

Poster 2001

Title: “Nos2A Evaluation of *Nos2A* & *SLC9A3R1* as positional and biological candidate genes for litter size in pigs”

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Abstract

The aim of the present study was to identify the responsible genes of two epistatic QTL detected on SSC12 for litter size trait in a Meishan x Iberian F2 cross. Two functionally related genes, *Slc9a3r* and *Nos2a*, have been selected as functional and positional candidates to underlay these QTL affecting number of piglets born alive and total number of piglets born. *Slc9a3r* is a factor required for *Nos2* isoform activity and both, *Slc9a3r* and *Nos2a* variants have been related with reduction of the litter size in rodents. The complete cDNA sequences of both porcine genes have been characterized in four F2 animals. Nine SNPs were detected in *Slc9a3r*, four of them changing aminoacid composition; and 13 SNPs and three INDELs were detected in *Nos2a*, five out of them changing aminoacid composition. Physical (IMpRH) and linkage mapping were carried out and both genes were mapped on porcine chromosome 12, within epistatic QTL intervals. Significant associations for *Nos2a* and *Slc9a3r* polymorphisms on piglets born alive and total number of piglets born have been found with marker assisted association tests.

Poster 2002

Title: Genetic study of common variants at the ABCA1 and CETP genes in vervet monkeys

Authors: Gift Chauke and Jürgen Seier

Presenting Author: Gift Chauke, PO box 19070, MRC Primate Unit, Tygerberg, Cape Town, South Africa

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1. Jürgen Seier

Abstract:

In the last few years there has been growing evidence of the influence of genetic variation in the

determination of plasma lipid concentrations, especially for genes involved in lipid transport and metabolism. More recently, a pharmacogenetic approach has been applied to polymorphisms in these genes and demonstrated that they are also associated with response to lipid-lowering drugs. The objective of this study was to identify common variations in 2 important genes related to lipid transport and with potential to be genetic determinants of niacin (nicotinic acid) responsiveness in vervet monkeys (*Cercopithecus aethiops*).

20 monkeys were genotyped for polymorphisms in the genes encoding ATP-binding cassette transport A1 (ABCA1) and cholesteryl transfer ester protein (CETP). An average of 5 single nucleotide polymorphisms (R219K, V771M, V825I, I883M and I405V) was genotyped within these genes. Based on the genotype results, 4 monkeys were selected from the 20 and treated with niacin at an escalating dosage. Their mean lipid-lowering response following drug therapy was examined, compared to those with the same genotype in a placebo group. Data obtained from this study will contribute to potential benefits of using the vervet monkey as a model to evaluate the mechanism of niacin as a therapeutic approach that can exploit the benefits of HDL in coronary heart disease.

Poster 2003

Title: Identification of positional functional candidate genes for protein percentage and lactation persistency

Presenting Author: Mini Singh, University of Sydney, NSW 2570

Other authors (name only):

1. Peter C. Thomson
2. Herman W. Raadsma

Abstract:

Positional and functional evidence for importance of genes for lactation persistency and protein percentage will be identified by integrating positional candidate genes from QTL fine mapping and genes identified against location of functional candidates obtained from transcript profiling. Previously identified QTL regions associated with these traits in sheep on OAR3 and OAR20 have been fine mapped using 28 additional markers (12 on a 10 cM region in OAR3 and 16 on a 20 cM region in OAR20) with Linkage Analysis/ Linkage Disequilibrium analysis (LA/LD) over 4 sire backcross and double backcross families of approximately 700 daughters. From standard QTL analyses animals with contrasting genotypes ‘Q-’ and ‘q-’ for desired QTLs have been identified. RNA extracted from tissue samples from mammary glands

of these animals at three different times corresponding to pre-, peak-, and post lactation, have been used in functional (transcriptome) analysis using the Affymetrix Bovine Genechip array to obtain a list of differentially expressed genes. The use of combining QTL mapping and functional analyses using animals of defined QTL genotype will allow a list of positional functional candidate genes for further investigation.

Poster 2004

Title: New splice variants in the porcine *PPARGC1A* gene

Presenting Author: Tim Erkens, Heidestraat 19, 9820 Merelbeke, Belgium

Other authors:

1. Karel Bilek
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Abstract:

Peroxisome proliferator-activated receptor γ coactivator 1 α (*PPARGC1A*) is a versatile coactivator with many functions, but is primarily involved in fat and energy metabolism. Therefore, it is a very interesting candidate gene for meat quality, which is one of the most important criteria in pig selection today. *PPARGC1A* however, does not have the same function in every tissue and there is still much unknown about its regulation. Therefore a detailed transcription profile of *PPARGC1A* in 34 tissues and 3 embryonic developmental stages in the pig was constructed in order to detect any alternative splicing. This was done by extracting mRNA from each tissue, cDNA synthesis and screening for splice variants with exon-spanning primers. Two new splice variants in the pig were discovered in which exon 8 was partly or completely spliced out. Both splice variants potentially give rise to a much shorter protein of respectively 359 and 337 AA, of which the first 291 AA would be the same compared to the complete protein (796 AA). Considering the functional domains of *PPARGC1A*, it is very likely these variants have a large impact on the function of the protein and could provide an explanation for the regulation of the diverse functions of *PPARGC1A*.

Poster 2005

Title: *Acox1*: A candidate gene for fatty acid composition in pigs

Presenting Author: María Muñoz - Departamento de Mejora Genética Animal -INIA. 28040 Madrid, Spain

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Abstract:

Two QTL related with backfat fatty acid composition were previously detected on SSC12 in an Iberian x Landrace F2 cross. *Acox1* was selected as candidate gene to explain one of these QTL, because human-pig comparative mapping shows the human localization could match in the QTL interval and *Acox1* is the first enzyme of the fatty-acid β -oxidation in peroxisomes. Alternatively spliced transcript variants encode two different enzymes with the exons 3-II and 3-I respectively. The complete porcine cDNA sequences of both isoforms were sequenced in two Iberian, two Landrace and three pigs of a composite Chinese-European line, and seven SNP were detected. Two missense SNP were genotyped in the Iberian x Landrace intercross, and a new linkage map of the chromosome based on the segregation of 11 markers was obtained. The *Acox1* gene mapped at 17.5 cM between the *S0143* and *GH* markers. New linkage analysis identified three significant QTL: QTL1 affects the percentage of myristic, QTL2 the average chain length and the percentages of palmitic, linolenic and gadoleic and QTL3 the percentages of palmitoleic, stearic and vaccenic. No evidence was obtained of association of analyzed missense *Acox1* SNP with fatty acid composition.

Poster 2006

Title: Fine Mapping of QTL for Parasite Resistance in Sheep

Presenting Author: NATASHA A ELLIS, Centre for Advanced Technologies in Animal Genetics and Reproduction (ReproGen), The University of Sydney, PMB3, Camden, NSW, 2570, Australia

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Abstract:

Gastro-intestinal nematodes (GIN) are estimated to cost Australian sheep producers over \$AUD200 million annually through loss of production and death. Effective control of GIN is compromised by parasite

resistance to drenches, so selecting sheep that are naturally resistant to parasitic infection is a feasible alternative to chemical control. We have fine mapped two QTL regions for parasite resistance in a backcross Indonesian Thin Tail x Merino population that were twice challenged with *H. contortus*. Twelve and fourteen new microsatellites were added to the 15 and 14 markers previously mapped on OAR1 and 3 respectively. QTL for FEC ($P < 0.01$) and body weight change ($P < 0.05$) at first challenge were detected on OAR1 around positions 169cM and 312cM using linkage analysis. QTL for FEC ($P < 0.05$) and PCV changes ($P < 0.05$) during the first challenge were also detected on OAR3, around positions 180cM and 160cM. Caution should be used when interpreting these positions as confidence intervals were large; LA/LD analysis will be used to address this. A meta-analysis is underway with the results to be used in a collaborative effort to further fine map with a targeted SNP panel. Additional positional candidate gene studies are being conducted by integrating mapping studies with gene expression analyses.

Poster 2007

Title: Mapping of Quantitative Trait Loci for lactation persistency traits in ewes

Presenting Author: Elisabeth Jonas; 425, Werombi Road; Camden 2570, NSW; Australia

Other authors (name only):

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Abstract:

The persistency is a trait of great consideration and well investigated in dairy cattle; but only a few studies were performed targeting the productivity in dairy sheep. It was found in different studies that cows with greater persistency tend to incur less feed, health, and reproductive costs and that cows affected with mastitis tend to have less persistent lactations.

In this study samples of animals of an Awassi-Merino backcross population were used for a linkage analysis using regression and maximum likelihood methods.

The curves of the milk, protein, fat, lactose and energy yield were calculated using the Wood model. The three parameters derived from the model were further biological interpreted to calculate the yields and persistence of the traits. QTL for milk, protein, fat, lactose and energy yield, the persistency of the different traits and additional for the somatic cell score could be detected on different ovine chromosomes with the most interesting region on OAR3. The results of the genome-wide detection leading to the first study

of persistency for milk composition traits in dairy sheep. Future fine mapping and candidate gene approaches may lead to the detection of marker for the improvement of the productivity and health in dairy sheep.

Poster 2008

Title: Deletion in a conserved CCR5 motif is associated with approximately doubling proviral load of maedi-visna/ovine progressive pneumonia virus

Presenting Author: Stephen N. White, USDA-ARS-ADRU, PO Box 646630, 3003 ADBF, WSU, Pullman, WA 99164

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Abstract:

CCR5 is a chemokine receptor that serves as a coreceptor for human immunodeficiency virus (HIV) and may have a role in regulating maedi-visna/ovine progressive pneumonia virus (OPPV) infection. Both of these lentiviruses are macrophage-tropic, have similar genomic structures, and cause lifelong persistent host infection. A human CCR5 coding deletion (termed delta-32) results in strong resistance to HIV infection by preventing expression of active CCR5 protein on the cell surface, and polymorphisms in CCR5 regulatory regions have been implicated in delayed progression to acquired immune deficiency syndrome (AIDS). To evaluate the impact of CCR5 on OPPV, proviral loads of OPPV were measured in 383 naturally exposed Rambouillet, Polypay, and Columbia sheep with approximately equal numbers from each breed. The ovine CCR5 genomic sequence was determined, and polymorphisms were obtained from the open reading frame and surrounding regulatory sites. One haplotype was associated with approximately doubling proviral load among positive animals ($P < 0.01$), and the haplotype contains a 4 base deletion within a conserved motif in a regulatory region. While none of the polymorphisms was associated with differing odds of infection, the association with reduced proviral load suggests CCR5 may play a role in restricting the degree of OPPV infection.

