

SESSION 35 GENETIC DIVERSITY

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 (ANAFIJ). 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 (ρ) 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, $\rho \approx r$ ≈ 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 = 0.54-0.830.84, (iv) non-linear trends across pairwise comparisons indicated that ρ 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.

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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.

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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:

