production purposes raise major concerns about the preservation of animal genetic resources and food security. This study aimed to identify signatures of adaptive selection related to the impact of environmental heterogeneity on the genome of Boutsko, a Greek native transhumant sheep. Global positioning system (GPS) devices were used to monitor a total of 296 purebred Boutsko sheep reared semi-extensively during summer in three flocks located in Northwestern Greece. These populations were georeferenced based on the farm coordinates for lowland winter locations and genotyped with the Ovine SNP50K Bead Chip v2. A genotype-environment association (GEA) analysis was conducted with high and low-altitude grazing locations implementing logistic regressions. Fifteen SNPs were significantly associated with the bioclimatic variable of precipitation seasonality (bio15) representing the variation of monthly precipitation totals over the course of the year. Five candidate SNPs located on chromosomes 6, 15 and 22 were positioned within annotated genomic regions and their spatial genotype distribution coincided with geographic variation of precipitation seasonality. Functional enrichment analyses detected gene sets related to climate response, such as hypoxia response, energy metabolism, cell proliferation and differentiation and body conformation traits. The association between specific genomic loci and precipitation variation in the contrasting eco-climatic regions could be attributed to water and relevant pasture availability and composition within grazing areas seasonally and altitudinally. These results can enhance the incorporation of adaptive-related SNPs in multi-trait selective breeding programs to face projected climate change.

8. George Wanjala, Zoltán Bagi, Putri Kusuma Astuti, Elena Ilisiu, Dinu Gavojdian, Husein Ohran, Eva Pasic Juhas, Bouabid Badaoiu, Dimitrios Loukovitis, Aldona Kawecka, Ruta Sveistiene, Beckei Zolt, Péter Strausz, Nelly Kichamu, Szilvia Kusza - *A genome-wide survey provides insights into the genetic evolution and climate change adaptation of native sheep breeds from different climatic zones* ***Submitted to Genetics Selection Evolution***

3 Conclusion

As described in details in D4.2 and D4.3, analyses performed during the project have led to a better understanding of the genetic structure and history of small ruminants and the identification of key genomic regions linked to their adaptations. At the end of the project, these works, performed by the partners of the SMARTER project are being prepared for publication in scientific journals.

4 Deviations or delays

The initial commitment was to produce at least one scientific paper presenting the population genetics analyses of the SMARTER database. Because of the COVID delays in gathering data, the unresponsiveness of some partners, and despite the extension granted, the production of such a paper was severely delayed. However, as is shown in the reports of deliverable 4.2 and 4.3, many analyses have progressed.

To date (March 2024), one paper has been accepted (Manunza et al., Sci Rep 2023) and the paper by Wanjala et al. has been submitted to Genetics Selection Evolution.

Other manuscripts are currently still under preparation for dissemination to the scientific community.