Poster 2009

Title: Whole Genome Scan in Commercial Angus Cattle for QTL Influencing Carcass Value Traits: Carcass Weight, Fat Thickness, Marbling, and Ribeye Area.

Presenting Author: Matthew McClure.
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Abstract:

We have genotyped two large Angus mapping populations for 415 microsatellites and 9 SNP markers for all *Bos taurus* autosomes and the X chromosome and have tested for quantitative trait loci (QTL) influencing marbling, ribeye area, carcass weight, and fat thickness. The first population is comprised of 1,678 AI sires born after 1955; the second is comprised of 36 halfsib families containing 2,546 commercial steers produced by the Circle A Ranch and MFA Inc. Data were analyzed using QTLEXPRESS under a halfsib design and by LOKI using the full pedigree. At a chromosomewise $P \leq 0.01$ significant level, every autosome was found to harbour multiple carcass related QTL. We estimate there to be as many as 22 QTL for carcass weight, 24 QTL for fat thickness, 21 QTL for marbling and 31 QTL for ribeye area within the Angus genome. Comparing our data to published results suggests that the majority of these QTL segregate within all *Bos taurus* breeds of cattle, but that novel QTL could exist within specific breeds. These results support our population-based approach to QTL mapping within commercially relevant populations

Poster 2012

Title: Association of SNPs with carcass measurements and taste panel assessed meat quality traits

Presenting Author: ...

Jennifer Gill, Roslin Institute, Midlothian, UK, EH25 9PS

Other authors (name only):

1. Stephen C Bishop
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Abstract:

Various single nucleotide polymorphisms (SNPs) have been shown to be associated with meat and carcass quality traits in cattle. Examples include leptin, associated with fatness, and calpain and its inhibitor calpastatin, associated with meat tenderness. SNPs from these genes have been incorporated into

commercial genetic tests for predicted meat quality. This study aimed to validate the effects of these six SNPs and to test two further SNPs, in the growth hormone receptor (GHR) and DGAT1 genes, also suggested to have effects on meat quality. Traits included a mechanical measure of tenderness (tenderometer), 22 carcass quality traits and a unique set of seven taste-panel-assessed sensory traits. Data were collected from 483 commercial Aberdeen Angus-cross animals. Statistical analyses used REML. Fixed effects included farm, sex, genotype and the genotype-sex interaction; random effects included the interaction of farm, sire and slaughter date. Significant associations between SNP alleles and traits were found for four SNPs ($p < 0.05$ for all). An association was confirmed between one of the calpain SNPs and tenderness, assessed by both taste panel and tenderometer, in agreement with previous findings. Other significant associations included a leptin SNP with overall liking and fat level, DGAT1 with fat level and GHR with steak odour.

Poster 2013

Title: Genetics markers involved in productive traits in Romanian swine breeds

Presenting Author: Costache Marieta, Splaiul
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Abstract:

Research of the gene polymorphisms with major economical effects is currently the best-known application of pig genome mapping in breeding practice. In swine breeds, two major genes are well-known: the ryanodine receptor 1 gene (RYR1), associated with meat quality, and estrogen receptor gene (ESR) whose polymorphism is correlated with reproduction traits.

Our objective was to develop straightforward methods to examine the two loci in swine breeds from Romania. We used the PCR-RFLP technique to identify two point mutations: one for RYR1 gene (C→T) correlated with malignant hyperthermia and another for ESR gene (T→G) associated with reproduction traits. DNA amplification was carried out through PCR using normal primers and amplicons were digested with restriction endonuclease *HhaI* and *AvaI*. Restricted products were analyzed via electrophoresis in agarose gel stained with ethidium bromide. Our results were confirmed through sequencing.

The major focus of this study was to identify the normal or affected homozygous and carrier individuals in Romanian swine breeds and to implement a useful diagnosis methodology in order to assist veterinarians and breeders in disease control. The method presented above is reliable, fast, and can be successfully applied in the wide-scale screening of different pig populations.

Poster 2014

Title: Genetic characterisation of Charolais and Limousin cattle breeds using microsatellites markers in Slovakia.

Presenting Author: Jakabová Daniela, The Breeding Services of the Slovak Republic, state enterprise, Hlohovská 5, 95141 Nitra- Lužianky, Slovakia

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Abstract:

The polymorphism of 11 microsatellite markers (*BM1824*, *BM2113*, *ETH3*, *ETH10*, *ETH 225*, *INRA23*, *SPS115*, *TGLA53*, *TGLA122*, *TGLA126*, *TGLA222*) was investigated in the Slovak population of 199 Charolais and 66 of Limousin cattle. DNA was amplified in one multiplex PCR for amplifying these microsatellites (StockMark for Cattle, AB). The automatic analysis was performed using an ABI 310 sequencer and size of analyzed DNA fragments was determined in base pairs using computer package GeneScan v.3.7 (AB). Statistical parameters were calculated using Powermarker v.3.25 software. Total 99 alleles were identified in the Charolais breed and 82 in the Limousin breed. The number of allele per each locus ranged from 4 (*ETH10*) to 13 (*TGLA53*, *TGLA122*) in Charolais breed and in Limousin from 4 (*BM1824*) to 12 (*TGLA227*). The average value of H_e was 0.714 and H_o was 0.752 in Limousin. In Charolais, heterozygosities were lower: H_e was 0.667 and H_o was 0.644. Mean PIC values were similar: 0.6371 in Charolais and 0.6737 in Limousin cattle breed. The combined exclusion probability was ($PE > 0,999$) in both breeds. The values of PE confirmed the usefulness of this set of microsatellite markers in parentage testing of Charolais and Limousin cattle in Slovakia.

Poster 2015

Title: Increasing accuracy of estimated breeding value using DNA parentage test information in beef natural service populations

Presenting Author: Z. Wang, Department of Agricultural, Food and Nutritional Science, University of Alberta, Edmonton, AB, Canada, T6G 2P5.

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Abstract:

Genetic markers have been used for assigning progeny to their sires in beef cattle populations where natural service is the norm. The genetic evaluation for such population can only use maternal information since the paternal information is unknown. Sufficient single nucleotide polymorphisms (SNP) are now available for efficient and accurate assignment of parentage. The value of such parentage assignment is discussed in reviews, but no quantified research can be found. The IGENITY cattle marker panel contains 200 SNP was used to assign sires in nine herds of seed stock beef cattle population. On average the success rate of the parentage test in this population was between 82-87%. A total of 2082 progenies out of 2558 were assigned to 110 bulls across the herds. The objective of this study was to quantify the increasing in accuracy of estimated breeding value by adding the parentage test information. Two performance traits, birth weight (BWT) and weaning weight (WWT) were analyzed in this study. By adding the paternal information, the accuracy of EBV for these two traits was increased by approx. 10% and 17% on average for young calves and sires respectively.

Poster 2016

Title: Mapping quantitative trait loci for embryo survival and litter size in pigs

Presenting Author: Silvia Hernandez, The Roslin Institute and Royal (Dick) School of Veterinary Studies, University of Edinburgh, Roslin, Midlothian EH25 9PS, Scotland, UK.

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Abstract:

Reproductive performance is a critical component of sustainable animal production systems. Increasing the number of viable and productive offspring per reproductive female reduces financial and environmental costs and improves the sustainability of the system. The Chinese Meishan breed is one of the most prolific pig breeds known, displaying greater litter size than commercial Western breeds. Meishan pigs deliver this increased litter size through higher levels of prenatal survival for a given ovulation rate. To date only a fraction of the molecular genetic variation that underpins the superior reproductive performance of the Meishan has been identified. In an earlier study we mapped a QTL for the related traits of litter size and embryo survival to the distal end of pig chromosome 8. Additional genetic markers were developed in the QTL region and genotyped across the three-generation Large White – Meishan F₂ population in order to improve the resolution with which the litter size and embryo survival QTL were mapped. The confidence interval for the QTL has been aligned with the emerging pig genome sequence and homologous regions of the human and murine genomes in order to reveal positional candidate genes (including secreted phosphoprotein 1, SPP1) for litter size and embryo survival.

Poster 2017**Title: A putative quantitative trait locus on chromosome 20 associated with bovine pathogenic disease incidence**

Presenting Author: Eduardo Casas, USDA, ARS, U.S. Meat Animal Research Center, P.O. Box 166, Clay Center, NE 68933. USA

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1. Gary D. Snowder

Abstract:

The objective was to detect QTL associated with the incidence of multiple pathogenic diseases. Four F₁ sires were used to produce offspring: Brahman × Hereford (BH; n = 547), Piedmontese × Angus (PA; n = 209), Brahman × Angus (BA; n = 176), and Belgian Blue × MARC III (BM; n = 246). Records for bovine respiratory disease, infectious keratoconjunctivitis (pinkeye), and infectious pododermatitis (footrot) were available for all the offspring. The incidences of these three microbial pathogenic diseases were combined into a single binary trait to represent an overall pathogenic disease incidence. Offspring diagnosed and treated for one or more of the pathogenic diseases were coded as a 1 for affected and 0 for untreated. A putative QTL was detected in the family derived from the BH sire. This was supported by evidence of a similar QTL on the family derived

from the PA sire. The maximum *F*-statistic ($F = 13.52$; $P = 0.0003$) was located at centimorgan 18. The support interval of the QTL spanned from centimorgan 9 to centimorgan 28. Offspring inheriting the Hereford allele, in the BH-sired family, and the Angus allele, in the PA-sired family, were less susceptible to incidence of pathogenic diseases.

