



coefficients (FG) in dairy cows. To this purpose, 95,540 Italian Holstein dairy cows with imputed SNP were analyzed. Cows were genotyped with 30 different SNP chips and imputed to 84,445 pre-selected SNP from the routine genomic evaluations of the Italian National Association of Holstein and Jersey Breeders (ANAFJ). All SNP were used, being already filtered for call rate <95%, parent-offspring SNP mismatch >0.01, minor allele (<0.02) and genotype (<0.001) frequencies and extreme deviation from Hardy-Weinberg Equilibrium ( $p < .005$ ). To test the effect of imputation on FG, four medium-density (MD) SNP chips (each with >10k genotyped cows; GeneSeek Genomic Profiler3,4,MD and LabogenaMD) and one high-density (HD) SNP chip (641 genotyped cows; GeneSeek Genomic ProfilerHD-150K) were selected. Moreover, FG was estimated with seven standardized methods: (i) based on homozygosity excess (F and FHAT2), variance of the additively recoded genotypes (FHAT1) and correlation between uniting gametes (FHAT3), available in PLINK v1.9, (ii) VanRaden's genomic relationship matrix (GRM; FGRM1, FGRM2), estimated with an in-house Python script and (iii) runs of homozygosity (ROH) across the genome (FROH), calculated in detectRUNS v. 0.9.5 R package. Pedigree-based inbreeding coefficients were also estimated (FPED). Pearson ( $\rho$ ) and Spearman ( $r$ ) correlations of imputed vs. genotyped cases across SNP panels were calculated. Results are summarized as follows: (i) on average, in the HD SNP panel,  $\rho \approx r \approx 1$  in pairwise comparison (imputed vs. genotyped) across FG; in MD SNP panels,  $\rho < r$  and with higher variability across FG, with LabogenaMD showing the highest consistency ( $\rho = 0.85 [0.77, 0.97]$ ,  $r = 0.95 [0.92, 0.97]$ ), (ii) FROH was most consistent across SNP chip ( $\rho = 0.94 [0.79, 1]$ ,  $r = 0.97 [0.93, 1]$ ), (iii) FROH had the highest relationship with FPED ranging between  $\rho = 0.48-0.83$  and  $r = 0.54-0.84$ , (iv) non-linear trends across pairwise comparisons indicated that  $\rho$  might not well reflect the relationship between FG and FPED;  $r$  provides an alternative estimate when a monotonic relationship exists, and v) re-ranking across FG methods was observed.

## O173

### Inbreeding depression in small ruminants: from pedigree to genomic estimation

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Nowadays genomic data represent an increasingly important tool to exploit the traditional pedigree information in order to create better mating schemes and preserve biodiversity. In the Italian small ruminants populations the use of such information could improve the estimation of genetic population parameters leading to a better

management of the genetic background of the different flocks. In this work we investigate the relationship between different methods of inbreeding calculation from traditional pedigree information (FPED) and from 50k SNP chip genomic data (FROH) on the estimation of inbreeding depression effect on daily milk production. We analyzed pedigree and genomic data from 980 individuals of Camosciata delle Alpi goat breed and 380 individuals of Massese sheep breed. Data were provided by the Italian association of small ruminants breeders (ASSONAPA) within the PSRN project Conservation, Health and Efficiency Empowerment of Small Ruminant (CHEESR). Editing of genomic data and calculation of FROH were performed with the PLINK software, while calculation of FPED was performed with the R package Optisel. The relationship between the two inbreeding indicators and daily milk production were calculated using two different GLM models.

The results showed a similar mean value of FROH in the two populations sampled, 6.6% in Massese and 5.4% in Camosciata, but the mean FPED value in Massese (3.9%) was more than double than in Camosciata (1.5%). As expected, the correlation between FPED and FROH was much lower in Camosciata (0.19) than in Massese (0.39). In Camosciata, 30.6% of FPED values equal to zero were unreliable (they exceeded the UIC 95% of the intercept). In terms of inbreeding depression effect (%) on daily milk production, daily milk production decreased by  $-0.017 (+ -0.005)$  in Camosciata and  $-0.015 (+ -0.018)$  in Massese using FROH and by  $-0.005 (+ -0.005)$  and  $-0.049 (+ -0.03)$ , respectively, using FPED. What emerges from these analyses is that, in populations with low average inbreeding, there is a similar inbreeding depression based on FROH estimate, but the completeness and validity of the pedigree recordings greatly affect the values of FPED. Based on that, the reliability of the pedigree information has to be fully evaluated in the estimation of the inbreeding depression effect.

