OP144   Investigating introgression of river-buffalo loci in the genome of Brazilian Carabao swamp buffaloes. M. Barbato1, L. Colli2, M. Milanesi3, Y. T. Usunomiy4, J. R. V. Herrera1, L. Cruz5, P. Baruselli5, M. M. J. Amaral5, M. G. Drummond6, J. F. Garcia1, J. L. W. Williams2, International Buffalo Consortium3, and P. Ajmone-Marsan4.1Department of Animal Science, Food and Nutrition – DIANA, Università Cattolica del S. Cuore, Piacenza, Italy, 2Universidade Estadual Paulista “Julio de Mesquita Filho,” Faculdade de Medicina Veterinária de Araçatuba, Araçatuba, Brazil, 3International Atomic Energy Agency (IAEA), Collaborating Centre on Animal Genomics and Bioinformatics, Araçatuba, Brazil, 4Philippine Carabao Centre, Nueva Ecija, Philippines, 5Faculdade de Medicina Veterinária e Zootecnia, Universidade de São Paulo, São Paulo, Brazil, 6Instituto de Biociências, Letras e Ciências Exatas, Universidade Estadual Paulista, São José do Rio Preto, Brazil. 

Water buffalo was domesticated about 5,000–6,000 years ago and since then has become one of the most important livestock species in tropical and sub-tropical environments providing labor, meat and dairy products. There are 2 water buffalo species, swamp and river buffalo, which differ in morphology, production attitudes, geographic range and chromosome number. Despite the difference in chromosome number, these 2 subspecies are inter-fertile and the river type is sometimes used to upgrade milk production of the swamp type. Here we investigated admixture and local ancestry of 2 populations: Brazilian Carabao, which were established hundreds of years ago in Brazil, and the recently created Philippine cross bred population. Data were produced using the 90K buffalo SNPs array (Axiom Buffalo Genotyping Array – ThermoFisher scientific). In addition, worldwide pure river and swamp breeds were genotyped and used as reference. We identified shorter river type-derived haplotypes in Carabao compared with those found in the Philippine population, confirming an older event of admixture. Several genomic regions appeared to be conserved among Carabao individuals, which are putatively associated with adaptation and production traits. Local ancestry analysis revealed preferential introgression of river loci into the swamp genome in regions carrying genes related to dairy production and fertility.

Key Words: river buffalo, swamp buffalo, introgression, admixture


Polymorphic markers on the male-specific region of the Y chromosome (MSY) are widely used to infer historic demography. Because of its lack of recombination and the strictly male specific inheritance, the MSY perfectly mirrors male genealogies. However, the low sequence variation on the horse MSY impeded fine-grained haplotyping in this species. We recently established a workflow to predict the regions on the highly repetitive Y-chromosome that are suitable for unambiguous variant calling from NGS data. We generated a MSY haplotype tree based on biallelic variants ascertained from more than 200 horses on about 6 MB of the MSY. This robust MSY topology served as a backbone for a haplotype determination in a comprehensive data set. By linking MSY haplotypes and pedigree information, we nailed down the MSY haplotypes of several influential founder stallions – ranging from the Thoroughbreds, to Arabian, Spanish and some coldblooded sires. We show the potential of MSY haplotyping to trace the influence of particular sires within and across populations and give examples how MSY haplotyping can be revealing to illuminate the development and origin of horses by studying rural populations and historic/ancient remains. Our effort brings the accuracy of Y-chromosomal lineage tracing in horses similar to that in humans and therefore sire line screening could be implemented for forensic applications.

Key Words: horses and related species, phylogeny, sequence variation, Y chromosome, forensics

OP146   Introgression with domestic goats has expanded the genetic variability of the Spanish ibex. T. Figuereo-Cardoso1, R. Tonda1, M. G. Luis-Sierra2, A. Castello1, B. Cabrera1, A. Noce1, S. Beltran1, R. Garcia-Gonzalez1, A. Fernandez-Arias1, J. Felch1, A. Sanchez1, A. Clop1, and M. Amills1,2,3, *Centre for Research in Agricultural Genomics (CRAG), CSIC-IRTA-UAB-UB, Universitat Autonoma de Barcelona, Bellaterra, Barcelona, Spain, 2Centre Nacional d’Anàlisi Genòmica-Centre for Genomic Regulation (CRG), Bellaterra, Barcelona, Spain, 3Universitat Autonoma de Barcelona, Bellaterra, Barcelona, Spain, 4Instituto Pirenaico de Ecología (IPECSIC), Spain, 5Servicio de Investigacion Agroalimentaria, Spain, 6Centro de Investigación y Tecnología Agroalimentaria de Aragón, Zaragoza, Zaragoza, Spain.

