

generations to increase the frequency of the adapted alleles. Besides this genetic-based response short-term mechanisms based on phenotypic plasticity may mediate the response to environmental stresses. Phenotypic plasticity, which is the ability of a same genotype to express different phenotypes in different environments, may rely on the presence of epigenetic marks (such as DNA methylation) in the genomes, that regulate gene expression. This presentation focuses on the environment-related variation of DNA methylation patterns along the genome in goat (*Capra hircus*) and sheep (*Ovis aries*), living in field conditions in Morocco. For each species, we studied 2 groups of animals from environments with contrasted ambient temperatures (desert vs. Mediterranean climates). For this purpose, individuals methylomes were generated by sequencing of DNA methylated fragments, previously retrieved by immunoprecipitation. Then, we identified 5 and 2 differentially methylated genomic regions between the 2 groups for goat and sheep, respectively. We didn't find any homologous regions that are differentially methylated between the 2 species. Finally, we identified 4 genes for goats and 2 genes for sheep that could be differentially expressed in relation to the variation of ambient temperatures.

**Key Words:** DNA methylation, *Ovis aries*, *Capra hircus*, local acclimation

**P448 Introgression with domestic goats has expanded the genetic variability of the Spanish ibex.** T. Figueiredo-Cardoso<sup>1</sup>, R. Tonda<sup>2</sup>, M. G. Luigi-Sierra<sup>1</sup>, A. Castelló<sup>1,3</sup>, B. Cabrera<sup>1,3</sup>, A. Noce<sup>1</sup>, S. Beltrán<sup>2</sup>, R. García-González<sup>4</sup>, A. Fernández-Arias<sup>5</sup>, J. Folch<sup>6</sup>, A. Sánchez<sup>1,3</sup>, A. Clop<sup>1</sup>, and M. Amills<sup>\*1,3</sup>, <sup>1</sup>Centre for Research in Agricultural Genomics (CRAG), CSIC-IRTA-UAB-UB, Universitat Autònoma de Barcelona, Bellaterra, Barcelona, Spain, <sup>2</sup>Centre Nacional d'Anàlisi Genòmica-Centre for Genomic Regulation (CRG), Barcelona, Barcelona, Spain, <sup>3</sup>Universitat Autònoma de Barcelona, Bellaterra, Barcelona, Spain, <sup>4</sup>Instituto Pirenaico de Ecología (IPE-CSIC), Spain, <sup>5</sup>Servicio de Investigación Agroalimentaria, Spain, <sup>6</sup>Centro 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. victoriae* (CPV, center and northwest of the Iberian Peninsula), *C. p. lusitanica* (CPL, Galicia and north of Portugal) and *C. p. pyrenaica* (CPP, Pyrenees mountains). Hunting, epidemics and habitat loss caused the extinction of CPL (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 ( $\times 16.6$  coverage) and Pool-sequencing ( $\times 39$  coverage) of 30 CPH and 23 CPV individuals revealed an extensive sharing of SNPs (96%) between the CPP 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 heterozygous 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

**P449 A de novo mutation causes polledness and a modified shape of the skull in Fleckvieh cattle.** L. Gehrke<sup>1</sup>, M. Upadhyay<sup>\*2</sup>, K. Heidrich<sup>2</sup>, E. Kunz<sup>2</sup>, D. Seichter<sup>3</sup>, A. Graf<sup>2</sup>, S. Krebs<sup>2</sup>, A. Capitan<sup>4</sup>, G. Thaller<sup>1</sup>, and I. Medugorac<sup>2</sup>, <sup>1</sup>Christian-Albrechts-University Kiel,

Kiel, Schleswig-Holstein, Germany, <sup>2</sup>Ludwig Maximilians University Munich, Munich, Bavaria, Germany, <sup>3</sup>Tierzuchforschung e.V. München, Grub, Bavaria, Germany, <sup>4</sup>GABI, INRA, AgroParisTech, Paris, France.