## O174

### SNP-based characterization of Montecristo feral goats in a Mediterranean perspective

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The Montecristo wild goat is an endangered free-ranging goat population inhabiting the homonymous island in the Tuscan Archipelago. The origin of this feral population is still debated:

some authors suggest that goats were introduced during prehistoric times while others date their first occurrence between the 6th and 13th century of the Common Era when Montecristo island was occupied by a monk community. In this study, we assessed genetic diversity and population structure of two different nuclei of Montecristo goats, one sampled on the island, the other from an ex-situ conservation project on the Italian mainland. Furthermore, to investigate the origin and evolutionary relationships of Montecristo goats we assembled a 50K SNPs dataset including 44 breeds from the Mediterranean basin. Observed ( $H_o$ ) and expected ( $H_e$ ) heterozygosity were calculated to estimate diversity levels, while gene flow, population structure and relationships were assessed through multiple approaches, i.e. runs of homozygosity (ROHs), Principal Component Analysis (PCA), Neighbour-network reconstruction, Treemix software analysis.

Values of  $H_o$  and  $H_e$  for the insular Montecristo population were the lowest recorded in the dataset, highlighting reduced genetic diversity, while the nucleus from the mainland displayed a less severe reduction. Results from PCA, Neighbour-network and population structure analysis clearly separated the insular population from all other breeds, and further highlighted a remarkable distance between the two Montecristo nuclei. Also, Treemix software analysis pinpointed possible genetic contributions received by the two Montecristo goat populations from different sources. Lastly, ROHs indicated the occurrence of an ancient bottleneck or founder effect in the insular population, and of recent inbreeding in the continental nucleus.

Taken together, our results indicate that Montecristo goats faced several demographic fluctuations combined with admixture events over time, and also emphasized the marked genetic differentiation between the goats sampled on the island and those from the ex-situ nucleus. This evidence can represent a starting point for the implementation of a marker-assisted monitoring and conservation plans for the autochthonous feral goat of Montecristo.

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### O175

#### A comparative analysis of pedigree- and genomic-derived inbreeding parameters in Italian heavy pig breeds over the last decades

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Inbreeding is an important parameter that is monitored in selection and conservation programs in livestock, as an increased inbreeding

rate leads to increasing depression and to an increased frequency of deleterious alleles. Traditionally, the inbreeding coefficient is calculated using pedigree information and it is defined as FPED. Single nucleotide polymorphism (SNP) genotyping panels allow to estimate genomic inbreeding by identifying runs of homozygosity (ROH) that are long chromosome regions in which all adjacent SNPs are homozygous. Genomic inbreeding (FROH) is calculated as the fraction of the autosomal genome covered by ROH. The breeding program of Italian pig breeds started about 30 years ago and pedigree data are recorded at a depth that reaches more than 20 generations. In this work, we retrospectively analysed FROH over this period in Italian Large White (ILW), Italian Duroc (ID) and Italian Landrace (IL) heavy pig breeds and compared this parameter with FPED. A total of 3055 ILW pigs, 758 ID pigs and 1940 IL pigs born over the last 25 years were genotyped with the Illumina PorcineSNP60 BeadChip and with Illumina GGP Porcine HD arrays. Quality filtering and ROH identification were obtained with PLINK version 1.9. Animals were grouped by year of birth and groups with a minimum of 10 animals were considered. The averaged FROH and FPED were computed for each group and for each breed. FROH was higher in ID (the highest averaged FPED in this breed was 0.12) with respect to the other two breeds (0.07 in ILW and 0.06 in IL) and was constant over all analysed time windows. In the considered period, the two increasing trends of FROH and FPED were significantly different in ILW and in ID breeds with a higher rate for the FPED mean values. In IL, the increasing inbreeding rate of FPED was stronger with respect to FROH, even if not significantly different. The results indicated that mean FROH in all three breeds over the years was more stable than FPED. FROH can be an effective parameter that could substitute FPED in breeding and conservation plans.

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### O176

#### The impact of inbreeding in the Italian Holstein breed

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The Italian Holstein breed has been exposed to intense selection for milk production traits over the last 50 years, leading to an average of over 10,000 kg of milk produced per cow per lactation.