Poster 2018**Title: STRs vs. SNPs in horse parentage testing**

Presenting Author: Teruaki Tozaki*, Department of Molecular Genetics, Laboratory of Racing Chemistry, Utsunomiya, Tochigi 320-0851, Japan, ttozaki@lrc.or.jp

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Abstract:

At present, about 1.4 million SNPs were identified on the horse genome due to recent advances of the horse genome project. Interest in utilizing SNPs for parentage testing is rapidly increasing, but there is no enough information to evaluate the utility, such as their power of discrimination. In this study, two genotyping systems using each STRs and SNPs were used for the evaluation. At first, two multiplex assay groups were constructed to genotype 53 SNPs and genotypes of 95 thoroughbreds were determined using Sequenom's MassARRAY system. Total PE of the 53 SNPs was 0.99996, and MAF was 0.36 on average. The SNP system was applied for the questions of the ISAG 2006 horse comparison test, and 5 of the 53 SNPs excluded for a pseudo Sire-Foal combination. Next, 30 STRs used in our routine and additional parentage testing were genotyped using the 95 thoroughbreds. Total PEs were 0.99997 and 0.99991, respectively. Allele frequency was 0.721 on average. The STR systems were also applied for the same questions, and 5 and 6 STRs excluded for the pseudo combination, respectively. Those results indicate that about 50 SNPs are required to reach equivalent power of discrimination as well as the routine STR systems.

Poster 2019

Title: Nutrient-induced modulation of genome profile and genetic network

Presenting Author: Congjun Li, Bovine Functional Genomics Laboratory, USDA-ARS, 10300 Baltimore Ave, BARC EAST, Beltsville, MD 20705 USA

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Abstract:

Short-chain fatty acids participate in metabolism both as nutrients and as regulators of histone deacetylation. The major biochemical change that occurs in cells treated with butyrate is the global hyperacetylation of histones. Utilizing gene expression profiling, our studies indicated that butyrate induces many significant changes in the expression of genes associated with regulatory pathways that are critical to cell growth, immune response and signal transduction. We identified 450 genes significantly regulated by sodium butyrate at a very stringent false discovery rate (FDR) = 0%. When relaxing the stringency to FDR = 10%, there were 3662 genes significantly regulated ($3662/45383 = 8\%$). The functional category and pathway analyses of the microarray data revealed that four canonical pathways (Cell cycles checkpoint; pyrimidine metabolism; G1/S checkpoint regulation and purine metabolism) were significantly perturbed. The biologically relevant networks and pathways of these genes were also identified. *IGF2*, *TGFBI*, *TP53*, *E2F4*, and *CDC2* were established as being centred in these genomic networks. Butyrate induced biological effects in bovine cells provide an example of epigenetic regulation of genome and a basis for understanding the full range of the biological roles and the molecular mechanisms that butyrate may play in animal cell growth, proliferation, and energy metabolisms.

Poster 2020

Title: Relationship of the Bovine Growth Hormone Gene to Carcass Traits in Japanese Black Cattle

Presenting Author: Ken Tatsuda (K. Tatsuda), 1533 Befucyo Kasai-City Hyogo 679-0198, Japan, Tel. : +81-790-47-2430, Fax : +81-790-47-0549, E-mail : Ken_Tatsuda@pref.hyogo.lg.jp

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Abstract:

The bovine growth hormone gene (bGH) possesses three haplotypes, A, B and C, that differ by amino acid mutations at positions 127 and 172 in the fifth exon : (leucine₁₂₇, threonine₁₇₂), (valine₁₂₇, threonine₁₇₂) and (valine₁₂₇, methionine₁₇₂), respectively. The correlation between meat quality or carcass weight and these haplotypes was investigated in Japanese black cattle. Altogether, 940 bGH haplotypes were compared with respect to six carcass traits; carcass weight, longissimus muscle area, rib thickness, subcutaneous fat thickness, beef marbling score and beef color. The frequency of the B haplotype was higher (0.421) than that of A (0.269) and C (0.311). High carcass weight and low beef marbling were associated with haplotype A ($p < 0.05$ and $p < 0.01$, respectively), whereas beef marbling was increased by haplotype C ($p < 0.05$). Estimated regression coefficient of the A haplotype substitution effect for carcass weight and beef marbling score were 5.55 (13.1% of the phenotypic S.D.) and -0.31 (17.0%), respectively. That of the C haplotype for beef marbling score was 0.20 (11.0%). The other traits showed no relationship to the haplotypes examined. The results of this investigation suggest that information pertaining to bGH polymorphisms in Japanese black cattle could be used to improve the selection of meat traits.

Poster 2021

Title: Genome wide association study of feed efficiency in beef cattle.

Presenting Author: Laura Sherman, Department of Agricultural, Food and Nutritional Science, University of Alberta, Edmonton, AB, T6G 2P5, Canada

Other authors (name only):

1. Donald Nkrumah
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Abstract:

Feed is the highest variable cost in beef production making feed efficiency an important trait to study. Feed efficiency is measured as residual feed intake (RFI), which is the difference between actual feed intake of the animal and expected feed intake based on weight and growth rate. 2,633 SNPs covering the bovine genome were analyzed in 464 steers for associations with RFI. 150 SNPs were identified with $P < 0.05$. To create a panel of SNPs that were maximally informative for RFI from these 150 SNPs, two methods were tested. First, a sequential molecular breeding value (MBV) was created for each animal by sequential addition of the SNP effects and only SNPs which improved the model were included. This panel

contained 79 SNPs. Second, the SNPs were combined in a single multivariate model and a backward elimination was used until all SNPs left in the model were significant with $P < 0.05$, which left 32 SNPs. Regression of an MBV of the 32 SNPs on RFI produced $r^2 = 0.497$ and regression of the 79 SNP MBV produced $r^2 = 0.497$. These SNP panels provide progress towards identification of markers for use in marker-assisted selection of RFI in beef cattle.

Poster 2022

Title: Genetic effects mapped in a Charolais X Holstein cross cattle population.

BEATRIZ GUTIÉRREZ-GIL¹, JOHN L WILLIAMS², PAMELA WIENER³

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Abstract:

An experimental cattle population to identify QTL influencing cattle production traits was established at the Roslin Institute, starting in 1995 by crossing Charolais sires and Holstein dams. Phenotypic measurements for a wide range of traits were made on the 500 second-generation individuals of the herd (F2, Charolais Backcross and Holstein Backcross individuals). We present here a summary of the results obtained for the traits related to meat production, temperament and coat colour dilution. A QTL affecting proportion of bone in the carcass on chromosome 6 was the most significant QTL identified for production traits. Many other genome-wide significant QTL were detected for yield and carcass composition traits. Several QTL were also found with effects on technological properties of the meat (e.g. moisture, pH) and consumer satisfaction (taste panel traits and fatty acid composition). For the temperament traits studied, which assessed the fearfulness of the animals to approaching humans and to social separation, a single genome-wide significant QTL was found on chromosome 29. The coat colour records were used to map the *Charolais dilution* locus, and to assess the relationship of a mutation in the *SILVER* gene with the coat colour dilution characteristic of the Charolais breed

Poster 2023

Title: Searching QTL affecting milk traits on chromosome 22 in Spanish Churra sheep.

Presenting Author: ELSA GARCÍA-GÁMEZ,
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3. LUIS FERNANDO DE LA FUENTE
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Abstract:

After a preliminary genome scan carried out in Churra sheep to detect QTL influencing milk production traits, additional families are now being analyzed to confirm some of the identified effects. We present here the analysis performed on chromosome 22 for 15 additional families, which included 798 Churra ewes. Phenotypic measurements for milk yield, protein percentage, fat percentage and somatic cell scores (SCS) were used to calculate yield deviations used in the analysis. Eight microsatellite markers evenly distributed across the chromosome were genotyped for all the population and used to construct a linkage map. A multimarker regression method was implemented through the *QTL Express* software. Chromosome-wise critical values were calculated through 10,000 phenotype permutations. The average information content across the linkage map was 0.78. The across-family association analysis revealed a 5% chromosome-wise significant QTL for the SCS (p-value = 0.026), between markers INRA81 and TGLA429. None of the production related traits showed significant linkage association on this chromosome. The SCS QTL was found to be segregating in three of the 15 analyzed families. The joint analysis of these data with those generated through the genome scan experiment will be needed to confirm the QTL effect here reported.

Poster 2024

Title: Development of a Panel for use in Red Deer Maternity Assessment.

Presenting Author: María Marcela Martínez.
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2. Silvia Di Micco.
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4. María Marcela Cerruti.

Abstract:

In farms, the red deer mating programs consists of one male which services 30-40 hinds; working usually with 12 groups of enclosed hinds.

In our country, red deer maternity assessment is still based on visual observation during the post-parturient phase. The behaviour of hinds and calves, mainly mother care and nurse, is used to assess maternity relationships. Adoption and allosuckling can affect, however, the accuracy of this procedure, which also is very hard and expensive.

Like in other species, DNA typing by microsatellites could be the alternative to reduce the mismothering rate. Due to the absence of a set of markers for typing service in red deer, our lab developed a panel of 11 markers useful for identification and parentage testing. All the markers are of public domain, derived from cattle and deer genomes. A total of 130 alleles were found on 90 samples tested. Assuming one known parent, the individual exclusion power ranged between 0.17 and 0.73, while the cumulative exclusion probability reached 0.999947 for the whole panel. The panel was validated on a trial assay, involving 34 hinds, 32 calves and 7 males. The results of DNA parentage testing were confirmed by previous visual observation assignment.

Poster 2025

Title Polymorphisms in Positional Candidate Genes on BTA14 and BTA26 affect Carcass Quality in Beef Cattle

Presenting Author: Elisa Marques. Department of Agricultural, Food and Nutritional Science, University of Alberta, Edmonton, Alberta, Canada, T6G 2P5.

Other authors (name only):

1. Donald Nkrumah
2. Laura Sherman
3. Stephen Moore

Abstract:

Previous analysis on Bovine Chromosome 14 (BTA14) and 26 (BTA26) have indicated a number of quantitative trait loci (QTL) affecting meat quality traits. Association studies of 2, 4 dienoyl CoA reductase 1 (*DECRI*) in pigs and core binding factor alpha domain 2 (*CBFA2T1*) in humans have shown associations between polymorphisms in those genes with lipid metabolism. Sequencing analysis in cattle identified nine polymorphisms in *DECRI*; including four exonic, two of which produced changes, while four intronic SNPs were identified in *CBFA2T1*. Multiple sequence alignment of *DECRI* among cattle, human and mice showed that four mutations lie in conserved regions across these species. Single locus analysis produced associations ($P < 0.05$) with ultrasound marbling score (*CBFA2T1*) and ultrasound

backfat (*DECRI*). Recent studies have linked fibroblast growth factor 8 (*FGF8*) to several QTL affecting obesity in mice which indicated its potential for regulating adiposity in other species. In *FGF8*, four polymorphisms were identified, two intronic and two exonic. Single locus analysis resulted in significant associations ($P < 0.01$) with carcass backfat and lean meat yield. This study hopes to contribute to a growing list of genes with specific functions affecting the final meat quality in beef cattle.

Poster 2026

Title: Effect of SCD, SREBP and FASN genotypes on fatty acid composition in adipose tissue of Japanese Black cattle.

Presenting Author: Shinji SASAZAKI, Laboratory of Animal Breeding and Genetics, Graduate School of Agricultural Science, Kobe University, Kobe, Japan 657-8501.

Other authors (name only):

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2. Hideki OHSAKI
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4. Fumio MUKAI
5. Hideyuki MANNEN

Abstract:

Fatty acid composition of beef adipose tissue is one of important traits because high proportion of monounsaturated fatty acid is related with favorable beef flavor and tenderness. In this study, we investigated the effects of genetic factors such as stearoyl-CoA desaturase (SCD), sterol regulatory element binding protein (SREBP) and fatty acid synthase (FASN) on beef carcass traits including fatty acid composition using two cattle populations ($N = 417$ and 233 , respectively). Sire effect was significantly related to almost all traits except BMS, suggesting that the trait examined in this study is highly controlled by genetic factors. The effect of SCD genotypes on fatty acid composition was detected remarkably in both cattle groups, especially on stearic (C18:0), oleic (C18:1) acids and monounsaturated fatty acid (MUFA) content. FASN genotypes were significantly associated with almost all fatty acids (C14-18) except stearic (C18:0) and linoleic acid (C18:2). However, effect of SREBP genotypes was not identified in this study. Our results also suggested there were other genetic factors relevant to fatty acid metabolism. In conclusion, these results would contribute to bring an insight into molecular mechanism of fatty acid metabolism in cattle.

Poster 2027

Title: The ubiquitin ligase gene (WWP1) is responsible for the chicken muscular dystrophy.

Presenting Author: Hirokazu MATSUMOTO, Laboratory of Animal Breeding and Genetics, Graduate School of Agricultural Science, Kobe University, Kobe, Japan 657-8501.

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12. Hideyuki MANNEN

Abstract:

Muscular dystrophy is defined as a group of inherited diseases producing progressive weakness and degeneration of skeletal muscles. It is well known that abnormalities of muscle proteins to compose the linkage between sarcolemma and basal lamina lead to muscular dystrophies, but genes responsible for several muscular dystrophies and related diseases have not yet been identified. The gene responsible for chicken muscular dystrophy with abnormal muscle (*AM*) remains unclear, either. Our previous study revealed seven functional genes as *AM* candidate genes, none of which were determined to be responsible for other muscular dystrophies. In current study, sequence and expression comparison between normal and dystrophic chickens was conducted to detect a mutation responsible for the disease. We detected three synonymous mutations and one missense mutation in *AM* candidate genes, while no remarkable alteration of expression patterns was observed. The missense mutation was observed in *WWP1* and detected only in dystrophic chickens in several tetrapod species. These results strongly suggested *WWP1* would be responsible for chicken muscular dystrophy. The information must be useful for determining the corresponding human dystrophy and for providing new insights for understanding muscular dystrophies.

Poster 2028

Title: Development of SNPs markers for individual identification and parentage test in Japanese Black cattle population.

Presenting Author: Kazuhiro HARA, Laboratory of Animal Breeding and Genetics, Graduate School of Agricultural Science, Kobe University, Kobe, Japan 657-8501.

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Abstract:

Individual identification and parentage control are essential for consumer protection and efficient management of animal population. Since single nucleotide polymorphisms (SNPs) are abundant in genome, genetically stable and amenable to high-throughput automated analysis, these are expected to be efficient DNA markers for the diagnostic. This study describes the development of SNP markers in order to apply the identification and parentage test in Japanese Black cattle population. Amplified fragment length polymorphism method was employed to detect informative candidate markers and yielded 44 SNPs markers from the 220 primer combinations. Unlinked 30 SNPs out of them were finally selected as diagnostic markers. The allele frequencies for each marker were estimated by using PCR-RFLP in Japanese Black population. Based on the frequency data, estimated identity power of these markers was $1.18E-12$. Parentage exclusion probability was 0.97243 with only one sampled parent. Sire exclusion was 0.99747 when the dam's genotype was known. This panel of SNP markers is theoretically sufficient for individual identification of any individual in Japanese Black cattle. These markers could be useful for identification and parentage test, and would contribute to management of beef industry in Japan.

Poster 2029

Title: Polymorphisms of fatty acid related genes and association between genotypes and fatty acid composition in Japanese Black cattle.

Presenting Author: Shogo HOASHI, Laboratory of Animal Breeding and Genetics, Graduate School of Agricultural Science, Kobe University, Kobe, Japan 657-8501.

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Abstract:

The fatty acid composition of adipose tissue in cattle has become more important in the beef industry. Fat tissue containing abundant monounsaturated fatty acid (MUFA) reflects lower fat melting points, leading to favorable beef flavor and decreasing the blood concentration of LDL-cholesterol. In this study, we searched polymorphisms in full length CDS of the six genes, *FABP4*, *LXR α* , *CYB5*, *ACSL1*, *ACSL4* and *DGAT2*, associated with fatty acid and lipid metabolism. Sequence comparison among eight animals, including five Japanese Black and three Holstein cattle, revealed 12 single nucleotide polymorphisms. Four of them, I74V, V110M in *FABP4* and G51E, V133I in *LXR α* , were predicted to cause amino acid substitutions. We investigated associations between these genotypes and fatty acid compositions of carcass fat in Japanese Black cattle. The genotype I74V in *FABP4* was significantly associated with palmitoleic acids (C16:1, $P < 0.01$) content of intramuscular fat, while remarkable effect was not detected in the other genotypes. In conclusion, I74V in *FABP4* gene would contribute to improve fatty acid composition in beef cattle.

Poster 2030**Title: Assessment of genetic diversity in Percheron horses registered in Canada using genealogical records and multiloci microsatellite data.**

Presenting Author: Tara McParland, Maxxam Analytics Inc., 2 - 335 Laird Road, Guelph, ON, Canada, N1H 6J3

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1. Yves Plante

Abstract:

In North America, the Percheron played a significant role in the development of agriculture and forestry. The Percheron was also a very popular stage-coach horse. In Canada, between 1908 and 1950, the Percheron registry counted twice as many horses (41,130) than the period between 1951 and 2000 (19,997). In 1973, only 23 stallions and 74 mares were registered. Over the last ten years, the Percheron has regained some popularity with average yearly registrations of 318 stallions and 655 mares. Within a framework of genetic resource conservation, we make use of 25,851 available genealogical records and 1,104 multiloci microsatellite genotypes (equine parentage set) to assess temporal changes in genetic diversity in the Canadian registered Percheron population. Average inbreeding (F) was 1.99% (2.52% within inbreeds). The increase in inbreeding ranged from 0.21% (maximum generation) to 0.43% (equivalent generation). Observed and expected heterozygosities remained high since 1985 (0.71 ± 0.022 and

0.71 ± 0.011) and comparable to other major and popular horse breeds. Individual microsatellite allelic richness and the polymorphism information content stayed constant through time. In conclusion, the Canadian registered Percheron horse population exhibits a relatively high level of genetic diversity even though it has gone through a reduction in population size between 1950 and 1975.

Poster 2031**Title: Understanding *Haemophilus Parasuis* Infection in Porcine Spleen Through Transcriptomics Approach**

Chang-Chun Li, * Hong-Bo Chen, * Ming-Di Fang, * Meng-Jin Zhu, Xin-Yun Li, Rui Zhou, † Kui Li, † Shu-Hong Zhao *

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Abstract:

Haemophilus parasuis (HPS) is a prominent swine pathogen that causes Glässer's disease characterized by fibrinous polyserositis, meningitis and arthritis. The molecular mechanisms underlying disease pathogenesis related to HPS infection is poorly understood, particularly the host counteraction to HPS invasion by the immune system. In this study, we firstly investigated the global expression changes in spleen following HPS infection using the Affymetrix Porcine Genechip™. Differential expression of selected genes was confirmed by QRT-PCR analysis. Differentially expressed genes involved in subsystems/modules including inflammasomes, acute-phase proteins and complement, cell differentiation, adhesion molecules, transcription factors and many others which may play central roles in several cascades. Also, down-regulations of both MHC class I and MHC class II genes are likely the main mechanisms for HPS evasion. We further detected the expression change of differentially expressed genes in LPS or Poly (I:C) treated porcine PK-15 cells. Interestingly, mRNA levels of S100A8, S100A9, and S100A12 increased in a sustained manner within 48h after administered LPS and Poly (I:C) respectively, suggesting they may play important roles in porcine systematic inflammations. *In silico* mapping of differentially expressed genes to porcine immune trait QTL regions showed that acute-phase protein HP and transcription factor CEBPD may be important candidate genes which merit further investigation.

Altogether, our findings indicate previously unrecognized gene transcription changes in case of HPS infection in vivo and should provide new clues for identification of candidate genes related to HPS resistance.

Poster 2032

Title: FecX^R, a deletion in the BMP15 locus associated to increased prolificacy in the Rasa Aragonesa sheep breed.

Presenting Author: Luis V. Monteagudo Ibañez, Department of Anatomy, Embriology and Genetics, Faculty of Veterinary Sciences of Zaragoza (Spain), C/Miguel Servet 177. 50013-ZARAGOZA (Spain)

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Abstract:

Since 1990, the National Association of Rasa Aragonesa Sheep Breeders (ANGRA) maintains a genetic improvement program for prolificacy. Data on 1.5 million were obtained during the field production control. We selected a small group of ewes showing high prolificacy performances in the resulting database. The exons for GDF9 (Growth Differentiation Factor 9) and BMP15 (Bone Morphogenetic Protein 15) were PCR amplified and sequenced. We identified a 17 bp deletion in the second exon of the BMP15 pro-protein. This deletion modifies the open reading frame in the sequence, so that only the first 45 amino acids of the wild variant are conserved. Then, premature stop codons appear at the positions 100 and 101. As a consequence the region of the pro-protein conserved in the mature protein is not synthesized (it normally starts in the position 120). Ewes heterozygous for the mutation (named FecX^R) show increased prolificacy, due to an increase in the number of duplets and triplets. In fact, the highest prolificacy in the ANGRA records was obtained by one of these ewes (3.67 lambs as a mean in three births). At present we are extending our search to flocks all over Aragón to detect further carriers of the deletion.