The Spanish ibex (Capra pyrenaica) is a wild goat species distributed in the Iberian Peninsula. Based on phenotypic criteria, 4 subspecies have been defined: C. p. hispanica (CPH, south and east of the Iberian Peninsula), C. p. victorina (CPV, center and northwest of the Iberian Peninsula), C. p. lusitana (CPL, Galicia and north of Portugal) and C. p. pyrenaica (CPY, Pyrenees mountains). Hunting, epidemics and habitat loss caused the extinction of CPH (disappeared in the 19th century) and CPP (extinct in the year 2000) as well as severe population bottlenecks decreasing the diversity of CPV and CPH. By using a high throughput genotyping approach, we have demonstrated that interspecific hybridization with domestic goats has been an important source of novel variability for Spanish ibexes living in Tortosa-Beceite. Individual sequencing of one of the last CPP representatives (×16.6 coverage) and Pool-sequencing (×39 coverage) of 30 CPV and 23 CPH individuals revealed an extensive sharing of SNPs (96%) between the CPH individual and the extant CPV and CPH subspecies, thus suggesting that the extinction of CPP did not cause a major loss of diversity in Capra pyrenaica. Sequencing experiments also revealed that the genome of one of the last CPP representatives contains stop-gained mutations, with heterogeneous genotypes, in the WASF2, RBM17 and SERPINB10 genes. The inactivation of WASF2 and RBM17 causes embryonic lethality, while SERPINB10 belongs to a family of serin proteases with key roles in immunity and other biological processes. Our results suggest that the dramatic reduction of the CPP population during the 19th-20th centuries led to the progressive accumulation of mutations with harmful effects (genomic meltdown) that probably contributed to its extinction by limiting fitness and reproductive success.

Key Words: conservation, hybridization, goats and related species

OP147   Unraveling the genomic diversity and population structure of 20 autochthonous European pig breeds. M. Muñoz4, R. Bozzi1, J. M. García-Casco2, Y. Núñez2, A. Ribani3, M. Skrip2, R. Quintanilla1, M. J. Mercat2, J. Rigue2, J. Estelle2, M. Candek-Potokar1, A. I. Fernández2, L. Fontanesi4, and C. Ovilo4. 1Departamento Mejora Genética Animal, INIA, Madrid, Spain, 2DAGRI, Animal Science Section, Universidad de los Studi de Firenze, Firenze, Italy, 3Department of Agricultural and Food Sciences, University of Bologna, Bologna, Italy, 4Knetijski institut Slovenije, Ljubljana, Slovenia, 5IRTA, Programa de Genética y Mejora Animal, Barcelona, Spain, 6IFIP – Instituto del Porc, Le Rheu, France, 7INRA, Génétique Physiologie et Système

The Spanish ibex (Capra pyrenaica) is a wild goat species distributed in the Iberian Peninsula. Based on phenotypic criteria, 4 subspecies have been defined: C. p. hispanica (CPH, south and east of the Iberian Peninsula), C. p. victorina (CPV, center and northwest of the Iberian Peninsula), C. p. lusitana (CPL, Galicia and north of Portugal) and C. p. pyrenaica (CPY, Pyrenees mountains). Hunting, epidemics and habitat loss caused the extinction of CPH (disappeared in the 19th century) and CPP (extinct in the year 2000) as well as severe population bottlenecks decreasing the diversity of CPV and CPH. By using a high throughput genotyping approach, we have demonstrated that interspecific hybridization with domestic goats has been an important source of novel variability for Spanish ibexes living in Tortosa-Beceite. Individual sequencing of one of the last CPP representatives (×16.6 coverage) and Pool-sequencing (×39 coverage) of 30 CPV and 23 CPH individuals revealed an extensive sharing of SNPs (96%) between the CPH individual and the extant CPV and CPH subspecies, thus suggesting that the extinction of CPP did not cause a major loss of diversity in Capra pyrenaica. Sequencing experiments also revealed that the genome of one of the last CPP representatives contains stop-gained mutations, with heterogeneous genotypes, in the WASF2, RBM17 and SERPINB10 genes. The inactivation of WASF2 and RBM17 causes embryonic lethality, while SERPINB10 belongs to a family of serin proteases with key roles in immunity and other biological processes. Our results suggest that the dramatic reduction of the CPP population during the 19th-20th centuries led to the progressive accumulation of mutations with harmful effects (genomic meltdown) that probably contributed to its extinction by limiting fitness and reproductive success.