ISAG 2023

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Abstracts

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pects. In addition, dromedaries are so fascinating also from a biological point of view, being among the few big mammals to have evolved specific adaptations to extreme environmental conditions. Under the frame of the 2019 Illumina Agricultural Greater Good (AGG) initiative program which is aimed at supporting studies on sustainability, productivity, and nutritional density of agriculturally important crop and livestock species, a total of 179 dromedaries from the entire geographic distribution range were whole-genome sequenced (WGS). Raw reads were mapped against the dromedary reference genome (CamDro3) and the variants were called using the Illumina Dragen germline platform. From a total of 13,560,911 biallelic SNPs, after a multistep filtering approach aimed at ensuring SNPs evenness across chromosomes and removing SNPs with less than 0.05 minor allele frequency and 0.1 missing call rate, a subset of 61,208 SNPs was selected. The panel included 59,069 autosomic SNPs with an average distance of 32 kb, 1,230 SNPs on X chromosome and 77 mitochondrial SNPs. In addition, about 1,000 of loci from 47 genes with known functional relevance were enriched. The linkage-disequilibrium (LD) decay graph indicated that at r^2 value ranging from 0.3 to 0.5 we found pairs of loci separated 50 kb apart. This value resulted higher than that reported in other cattle commercial breeds and even higher than that observed in sheep breeds. This result confirms that the selection of SNPs with an average distance of 32 kb will perform well in linkage disequilibrium mapping approaches such as in looking for selection signatures or in genome-wide association studies. The panel is currently being validated and we are confident that will represent a further relevant step toward the understanding of dromedary genomics.

Key Words: Old World camelid, genome sequencing, genotyping, single nucleotide polymorphism (SNP)

OP78 Selection of an ovine SNP parentage panel for consideration as the ISAG comparison test panel. R. Ferretti*[1], K. Schutt[2], M. Dowling[2], J. Qiu[1], and R. Tait Jr.[1], [1]*Neogen GeneSeek Operations, Lincoln, NE,* [2]*Neogen Australasia, Ipswitch, QLD, Australia.*

Advances in medium- and high-throughput genotyping platforms have allowed for significant reduction in genotyping costs. A decade ago, this was a prohibiting factor for transitioning away from microsatellites over to single nucleotide polymorphism (SNP) technologies. At the 33rd International Society for Animal Genomics (ISAG) conference in 2012, this topic was raised and initial work was done by the International Sheep Genomics Consortium (ISGC) to adopt an official Ovine SNP comparison test (CT) panel using 88 autosomal SNPs and one male specific SNP. Here we propose an expanded panel of 201 SNP markers for consideration as the accepted ISAG Ovine Parentage CT panel. The aim for this parentage panel was to build off the original ISGC 89 SNP panel by incorporating SNP markers from newer iterations of academic and commercial parentage panels. Furthermore, to foster greater adoption we have considered attributes of a SNP panel including: 1) backward compatibility to multiple historic genotyping platforms; 2) global relevance across populations; and 3) the ability to be platform agnostic. To achieve this, we used a 3-step approach for SNP selection. First, the candidate SNPs should be available in the public domain. Second, SNPs should be represented on at minimum 2 genotyping platforms: Agena (Sequenom), KASP, Illumina, Affymetrix, GBS/NGS. Lastly, final SNP selection was made using SNPs displaying high Minor Allele Frequency (MAF) and highest average call rate across data sets and platforms. A total of >200,000 animals consisting of more than 20 breeds and sample representation from 6 different geographic regions were evaluated. From this data a subset of 200 highly informative SNPs from a candidate pool of 857 SNPs were selected.

Key Words: sheep and related species, animal breeding, genotyping, parentage

OP79 High-throughput detection of single nucleotide polymorphisms with flexible content panels. S. Camiolo[1], J. Yeakley[1], E. Clark[2], B. Seligmann[1], and J. McComb*[1], [1]*BioSpyder Technologies Inc., Carlsbad, CA,* [2]*Zoetis Inc., Kalamazoo, MI.*

Detection of single nuclear polymorphisms (SNPs) is a powerful tool for genetic selection and maximization of the breeding potential of farm animals. It can also be used to estimate disease susceptibility or for pathogen detection. Most approaches to SNP calling, however, have significant limitations. Microarrays can measure many SNPs simultaneously but come with fixed content that cannot be customized or easily expanded without distorting original performance. Due to high costs of creating and qualifying each production lot, microarrays are usually available only for a subset of species, and often suffer from significant levels of lot-to-lot variability. qPCR detection is work intensive and severely limited in the number of samples and gene targets that can be evaluated simultaneously. Direct sequencing is expensive and produces data that is difficult to interpret correctly. TempO-SNP is a novel targeted assay capable of inexpensive high-throughput and high-plexity detection of SNPs from any species. It relies on direct hybridization of 2 adjacent barcoded oligomers to the target DNA, which are ligated into the reporter probe only if the correct SNP base is present. The content of such SNP panels is flexible as new probes can be added to the mixture easily and without affecting prior content. The assay does not require specialized instrumentation and the TempO-SeqR software pipeline makes SNP calling and report creation straightforward and painless. In partnership with Zoetis, we demonstrate TempO-SNP detection of hundreds of targets from hundreds of samples simultaneously, across multiple species. We also show that the assay can measure SNPs from crude tissue lysates or without need for DNA extraction, as well as from hair and blood. TempO-SNP shows excellent call and accuracy rates in a side-by-side comparison of data from the same samples produced by Zoetis' current microarray approaches. Additionally, TempO-SNP can be combined with existing commercial TempO-Seq technology to obtain RNA expression data from the same tissue lysates. Robust samples like dried blood spots on paper can also be used for both RNA and DNA readouts.

Key Words: SNP, genotyping, genetics, parentage, RNA

OP80 Genetic differentiation of *Camelus bactrianus* from Kazakhstan. K. Dossybayev*[1,2], D. Ualiyeva[1], M. Amandykova[1,2], T. Kapasuly[1,2], A. Mussayeva[1], Z. Orazymbetova[1], G. Shaltenbay[1,2], and B. Bekmanov[1,2], [1]*Laboratory of Genetics and Cytogenetics, Institute of Genetics and Physiology, Almaty, Kazakhstan,* [2]*Faculty of Biology and Biotechnology, Al-Farabi Kazakh National University, Almaty, Kazakhstan.*

The Bactrian camel represents an Old World camel that is well adapted to the cold and dry deserts of Middle and Central Asia. It is used to be the main source of food and logistics for the nomadic tribes of people since ancient times. Nowadays camels are bred worldwide for meat and dairy products. Recently, in Kazakhstan camel farming has been growing rapidly, particularly, in 2022 there registered around 272 thousand camels. The successful development of animal husbandry mainly depended on the genetic characteristics of farm breeds. Mitochondrial DNA is an optimal molecular marker which due to the high mutation rate allows for tracing the evolutionary history of matrilines as well as determining the speciation process. Nowadays, the genetic differentiation of local camels is poorly understood. Thus, to investigate the evolutionary relationships of domesticated Bactrian camels from Kazakhstan with extant populations spread worldwide, we determined the sequences of mitochondrial D-loop region from 13 camels, of the Almaty population. Totally, the analysis involved 50 samples including the sequences from GenBank. The targeted mitochondrial region consisted of a total length of 321 bp, which was analyzed by the Sanger method. The phylogenetic analysis recovered 2 main clusters representing the basal position of the monophyletic clade of Kazakhstani Bactrian camels with Arabian Dromedary camels, and a polyphyletic clade of *Camelus ferus* and *Camelus bactrianus* from Eastern Central Asia (Chi-

na, Mongolia). These results were supported by the haplotype network analysis as well with detection of 3 haplogroups. The obtained results suggest the possible past admixture and origin of a common ancestral form of the Central Asian population from the Arabian Peninsula. The current research may play a crucial role in the future investigations of the evolutionary history of the species. This research was funded by grant AP14870678 of the Ministry of Sciences and Higher Education of the Republic of Kazakhstan.

Key Words: *Camelus bactrianus*, Kazakhstan, mtDNA, phylogeny

OP81 Genetic diversity and population structure among Central European native sheep breeds using microsatellite markers. Z. Sztankoova, M. Milerski, M. Brzáková, J. Rychtárová, and J. Kyselova*, *Institute of Animal Science, Praha-Uhrineves, Czech Republic.*

Analysis of microsatellite loci is highly informative in reconstructing the historical processes underlying the evolution and differentiation of animal populations. This study used 13 polymorphic microsatellite markers recommended by FAO and ISAG to analyze the genetic diversity, genetic structure, variation, and phylogenetic relationship of 6 Central European sheep breeds (Czech Wallachian, CWA, n = 36, Sumava, S = 46, Slovak Wallachian, SWA, n = 59, Improved Wallachian, IPW, n = 59, Swiniarka, SWI, n = 35, and Uhruska sheep UHR, n = 19). The 172 alleles were observed in 254 animals. The number of observed alleles per locus varied from 7 to 17 per locus (average of 13.23). The mean number of effective alleles per locus was 5.77, with PIC ranging from 0.613–0.907 (equal to 0.77). Fst within subpopulations showed a low level of inbreeding. Nei's genetic distances between breeds were calculated, and results showed that the smallest distance was recorded between CWA and SWA (0.108). The largest was between the polish SWI and UHR sheep breeds (0.283). Principal component analysis showed that Czech and Slovak sheep breeds are closely related compared with Polish sheep breeds, specially SWI. Analysis of molecular variance showed a 6% variance among breeds and a 94% variance within populations. The ΔK value indicated that the most suitable group number was K = 4. These results showed genetic diversity, which is essential for future selection, animal breeding, and keeping the genetic diversity of native breeds. On the other hand, these results could help preserve genes in these breeds, thereby ensuring their preservation in the Czech and Slovak Republic and Poland. Therefore, future study is recommended to screen other middle European sheep breeds for comparison purposes.