Poster 2033

Title: Biosafety assessment of markers genes in transgenic cloned cattle

Presenting Author: Yan Liu, State Key Laboratory for Agrobiotechnology, China Agricultural University, Yuanmingyuan West Road 2, Beijing, China

Other authors (name only):

1. Ning Li

Abstract:

Both enhanced green fluorescence protein (EGFP) and neomycin phosphotransferase type II (NPTII) are widely used in transgenic studies, but their side effects have not been extensively investigated. In this study, we evaluated the expression profiles of the two marker genes and the relationship between their expression and organ abnormality (dead cattle)/health profile (living cattle). In dead cattle, EGFP and NPTII protein expression level ranged from 0.3 to 5ug/g in heart, liver and lungs, and the expression profiles exhibited differential or mosaic pattern between the organs, the pathologic symptoms of which were identified, but were similar to those of age-matched cloned cattle ($p>0.05$). All data indicated that the expression of EGFP and NPTII is not associated with organ abnormality in transgenic cloned cattle. With respect to living cattle, all parameters derive from serum samples in which EGFP and NPTII expression will be measured by RIA and ELISA. Meanwhile, hormones and clinical parameters, such as complete blood counts and biochemical analyses will be checked every two months during half a year. All the parameters from transgenic cloned cattle will be compared with the ones from age-matched cloned cattle and age-matched normal cattle. The study is in progress.

Poster 2034

Title: Identification of differentially expressed genes in rumen tissues of dairy cattle fed a low or high grain diet

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Abstract:

A fundamental understanding of molecular mechanisms of ruminal epithelial cell adaptation to highly fermentable diets is essential to prevent ruminal acidosis (RA). In this study, we aimed to gain a broad

overview of the involved molecular mechanisms by detecting differentially expressed genes (DEG) in rumen tissues from lactating dairy cows fed a low (8%) or a high grain diet (64%) using microarray analysis (a bovine 24K panel). A total of 7035 elements were detected as DEG with >1.5-fold expression change ($P < 0.05$): 2993 and 4042 were up- and down-regulated in the cows fed a high grain diet, respectively. Molecular function of the DEG analyzed with gene ontology and KEGG databases indicated that 49 (22 up/27 down), 26 (8 up/18 down) and 19 DEGs (9 up/10 down) are involved in MAPK, calcium, and insulin pathways, respectively. The down-regulated gene encoding cAMP-dependent protein kinase subunit beta (*PRKACB*) was common among the three pathways. This suggests that intracellular signals induced by cAMP may be negatively affected in cows fed highly fermentable diets. Functional analyses of DEG to elucidate the interactions between diet fermentability and metabolic adaptation by animals, leads to a better understanding of the role of absorptive metabolisms in mitigating RA.

Poster 2035

Title: Molecular analysis and SNP development of three candidate genes (*IL6*, *IL10*, *TF*) for disease resistance in pigs

Presenting Author: Emilia Danilowicz, Institute of Genetics, University of Berne, Bremgartenstrasse 109a, 3001 Berne, Switzerland

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Abstract:

Different cytokines such as interleukin 6 (*IL6*) and *IL10* are secreted in response to specific microbial molecules referred to as pathogen associated molecular patterns (PAMPs) and play a central role in the immunological response. Transferrin-mediated provision of iron is essential for bacterial growth and iron-depletion of transferrin is a first line defense against bacterial infections.

We obtained the genomic structure and complete DNA sequence of the porcine *IL6*, *IL10* and *TF* genes and identified polymorphisms on a panel of ten different pig breeds. Comparative intra- and interbreed sequence analysis revealed 8 polymorphisms in the porcine *IL6*, 21 in the *IL10*, and 57 in the *TF* gene, which include single nucleotide polymorphisms (SNPs) and insertion deletion polymorphisms (indels).

In the *IL6* gene one polymorphism was located in an exon (c.92G>T) and predicted to cause an amino acid exchange (p.R31L). In the *IL10* gene, two polymorphisms were located in the coding sequence, and one (c.371A>T) was predicted to cause an amino acid exchange (p.Q124L). In the *TF* gene 7 of the 57 polymorphisms were located in the coding sequence. Two SNPs (c.1417A>G, c.1810A>C) were predicted to cause amino acid exchanges (p.K473E, p.N604H). All polymorphisms were submitted to the NCBI dbSNP.

Poster 2036

Title: Assignment of the swine MHC alleles and biological traits in selective breeding Duroc pigs

Presenting Author: ASAKO ANDO, Department of Molecular Life Science, Division of Basic Medical Science & Molecular Medicine, Tokai University School of Medicine, 143 Shimokasuya, Isehara, Kanagawa 259-1193, Japan

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Abstract:

Pigs with defined swine major histocompatibility complex (MHC: SLA) haplotypes and characterized immunological traits are useful for studies of transplantation and immune responses. To develop SLA homozygous pigs with novel SLA haplotypes, a pair of pigs was chosen from a commercial Duroc pig line, and substantial breeding within progenies was carried out for eight generations. The theoretical inbreeding coefficient at the eighth generation was 78.5%. In the selective breeding Duroc (SBD) pigs, only two SLA haplotypes including novel two class I haplotypes, Hp-27.30 and Hp-60.13 were assigned by SLA-DNA typing methods. The haplotypes were also identified by genetic polymorphisms of 36 microsatellite markers within the SLA region in the SBD pigs. Despite inbreeding for eight generations, litter sizes of the SBD pigs in the all generations were comparable to those of non-SBD pigs. Weaning weights from the fifth to eighth generation produced progenies significantly lighter ($p < 0.01$) than those in the non-SBD pigs. Concerning the lymphocyte subsets, the percentage of CD4+ T-lymphocytes in

peripheral blood of the SBD pigs was significantly higher than that in the non-SBD pigs. Our newly established SBD pig lines with novel SLA haplotypes will be useful in further studies on immune responses against various foreign antigens.

Poster 2037

Title: Analyses of serum antibody titers against swine erysipelas vaccines and hepatitis E virus infection in SLA-defined selective breeding Duroc pigs

Presenting Author: HITOSHI KITAGAWA, Laboratory of Veterinary Internal Medicine, Faculty of Applied Biological Sciences, Gifu University, 1-1 Yanagido, Gifu 501-1193, Japan

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Abstract:

To clarify the relationship between swine leukocyte antigen (SLA) haplotypes assigned by DNA typing techniques and antibody productivity, we determined antibody titers against swine erysipelas live and inactivated vaccines and natural infection of hepatitis E virus (HEV) in SLA-defined selective breeding Duroc pigs with 2 SLA haplotypes; Hp-27.30 and Hp-60.13. Serum antibody titers against a swine erysipelas live vaccine determined by a viable cell agglutination test were lower in order in Hp-27.30 homozygotes, Hp-27.30/60.13 heterozygotes and Hp-60.13 homozygotes. Mean titer in the homozygous pigs with Hp-27.30 was significantly ($P < 0.05$) lower than that in non-selective breeding pigs. Titers against inactivated vaccine for swine erysipelas were determined by ELISA. Mean titer in Hp-27.30 homozygous pigs was lowest, and significantly ($P < 0.05$) lower than in Hp-27.30/60.13 heterozygous pigs. Antibody titers against HEV were determined by ELISA in 89 SLA-defined pigs at the ages of 20 to 50 weeks. Homozygous pigs with Hp-27.30 haplotype had the lowest mean titer followed by the heterozygous pigs and homozygous pigs with Hp-60.13 haplotype. Thus, the homozygous pigs with Hp-27.30 tended to exhibit rather low antibody titers, suggesting the associations among SLA-class I Hp-27.0 and/or class II Hp-0.30 haplotypes and low responsiveness against immune stimulation by these foreign antigens.

Poster 2038

Title: Syndactyly in a German Holstein cow – a case report

Presenting Author: Claudia Floren, Institute of Veterinary Medicine, University of Goettingen, Burckhardtweg 2, 37077 Goettingen, Germany

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Abstract:

Syndactyly in Holstein cattle, also called Mulefoot disease (MFD), is an autosomal recessive abnormality characterized by the fusion of the functional digits. An identity-by-descent linkage study mapped the disorder to BTA 15. The region is homologous to a segment of murine chromosome MMU 2 containing the low density lipoprotein receptor-related protein 4 gene (*LRP4*). Studies in mouse showed that *LRP4* plays an essential role in the process of digit differentiation in mammalian species. Recently, six mutations associated with syndactyly have been detected in cattle. All newly described mutations affect different conserved protein domains but do not explain all analyzed cases of syndactyly. Here, an affected Holstein cow, with two syndactylous hind foot claws has been investigated for these six mutations. In addition, exons 2 to 38 of *LRP4* and the respective splice/donor sites were comparatively sequenced using the affected animal, six unaffected randomly chosen animals and one heterozygous carrier for MFD. Surprisingly, the affected cow did not reveal any of the described SNPs associated with the phenotype. This confirms the supposition of additional mutations causative for syndactyly. Further experiments will focus on the 5'UTR and exon 1 to clarify whether these regions might be involved in this case of syndactyly.

Poster 2039

Title: SNP within the porcine *NAMPT/PBEF1/visfatin* gene is associated with fat and carcass traits

Stanislav Cepica¹, Heinz Bartenschlager², Cristina Ovilo³, Almudena Fernandez³, Jana Zrustova⁴, Martin Masopust¹, Ales Knoll⁴, Hermann Geldermann²

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Abstract:

NAMPT (visfatin) is an essential enzyme in the NAD biosynthetic pathway. Findings obtained in rodents and man suggests a role of visfatin in glucose metabolism and pathogenesis of type 2 diabetes. On the basis of porcine RNA sequence of *NAMPT* (NM_001031793) we cloned a 1417 bp fragment encompassing partial exons 9 and 10 and intron 9. Using IMPRH panel the gene mapped close to *SW944* (0.52 R, LOD=7.53) located at position 83.3 cM on SSC9 map. In Wild Boar x Meishan F₂ family (N=333) *TT* genotype of AM999341:g.669T>C is associated (GLM) with lower fat cell volume, smaller fat area on *m.l.d* at 13th/14th rib, lower loin and neck meat weight, higher weight of ham relative to half carcass weight, lower growth rate and higher drip losses ($P<0.01$) as compared to *CT* genotype. In commercial Landrace x Chinese-European synthetic (N=447) *TT* is associated (animal model) with higher backfat thickness at 160d, lower shoulder weight, IMF % ($P<0.05$), Minolta *a** and Minolta *b** values ($P<0.01$) while *CT* is associated with lower C16:0 and higher C18:2 content in backfat. *TT* is associated (GLM) with higher and lower backfat thickness in Large White (N=216, $P<0.05$) and Black Pied Prestice (N=97, $P<0.05$), respectively.