Genetic heterogeneity refers to the phenomenon where mutations in different loci (locus heterogeneity) or within the same locus (allelic heterogeneity) lead to a similar phenotype. In cattle, allelic heterogeneity is observed for the polled condition. In fact, at least 3 different alleles at the *polled* locus have been identified in cattle. In this study, we describe a case of a polled Fleckvieh bull born to horned parents that also implies locus heterogeneity of polledness. Genotyping of the case bull, its sire, grandsires and its polled and horned offspring was carried out using the bovine50K SNP array to determine the genetic basis of the de novo polledness condition. Additionally, Illumina paired-end and Oxford Nanopore sequencing technologies were employed to identify the exact candidate mutation for the polledness. Later, sanger sequencing technology was also used to validate the candidate mutation. The approach identified an 11-bp de novo deletion as the candidate mutation for the polled condition that first arose in a Fleckvieh bull and later passed onto its offspring. The 11-bp deletion event encompassed the second exon of the *ZEB2* gene and led to a translational frameshift. The frameshift caused a premature termination of translation, leading to a truncated protein. Compared with the wild type, the truncated *ZEB2* protein is predicted to be shortened by about 91%. Mutations in the *ZEB2* gene cause multiple congenital anomalies in humans as well as in cattle. However, apart from displaying polledness, a modified shape of the skull and presumably a short stature, the individuals carrying the 11-bp deletion in *ZEB2* gene did not display any other clinical symptoms. Because the *ZEB2* gene encodes a Smad Interacting Protein 1 (SIP1) that plays a vital role in epithelial-mesenchymal transition, it can be hypothesized that the truncated *ZEB2* protein might have lacked essential domains associated with the differentiation of horn buds. To conclude, the results of this study point toward a complex genetic pathway involved in bovine polledness that requires further investigation.

**Key Words:** cattle, polledness, *ZEB2* gene, de novo deletion, frameshift

**P450 Follows P231**

**P451 Follows P167**

**P454 A draft genome of Drung cattle (*Bos frontalis*) and its adaption to life at high altitude.** Y. Chen<sup>\*1</sup>, X. Gao<sup>1</sup>, J. Li<sup>1</sup>, T. Zhang<sup>1</sup>, W. Yang<sup>2</sup>, W. G. Zhang<sup>1</sup>, and B. Su<sup>3</sup>, <sup>1</sup>Institute of Animal Science, Chinese Academy of Agricultural Sciences, Beijing, China, <sup>2</sup>The Co., Ltd. of 1 Gene Technology, Hangzhou, China, <sup>3</sup>Drung Cattle Nature Reserve, Gongshan, Yunnan China.

Drung cattle (*Bos frontalis*), locally called "Panda cattle," is a unique semi-wild bovine species that mainly inhabits the rain-forests of "Grand Canyon of the East" located in the Gaoligong Mountains and Drung River Basin of Yunnan Province, China. Here, we reported the ~2.74 Gb draft genome sequence of an adult male Drung cattle using Illumina, PacBio and 10X Genomics sequencing platforms. Compared with the previous gayal genome assembly, the scaffold N50 raised from 2.74 Mb to 4.08 Mb and the contig N50 boosted from 14.41 kb to 157.67 kb, respectively corresponding to ~2.6-fold and 10.9-fold improvement. Speciation time estimation showed that Drung cattle diverged ~1.20 Mya earlier than the clade of domestic cattle, indicating that *Bos frontalis* is clearly distinct from *Bos taurus*. This result strongly supports the contention that gayal is not the modern domestic cattle in lineage. To characterize the mechanisms underlying a chromosome fusion event leading to the formation of gayal chromosome 1, we analyzed bovine satellite I sequences and found concentrated tandem repeat regions located at the terminal end of 2 chromosomes in *Bos taurus*, corresponding to 29 satellite repeats at BTA2 and 57 repeats at BTA28, respectively. In our Drung cattle assembly, we identified one scaffold sequence (Frag scaffold60) having the presence of cattle satellite I with 28 tandem repeats, in which 17 genes were detected to be aligned to BTA2