Key Words: native sheep, gene resource, gene diversity, population structure, microsatellite

OP82 Genome-wide association study between copy number variations and economically important traits in American mink. P. Davoudi*[1], D. Ngoc Do[1], B. Rathgeber[1], S. Colombo[1], M. Sargolzaei[2,3], G. Plastow[4], Z. Wang[4], G. Hu[1], S. Valipour[1], and Y. Miar[1], [1]*Department of Animal Science and Aquaculture, Dalhousie University, Truro, NS, Canada,* [2]*Department of Pathobiology, University of Guelph, Guelph, ON, Canada,* [3]*Select Sires Inc., Plain City, OH,* [4]*Livestock Gentec, Department of Agricultural, Food and Nutritional Science, University of Alberta, Edmonton, AB, Canada.*

Copy number variations (CNVs) are structural variants consisting of duplications and deletions of DNA segments, which are known to play important roles in the genetics of complex traits in livestock species. However, CNV-based genome-wide association studies (GWAS) have not been reported in American mink. Therefore, the purpose of the current study was to investigate the association between CNVs and complex traits in American mink. A CNV-based GWAS were performed with the ParseCNV software program using deregressed estimated breeding values of 27 traits as pseudophenotypes, categorized into traits of growth and feed efficiency, reproduction, pelt quality, and Aleutian disease tests. The study identified a total of 10,137 CNVs (6,968 duplications and 3,169 deletions) using the Affymetrix Mink 70K single nucleotide polymorphism (SNP) array in 2,986 American mink. The association analyses identified 353 CNV regions (CNVRs) associated with at least one of the studied traits. These CNVRs overlapped with a total of 321 potential candidate genes, and among them several genes have been known to be related to the traits such as *ARID1B*, *APPL1*, *TOX*, *CXCL12*, and *PHYHIPL* (growth and feed efficiency traits); *DLGAP2*, *UNC5D*, *GRM1*, *SYCP2L*, *ARF1*, *RNASE9*, *RNASE10*, *WNT3*, *WNT3A*, and *WNT9B* (reproduction traits); *MYO10*, and *LIMS1* (pelt quality traits); and *IFNGR2*, *APEX1*, *UBE3A*, and *STX11* (Aleutian disease tests). Overall, the results of the study provide potential candidate genes that may regulate economically important traits and therefore may be used as genetic markers in mink genomic breeding programs.

Key Words: copy number variation (CNV), genome-wide association, complex trait, candidate gene, animal breeding

Domestic Animal Sequencing and Annotation

OP83 Invited Workshop Presentation: The human genome is finally complete, now what? S. Koren*, *National Human Genome Research Institute, National Institutes of Health, Bethesda, MD.*

Since its initial release in 2000, the human reference genome has covered only the euchromatic fraction of the genome, leaving important heterochromatic regions unfinished. Addressing the remaining 8% of the genome, the Telomere-to-Telomere (T2T) Consortium recently completed the 3.055 billion–base pair sequence of a human genome, T2T-CHM13. The completed regions include all centromeric satellite arrays, recent segmental duplications, and the short arms of all 5 acrocentric chromosomes, unlocking these complex regions of the genome to variational and functional studies. Building on this largely manual effort, we have since improved and automated this strategy in Verkko, an iterative, graph-based pipeline for assembling complete, diploid genomes. Verkko begins with a multiplex de Bruijn graph built from long, accurate reads and progressively simplifies this graph by integrating ultra-long reads and haplotype-specific markers. The result is a phased, diploid assembly of both haplotypes, with many chromosomes automatically assembled from telomere to telomere. Verkko has been used to generate multiple draft T2T genomes, including human as well as important agricultural species, such as tomato. The complete assembly of diploid genomes is a critical step toward the construction of comprehensive pangenome databases and chromosome-scale comparative genomics.

OP84 ISAG Bursary Award: An organism-wide ATAC-Seq peak catalogue for the bovine and its use to identify regulatory variants. C. Yuan*[1], L. Tang[1], T. Lopdell[2], C. Oget-Ebrad[1], G. Costa Monteiro Moreira[1], J. L. Gualdron[1], Z. Cheng[3], M. Salavati[3], D. C. Wathes[3], M. A. Crowe[4], W. Coppieters[1], C. Charlier[1], T. Druet[1], M. Georges[1], H. Takeda[1], [1]*GIGA Institute, University of Liège, Liège, Belgium,* [2]*Livestock Improvement Corporation, Hamilton, New Zealand,* [3]*Royal Veterinary College, Herts, UK,* [4]*School of Veterinary Medicine, University College Dublin, Dublin, Ireland.*

We herein report the generation of an organism-wide catalog of 976,813 *cis*-acting regulatory elements detected by ATAC-Seq. We regroup these regulatory elements in 15 tissue-specific and one tissue-shared components by nonnegative matrix factorization. Correlation between the genome-wide density of peaks and transcription start sites, between peak accessibility and expression of neighboring genes, and enrichment in transcription factor binding motifs supports their regulatory potential. Using a previously established catalog of