Supported by the Czech Science Foundation (523/07/0353).

Poster 2040

Title: Transcriptomic analysis of porcine skeletal muscles exposed to a single bout of endurance exercise

Presenting Author: Jeanette Hedegaard Hansen, department of Genetics and Biotechnology, Faculty of Agricultural Sciences, University of Aarhus, Tjele, Denmark

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3. Bo Thomsen

Abstract:

The phenotype of skeletal muscle cells is highly adaptive to different types of stimulus such as stress or exercise. The aim of the present study is to investigate the impact of a single bout of endurance exercise on global transcriptional profiles in porcine skeletal muscles measured immediately after, 1 hour after and 3 hours after exercise. The transcriptional profiles are

obtained using porcine oligonucleotide microarrays consisting of 27.648 oligonucleotides: 2.438 control oligonucleotides and 25.210 oligonucleotides representing approximately 19.000 genes. To emulate endurance exercise, 3x10 pigs ran on a treadmill until they were exhausted. Biopsies were taken at slaughter from the muscle *longissimus dorsi* immediately after exercise (T0, 10 pigs), 1 hour after (T1, 10 pigs), and 3 hours after (T3, 10 pigs). Biopsies were also taken from 10 pigs that had not been exercising (control animals). Analysis of the transcriptional profiles revealed a large number of genes to be differentially expressed. Metabolic and regulatory pathways associated with muscle exercise were inferred from KEGG classification of the differentially expressed genes. Furthermore, gene ontology term enrichment analysis was used to identify molecular function, cellular component, and biological processes that were significantly enriched among the differentially expressed genes.

Poster 2041

Title: Genetic variability in Acetil-CoA carboxylase alpha in Spanish Churra sheep

Presenting Author: Marta García Fernández. Dpto. Producción Animal. Facultad de Veterinaria. Campus de Vegazana s/n, 24071 León (Spain)

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Abstract:

The Acetyl-CoA Carboxylase (ACACA) is a key enzyme in the biosynthesis of fatty acids in the mammary gland. This enzyme catalyzes the conversion of acetyl-CoA to malonyl-CoA, which is the activated donor of two-carbon units for fatty acids elongation. Therefore, ACACA is an interesting functional candidate to be studied in relation with the phenotypic variation observed on milk fat traits in dairy sheep. For this purpose an experiment searching SNPs was performed using a total of 24 non-related Churra sheep. Whole RNA was extracted from mammary gland and retrotranscribed in cDNA. Individual cDNA was sequenced to detect SNPs. A total of sixteen overlapping fragments were used for scanning the 9422 bp of the complete ACACA cDNA sequence. Up to now we have analyzed about 40% of the total gene sequence and 16 SNPs were detected. Three of these SNPs resulted in transversions and the other thirteen were transitions. The polymorphism situated at 2399 position (A/G) induced an amino acid change in the protein sequence (Met/Val) while the

other variations were synonymous. These and future results will be the base of subsequent statistical analyses that will test the possible influence of the identified polymorphisms on milk production traits.

Poster 2042

Title: SNaPshotTM assay successfully determines SNP for chestnut coat color in historic Thoroughbred horses

Presenting Author: *M. Whitten (Max-Planck Institute for Evolutionary Anthropology, Deutscher Platz 6, D-04103, Leipzig, Germany)

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*Mark Whitten and Michael G. Campana are co-presenting this poster at ISAG 2008. Mark Whitten shall be listed as first author, with M.G. Campana being second.

Abstract:

A single missense mutation at position C901T in the melanocyte-stimulating hormone receptor gene (*MC1R*) causes chestnut coat color in horses. Since detailed historic records have been kept by Thoroughbred horse breeders from the 17th century, we used this mutation to address the reliability of the SNaPshotTM technology, a single base extension SNP-genotyping assay, on ancient DNA extracts from the bones and teeth of historic Thoroughbred horses. In a blind test, we compared the coat colors recorded in Thoroughbred pedigree records with the genotypes obtained from bones and teeth of museum specimens, dating from the mid-18th century to the mid-20th century. In all cases, the genotypes of the historic Thoroughbred horses were consistent with the phenotypes derived from historical records. We propose that using a SNaPshotTM-genotyping assay is a simple, robust and reliable method for recording SNP data in historic samples. This methodological development opens the door to in-depth studies of past phenotypic and genotypic diversity, and allows us to address questions such as the prevalence of inheritable disease and selective breeding in the past.

Poster 2043

Title: Polymorphism of 17 microsatellite loci in Akhal-Teke, Arabian and Thoroughbred horses in Russia

Presenting Author: LYUDMILA KHRABROVA, The All-Russian Research Institute for Horse Breeding, Laboratory of Genetics, P.O. Divovo, Ryazan region, Russia

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Abstract:

For studying genetic diversity within three famous horse breeds, Akhal-Teke (n=109), Arabian (n=125) and Thoroughbred (n=476), a set of 17 microsatellites (StockMarks®) was used. Comparison between these populations demonstrated that each breed has some alleles, which were not found in other breeds yet. The oldest Akhal-Teke breed had the highest genetic variability – 139 alleles (among them 28 unique alleles) in 17 loci. There were more individual genetic variations in Akhal-Teke horses ($A_e=3,72$, $H_o=62,7\%$), than in other two breeds. In Arabian and Thoroughbred horses 102 alleles were found, among them 4 and 7 unique ones, respectively. The population of Arabian horses in Russia, traced to Tersk stud, was characterized by high consolidation ($A_e=2,92$). Horses of three studied breeds had specific allele spectrum of satellite DNA for 16 loci and similarity was found only in locus HMS2. Estimated coefficients of genetic similarity demonstrate more close relationship between Arabian and Thoroughbred horses (0,780). Akhal-Teke horses show less significant resemblance to Thoroughbred (0,727) and Arabian breeds (0,729). High polymorphism of microsatellite, blood group and biochemical markers in Akhal-Teke horses proves the ancient origin of the breed.

Poster 2044

Title: Genomic DNA markers analysis in hybrid lines of pigs

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Abstract:

Genetic variability of four porcine genes (*FUT1*, *MC4R*, *MYF4* and *RYR1*) with considerable economic importance and ten microsatellite markers (*S0068*, *SW24*, *S0107*, *S0355*, *S0386*, *SW353*, *SW936*, *SW72*, *S0070* and *TNFB*) was studied. SNP and microsatellite genotyping was done in 59 animals of crossbred pigs (CL x CLW, CLW x P and CLW x D). The following allele frequencies were observed: 0.25 for allele A and 0.75 for allele G of *FUT1* gene, 0.50 for allele A and 0.50 for allele B of *MC4R* gene, 0.79 for allele A and 0.21 for allele B of *MYF4* gene and 0.92 for allele N and 0.08 for allele n of *RYR1* gene. A total number of 72 distinct alleles were obtained for microsatellite loci. The number of alleles at individual loci ranged from 4 (*S0355*) to 10 (*TNFB*). The highest heterozygosity (above 80%) was observed for locus *S0107*, *SW24*, *S0070* and *TNFB*. The highest polymorphism information content (PIC>0.80) was determined for locus *S0107* and *S0070*. The probabilities of paternity exclusion/one parental genotype unavailable/and parentage exclusion were for this microsatellite panel 99.96%/99.00%/99.99% in all hybrid pigs. *The study was supported by grants of Ministry of Agriculture of the Czech Republic 1G58073 and QG60045.*

Poster 2045**Title: Mapping genes associated with Recurrent Airway Obstruction (RAO) in horses**

Presenting Author: June Swinburne, Centre for Preventive Medicine, Animal Health Trust, Lanwades Park, Kentford, Newmarket, Suffolk CB8 7UU, UK

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2. Helen Bogle
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4. Elizabeth Temperton
5. Vince Gerber

Abstract:

RAO is a severe inflammatory airway disease typically affecting middle-aged horses and resulting in coughing and exercise intolerance due to increased mucus production and bronchoconstriction. It is initiated by the inhalation of dust and may be a model for human asthma. Once sensitised, a horse becomes hypersensitive to airborne particles and requires careful management. RAO is a common disease caused in part by inherited factors and influenced by environmental conditions.

We have undertaken extensive sampling from Swiss Warmblood horses consisting of two half-sibling families based around RAO-affected stallions. These offspring (n=132; n=98) have been graded using a general scale 1-4 which incorporates the typical

clinical symptoms, and also for individual symptoms such as coughing and nasal discharge. We have performed a whole genome scan, using a panel of microsatellite markers (average spacing 8.5cM), in order to locate genes influencing this condition. The genotyping data is being analysed using a regression interval QTL mapping method for out-bred half-sib families. We will present our data showing the positions of QTLs associated with this condition. In addition unrelated horses, and other European Warmblood breeds, have been collected in order to perform an association study using the recently available 60,000 SNP chip.

Poster 2046**Title: Analysis of gene expression in horned and polled tissue from Brahman cattle using the Agilent Bovine microarray.**

Presenting Author: Maxy Mariasegaram, CSIRO, St Lucia, Australia

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Abstract:

We present results from a study examining the gene expression differences involved in the development of horned and polled phenotypes in Brahman cattle. Tissue biopsies were obtained from the skull region of new born calves to obtain a total of 11 samples representing phenotypes corresponding to 3 horned, 4 scurred and 4 polled animals (3 treatments). Hybridization against the 4x44K Agilent bovine microarray was conducted by the SRC microarray facility (IMB, University of Queensland). Each chip contains 44,000 unique 60mer oligos representing 21,475 individual genes which are deposited using a non-contact inkjet system. The analysis was conducted using a mixed model approach with array, chip, dye and sex as fixed effects; probe, probe*treatment, probe*sex and residual as random effects. A total of 733 differentially expressed probes were identified across various contrasts of interest i.e. Poll vs. Horn, Poll vs. Scur, and Horn vs. Scur. Preliminary inspection points to the existence of several differentially expressed pathways specific to the polled phenotype. One pathway of significant interest relates to genes involved in cell adhesion such as desmocollin (DSC), desmoglein (DSG) and related cadherin genes that appear to be up regulated in polled animals versus horned.

