

software was used to identify CNV on 28 chromosomes, and the R package HandyCNV was used to generate and visualise the CNVR, the chromosome location and the gene annotation.

A total of 4266 CNV were identified with the highest number in Ancona (480), Valplatani (408) and Bianca di Saluzzo (348), and the lowest in Polverara Nera (41) and Robusta Lionata (40). To make the interbreed CNVR frequency comparison possible, core CNVR were identified using CNV that had overlapping chromosomal locations in different breeds. This resulted in 292 interbreed CNVR with frequency >15% in at least one breed. Chicken quantitative trait loci datasets and Ensembl gene annotations were used to estimate potential phenotypic effects of CNVR on breed-specific traits. Various CNVR identified in the present study overlapped with genes involved in skeletal muscle development and function, fatty acid deposition, heat stress response and climate change adaptation.

The distribution of the CNV and the comparison of the differences among the CNVR of the 23 local chicken breeds reported in the present study allowed a comprehensive characterization of the breeds. However, further research is needed to confirm if the CNV observed in the breeds are also linked to variations in the specific phenotypic aspects. Further experimental and functional validation of CNVR would help in the conservation of Italian local chicken breeds.

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# Mapping of heterozygosity-rich regions in Italian and worldwide goat populations

Giorgio Chessari<sup>a</sup>, Andrea Criscione<sup>a</sup>, Paola Crepaldi<sup>b</sup>, Donata Marletta<sup>a</sup>, Baldassare Portolano<sup>c</sup>, Filippo Biscarini<sup>d</sup> and Salvatore Mastrangelo<sup>c</sup>

<sup>a</sup>Dipartimento Agricoltura, Alimentazione e Ambiente, University of Catania, Catania, Italy

<sup>b</sup>Dipartimento di Scienze Agrarie e Ambientali, University of Milan, Milano, Italy

<sup>c</sup>Dipartimento di Scienze Agrarie, Alimentari e Forestali, University of Palermo, Palermo, Italy

<sup>d</sup>CNR, Institute of Agricultural Biology and Biotechnology, Lodi, Italy

The heterozygosity-rich regions (HRR) are genomic regions with high variability, which can provide information on populations' diversity and may harbor genes related to functional traits such as immune response, adaptation or fertility. To our knowledge, there are few studies characterizing HRR in goat species. Italy provides a high source of information for caprine genetic resources with its 36 recognized breeds, mainly reared in small farms, in semi-extensive systems, and exploiting marginal areas. Italian (n=30) and Worldwide (n=19) goat populations, for a total of 1287 individuals genotyped with the Illumina GoatSNP52k

BeadChip, were investigated for population structure and HRR patterns using both Sliding Windows (SW) and Consecutive Runs (CS) methods (common parameters: minSNP = 10, no missing or opposite genotypes and minLength =250kb). The amongbreeds relationship analyses clustered the populations in accordance with their geographical distribution, highlighting Asiatic goats as the most distant cluster. The HRR analysis reported overlapping results by both detection methods with high correlation coefficients (r = 0.996-1) for the descriptive parameters of number, length and diversity rate. The Alpine populations showed the highest mean number of HRR  $(N_{HRR})$  per individual (12.70) followed by the goats of central-southern Italy (11.59), while African and Asian goats showed relative low values (8.60 and 4.88, respectively). CR and SW reported the same total number of HRR (1130) corresponding to 166 HRR islands. CHI01, CHI11, CHI12 and CHI18 showed 66% of the islands, involving 33, 16, 22 and 30 populations, respectively. In particular, 67% of the populations shared the same hotspot in CHI01, including STAG1 and PCCB reproduction-related genes. Almost all the Italian and Alpine goats had the CHI18 island in common, related to the fiber production from fleece. The hotspot in CHI12 shared by some Alpine and Italian, as well as African and Asian breeds, harbor genes involved in productive and adaptation to climate traits. The results highlighted similar genomic patterns between geographically close breeds probably due to gene flow. Moreover, HRRs across breeds and common HRR hotspots underlined a high level of genetic diversity that could give this species the possibility of facing new challenges linked to the onset of diseases or climate change.

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### **O374**

## An ancient DNA perspective on the genomic variation of present day Sardinian cattle

Licia Colli<sup>a</sup>, Fabio Correddu<sup>b</sup>, Mariantonietta Palazzo<sup>a</sup>, Elisa Somenzi<sup>a</sup>, Elisa Eufemi<sup>a</sup>, Alberto Cesarani<sup>b</sup>, Marco Zedda<sup>c</sup> and Nicolò Pietro Paolo Macciotta<sup>b</sup>

<sup>a</sup>DIANA Dipartimento di Scienze Animali, della Nutrizione e degli Alimenti, Università Cattolica del S. Cuore, Piacenza, Italy <sup>b</sup>Dipartimento di Agraria, Sezione Scienze Zootecniche, Università degli Studi di Sassari, Sassari, Italy <sup>c</sup>Dipartimento di Medicina Veterinaria, Università degli Studi di Sassari, Sassari, Italy

The Sarda cattle is an autochthonous breed from the island of Sardinia. This population has never been subjected to artificial selection or strong phenotypic standardization and its evolution has been mainly driven by environmental adaptation, leading to