Poster 2047

Title: miR-26a targets the histone methyltransferase *Enhancer of Zeste homolog 2* during myogenesis

Presenting Author: Ross L. Tellam, CSIRO Livestock Industries, Queensland Bioscience Precinct, 306 Carmody Rd, St Lucia 4067, QLD, Australia

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Abstract:

MicroRNA (miRNA) are important regulators of many biological processes. However, the biological targets for most miRNA are still poorly defined. In this study we profiled the expression of miRNA during myogenesis from proliferating myoblasts through to terminally differentiated myotubes. Microarray results identified six significantly differentially expressed miRNA that were more than two-fold different in expression level in myotubes compared to proliferating myoblasts. From this list miR-26a was further examined. Over-expression of miR-26a in murine myogenic C2C12 cells induced creatine kinase activity, an enzyme that markedly increases during differentiation of the cells into myotubes and the myogenic transcription factors *myoD* and *myogenin*. These results indicated that increased expression of miR-26a promoted myogenesis. Through a bioinformatics approach we identified the histone methyltransferase, *Enhancer of Zeste homolog 2 (Ezh2)*, as a potential target of miR-26a. Over-expression of miR-26a in C2C12 cells suppressed the activity of a luciferase reporter construct fused with the 3' untranslated region of *Ezh2* but not when the putative miR-26a binding site in *Ezh2* was mutated. In addition, miR-26a over-expression caused a decrease in *Ezh2* mRNA. These results reveal a novel model of myogenic regulation whereby the up-regulation of miR-26a during differentiation acts to post-transcriptionally repress *Ezh2*, a known suppressor of skeletal muscle cell differentiation. This research links a miRNA with epigenetic mechanisms controlling myogenesis.

Poster 2048

Title: Genetic diversity and individual assignment in major cattle breeds registered in Canada.

Presenting Author: Kevin Lang, Genserve, Saskatchewan Research Council, 15 Innovation Blvd., Saskatoon, SK, Canada, S7N 2X8.

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1. Yves Plante

Abstract:

Base-line knowledge of genetic diversity in food producing animals provides valuable information to genetic conservation programs. Surprisingly, large population surveys of major beef and dairy cattle breeds are lacking in Canada. Here we explore genetic diversity in the Angus, Blonde d'Aquitaine, Gelbvieh, Hereford, Holstein, Limousin, Simmental, and Belgian Blue breeds. A total of 3,548 individuals were genotyped at twelve ISAG recommended bovine microsatellites. The effective number of alleles ranged from 3.00 (Hereford) to 3.92 (Belgian Blue). The average allelic richness ranged from 5.36 (Hereford) to 6.76 (Gelbvieh), whereas Nei's gene diversity indices ranged from 0.64 (Hereford) to 0.72 (Belgian Blue). Eighteen private alleles were detected with at least one such allele found in any given breed. A frequency-based approach could assign 98.6% of Angus cattle tested to the predefined breed but only 84.4% of the Limousin cattle were properly assigned. A Bayesian approach identified eight natural clusters. Over 98% of Holsteins could be assigned to a unique cluster, but only 76% of the Blondes and Limousin cattle could be assigned to unique groups. Genealogical records and multiloci genotype databases are now used to monitor temporal changes in genetic diversity in these and other minor breeds registered in Canada.

Poster 2049

Title: Associations of *IGF1* and *FASN* positional candidate genes with fat deposition and carcass merit traits in three unrelated beef cattle populations

Presenting Author: Changxi Li, Agriculture and Agri-Food Canada, Lacombe Research Centre, 6000 C&E Trail, Lacombe, Alberta, Canada T4L 1W1, and Department of Agricultural, Food and Nutritional Science, University of Alberta, Edmonton, Alberta, Canada, T6G 2P5

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Abstract:

Two positional candidate genes for fat deposition and carcass merit traits, insulin-like growth factor-1 (*IGF1*) on bovine chromosome 5 and fatty acid synthase (*FASN*) on bovine chromosome 19, were examined for associations with hot carcass weight, carcass average fat, lean meat yield, carcass ribeye area and carcass marbling score in three unrelated beef

cattle populations. Two gene-specific single nucleotide polymorphisms (SNPs), one for each gene, were genotyped on 463 beef steers from a hybrid population, 206 purebred Angus, and 187 purebred Charolais which had the phenotypic traits recorded. A preliminary association analysis showed that the *IGF1* SNP was significantly associated with carcass average fat and lean meat yield in the Angus population, and animals with the "TT" genotype had significantly lower carcass average fat and higher lean meat yield ($P < 0.05$). However, no association was found between the *IGF1* SNP in the other populations. The *FASN* SNP showed no associations with carcass traits in any of the populations examined. Different *IGF1*, *FASN* SNP, as well as SNP in other genes will need to be developed to further characterize genetic control of fat-related carcass traits in beef cattle.

Poster 2050

Title: Polymorphisms in porcine *TLR1*, *TLR2*, and *TLR6*

Presenting Author: Ingrid-Maria Bergman, School of Pure and Applied Natural Sciences, University of Kalmar, S-391 82 Kalmar, Sweden

Other authors (name only):

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Abstract:

The Toll-like receptors (TLRs) recognize pathogen-associated molecular patterns (PAMPs) and initiate the innate as well as the adaptive immune defences. Several *TLR* polymorphisms associated with susceptibility to infectious diseases have been identified in man. The complete coding sequences of porcine *TLR1-10* have been reported, as well as several non-synonymous SNPs in *TLR1*, 2, 4, 5, and 6. So far, there are few studies on associations with disease in pigs, but the *TLR2/6* heterodimer is known to be activated by *Mycoplasma hyopneumoniae*. The present study aims to investigate polymorphisms in porcine *TLR* genes mapping to chromosome 8, close to earlier identified QTLs for immune-related traits. Currently, polymorphisms in *TLR1*, *TLR2*, and *TLR6* are being investigated amongst animals of different breeds (Wild boar, Hampshire, Landrace, and Yorkshire). Our preliminary results indicate that fewer polymorphisms are present in *TLR2* than in *TLR1* and *TLR6*. So far, no polymorphisms have been detected in *TLR2* in the Wild boars. In *TLR6*, most of the SNPs detected are clustered in the part of the sequence corresponding to the transmembrane domain and its vicinity, while polymorphisms are evenly distributed along *TLR1* and *TLR2*.

Poster 2051

Title: Microsatellite markers for resistance to facial eczema disease in New Zealand cattle.

Presenting Author: C.A. Morris, AgResearch, Ruakura Research Centre, PB 3123, Hamilton New Zealand.

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Abstract:

Facial eczema (FE) is caused by the toxin, sporidesmin, from the *Pithomyces chartarum* fungus, found on pastures in summer/autumn in the North Island of New Zealand. In susceptible dairy cows, sporidesmin causes liver injury, and reduced milk production and survival. A secondary effect is photosensitisation, leading to visible eczema symptoms; hence the common name of the disease. Susceptibility (liver injury), measured using the enzyme GGT from peripheral blood, is a heritable trait in cattle (latest estimate 0.40 ± 0.04). In 2004-08, 11664 commercial animals in 55 Holstein-Friesian (F), Jersey (J) and FxJ herds experiencing FE following natural challenge were blood-sampled (to assay GGT activity and to extract DNA). Pedigree information was obtained from the herd owners. Breeding Values for susceptibility were returned to owners of the cows and artificial-insemination sires. Earlier genomics work in a research beef herd affected with FE had pinpointed two chromosomal regions; 384 elite dairy cows and cow-sires were then genotyped using 26 and 12 surrounding microsatellite markers. Significances of the best markers/chromosome associated with phenotype were respectively $P < 0.002$ and $P < 0.008$ overall ($P < 0.0008$ and $P < 0.02$, within breeds). The best two markers here could be part of an FE test, but additional markers are sought.

Poster 2052

Title: Fishy-egg tainting by brown-shelled layers is recessively inherited under typical commercial conditions

Presenting Author: Fiona Buchanan, Department of Animal and Poultry Science, University of Saskatchewan, 51 Campus Drive, Saskatoon, Sask., Canada S7N5A8

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Abstract:

Fishy-egg taint has long been a problem associated with feeding canola meal (CM) to brown-shelled laying hens. It is caused by a SNP in the *flavin-containing monooxygenase 3* gene (*FMO3 984c.A>T*). This mutation prevents the fishy-smelling trimethylamine (TMA) from being oxidized to the non-odorous trimethylamine *N*-oxide (TMAO), leading to an accumulation of TMA in developing egg yolks. TMA is produced from the bacterial fermentation of choline in the gut. Conflicting results from previous studies have found egg tainting to be recessive or additive. Both studies fed high concentrations of choline chloride to induce tainting, which does not reflect commercial production practices. Our objective was to characterize the inheritance pattern of fishy-egg tainting when hens are fed CM, reflecting typical industry practices. Diets consisting of 0, 6, 12, 18, or 24% CM were fed to 6 hens per genotype per diet (n=90). Three eggs were collected per hen and the yolks were analyzed for TMA concentration. The effects of diet, genotype, and their interaction were all significant ($P < 0.0001$). Only hens of the TT genotype displayed increasing yolk TMA concentration with increasing CM. We therefore conclude that fishy-egg tainting is recessively inherited under standard CM feeding practices.

Poster 2053

Title: Analysis of DNA sequence variants in candidate genes for bovine spongiform encephalopathy (BSE) susceptibility located in a QTL region on bovine chromosome 17

Presenting Author: Bertram Brenig, Institute of Veterinary Medicine, University of Göttingen, D-37077 Göttingen, Germany

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Abstract:

With the occurrence of the BSE crisis, several studies have evaluated sequence variations in the prion protein gene (*PRNP*) to assess their associations with BSE. In contrast to other mammals no functional polymorphisms in *PRNP* leading to a

susceptibility/resistance towards BSE have been reported. Recently, associations between polymorphisms in the regulatory region of *PRNP* and BSE have been shown. In addition, several genome-wide DNA marker scans have been performed and quantitative trait loci (QTL) regions significantly linked to BSE susceptibility (BTA 5 and 17) were detected. We have analyzed functional and positional candidate genes located in one of these QTL regions on chromosome 17q23-q24. The region flanks several cM upstream and downstream marker *INRA025*, which showed a genome-wide significant linkage disequilibrium in BSE positive cattle. DNA from 139 BSE cattle and 484 unaffected control cattle (Holstein, Fleckvieh, and Braunvieh) were genotyped for mutations in *RNP24*, *PSMD9*, *PITPNM2*, and *B3GNT4*. SNPs were detected in all four genes, and allele frequencies were calculated for each SNP stratified according to disease. Different statistical analysis showed that several of the SNPs are significantly associated with BSE susceptibility/resistance. **Acknowledgments: The study was supported by the German Research Foundation DFG (BR992/14-1,2) and German TSE platform.**

Poster 2054

Title: Polymorphisms associated with residual feed intake in growing cattle

Presenting Author: Denis Fidalis N. Mujibi, Department of Agriculture, Food and Nutritional Science, 4-10 AgroForestry Centre, University of Alberta, Edmonton, T6G2P5, AB, Canada.

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Abstract:

Residual feed intake (RFI) has a moderate heritability. However, collection of feed intake data is expensive. Effective selection would therefore be enhanced if marker assisted evaluation were to be used. In this study 44 SNP, previously identified to be of interest were evaluated for association with RFI. The data consisted of 464 steers tested for feed efficiency over a period of 3 years. A mixed inheritance model was used for analysis. Marker assisted breeding values (MEBV) were calculated as the sum of polygenic EBV and SNP effect whereas accuracy of prediction was estimated as the correlation between the (M)EBV and phenotype. Three SNPs were found to be highly associated with RFI ($P < 0.05$). The allelic substitution effects for these SNP were -0.19 ± 0.07 , -0.21 ± 0.06 and -0.23 ± 0.08 kg/d. Genotypic effects were

substantial with a difference of 0.59, 0.48 and 0.55 kg/d between the two homozygotes of the SNP. Together, the SNP accounted for 21% of the total phenotypic variation for RFI. Single trait heritability for RFI was increased by 13% when all three SNP were included in the model. Validation of these SNP in a different population will determine their utility for marker-assisted selection.

Poster 2055

Title: Mitochondrial DNA - an imperfect tool to study the origin of horse populations

Presenting Author: Iwona Głazewska, Dep.of Genetics, University of Gdańsk, Kładki 24 80-822 Gdańsk, Poland,

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1. Barbara Gralak

Abstract:

A popular tool used in the study of the origin of breeds and species is mtDNA analysis. However, since mtDNA is passed exclusively through the female line, it is an incomplete source of information on present-day population ancestors. The genetic analysis of Polish Arabian horses highlights some of the limitations of this method:

- The percentage of the mares from which mtDNA originates is only 7.0% of the total number of population founders, and the contribution of their genes to the present gene pool is 8.3%.
- The presence and frequency of haplotypes in the population is strictly dependent on the decisions of breeders as to preferred dam lines. This is reflected both in the different number of haplotypes in particular studs and in the large disproportion in haplotype frequency (0.2% - 31.6%).
- Genetic relations between particular dam lines revealed by mtDNA analysis are not reflected in protein or microsatellite marker analyses. If differences between the results of the latter two can be attributed to distinct selection pressure, the discrepancy between the dendrograms constructed using mtDNA data and those based on the remaining markers seems to arise from the imperfection of the mtDNA method.

Poster 2056

Title: Y-Chromosome diversity in cattle.

Presenting Author: Luis J. Royo; SERIDA-Somió, Camino de los Claveles 604, E-33203 Gijón (Spain)

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Abstract:

A total of 350 male samples belonging to 41 cattle populations of Europe (244), Africa (46) and Asia (60) were genotyped for 5 Y-specific microsatellites totaling 6 different loci. The microsatellites used were tested for non-amplification on female DNA. The number of alleles identified was 6, 3, 11, 7, 8 and 6, respectively, for locus A, B, C, D1, D2 and E. Analyses allowed identifying a total of 55 different haplogroups with 182 samples (52%) having the same haplotype (H55). Most of the haplotypes found (21) were unique. Correspondence analysis carried out on the Y-chromosome haplotypes allowed the identification of three different factors explaining, respectively, 11.7%, 9.9% and 7.4% of the variance. These factors let us separate haplotypes of zebuine origin (11) from those of taurine origin (44). Within the taurine cluster the correspondence analysis would allow separating two subgroups: the main subgroup included most individuals belonging to Continental European, Iberian and African cattle breeds, whereas the second subgroup was more frequent in Northern European cattle breeds. This grouping pattern was confirmed via Network analysis. Median-joining trees informed that both the zebuine and the main taurine clusters resembled star-like shapes. Research partially funded by grants MEC CGL2005-03761/BOS and USDA-NRI 2002-35205-11627.

Poster 2057

Title: Further characterization of equine *KIT* mutations in horses with dominant white or variable white coat color

Presenting Author: Bianca Haase, Institute of Genetics, Vetsuisse-Faculty, University of Bern, Switzerland

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Abstract:

We recently ascertained the genomic structure of the equine *KIT* gene and demonstrated that multiple independent mutations within this gene are responsible for the dominant white coat color in several modern horse breeds. Sequencing of the coding regions of *KIT* in additional unrelated Thoroughbred, Quarter Horse, and draft horse families revealed new mutations associated with depigmentation phenotypes.

In the mouse, *KIT* mutations that lead to white or white spotted coat color phenotypes are often associated with pleiotropic effects such as anemia and male sterility. We analyzed various blood parameters in dominant white Franches-Montagne horses heterozygous for the *KIT* Y717X mutation. The results showed that Franches-Montagnes horses carrying the *KIT* Y717X did not have any significantly altered blood parameters with respect to age-matched solid colored Franches-Montagnes horses. Our data indicate that *KIT* mutations may have different effects in mice and horses, and that at least the *KIT* Y717X mutation does not have a major negative effect on the hematopoietic system of dominant white horses.

Poster 2059

Title: Fine-mapping of ATPP in Japanese Black cattle and search for candidate genes with microarray

Presenting Author: Takashi Hirano, Shirakawa Institute of Animal Genetics, Odakura, Nishigo, Nishi-shirakawa, Fukushima 961-8061, Japan

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Abstract:

Abnormal teat patterning phenotype (ATPP) is characterized by one (moderate form) or two (severe form) absent teat(s) in cattle. We have mapped ATPP to the centromeric region on BTA17 (*ATPP-1*), and the telomeric and centromeric regions on BTA1 (*ATPP-2* and *-3*) with 152 affected progenies and 454 normal progenies of Japanese Black Bull A (Ihara *et al*, 2006; *Anim. Genet.* 38, 15–19). In this study, we performed a fine-mapping of the *ATPP-1* region and search for candidate genes.

Because severe ATPP animals had the *ATPP-1* risk-haplotype from Bull A, we searched shared risk-haplotype in paternal haplotypes of all 136 severe

ATPP animals, which could narrow down the *ATPP-1* region to a 3.15 Mb region. To search for genes relating with teats development, we performed a microarray analysis using teats and skin of three-day-old female mice. In the corresponding region to the *ATPP-1*, 2 genes exhibited expression differences between teats and skin with the microarray analysis. These genes may be candidate genes for ATPP.

Poster 2060

Title: Gene discovery for reproduction rate in tropically adapted Australian beef cattle

Presenting Author: Rachel Hawken, CRC for Beef Genetic Technologies, University of New England, Armidale NSW, 2351 Australia and CSIRO Livestock Industries, Queensland Bioscience Precinct, 306 Carmody Road, St Lucia, QLD, 4064, Australia

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Abstract:

Female reproductive performance is a key driver of profitability of beef cattle systems, but is generally lowly heritable and only expressed relatively late in life. The Cooperative Research Centre for Beef Genetic Technologies (Beef CRC) has established a research population consisting of both Brahman and Tropical Composite cows and various productive, reproductive, adaptive and feed efficiency measures have been recorded.

Two key components of reproduction rate of interest to industry are age at puberty and post partum anoestrus interval. Age at puberty is an issue for Brahmans in Northern Australia where they can be 3 or even 4 years of age before their first parturition. Furthermore, post partum anoestrus can delay ovulation beyond the mating season. Both of these traits result in increased production costs associated with the maintenance of barren cows and affecting their lifetime reproductive performance. The Beef CRC cattle resource, and research to identify genetic markers for components of reproduction rate will be described.

Poster 2062

Title: The Single Nucleotide Polymorphisms of Chicken 70-kilodalton Heat Shock Protein (HSP70) Gene

Qingbin Luo, Xiquan Zhang, Guanfu Yang
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Abstract:

The heat shock protein 70, which coded by the 70-kilodalton heat shock protein (HSP70) gene, is very important for the genesis of the heat tolerance of chicken. In the research of resistance breeding, HSP70 gene can be considered as the candidate loci of improving the sensibility of heat shock. But in present, researches about single nucleotide polymorphisms (SNPs) exist in chicken HSP70 gene are very limited. In this research, 591 individuals from 20 different Chinese native chicken breeds were studied. Denaturing High Performance Liquid Chromatography (DHPLC) technology was used to screen unknown SNPs existed in chicken HSP70 gene sequence. From every typical DHPLC peak, 2 individuals were selected to sequence. Homologous analysis was carried out based on the sequence information of samples that amplified from same primer to identify new unknown SNPs. From the compare of the typical DHPLC peak and sequence information, the genotype of every individual in each SNPs site was deduced. The genotype and gene frequency of every SNPs site in different chicken breeds were calculated and the difference exist among different breeds were tested. Average heterozygosity of different breeds, genetic diversity and its associate parameters, standard genetic distance (D_s) and genetic identity parameters were calculated to measure the genetic variation exist among and between different chicken breeds in HSP70 gene. Based on the genotype of every individual in each SNPs sites, the PHASE program, which compiled by the theory of most likelihood, was used to affirm the haplotype and the distribution frequency of every haplotype in different chicken breed was calculated. In the PCR products amplified by primer A5, A6, A7 and A4, 10 SNPs sites (A258G, C276G, C507T, C1040A, G1044A, C1431A, T1476C, G1500A, A1529G and C1722T) were detected. All of these SNPs exist in the code region of HSP70 gene. Especially, the mutation of C→A happened in 1040 site lead to a substitution of the amino acid histidine for proline at residue 347 and the mutation of

A→G happened in 1529 site lead to a substitution of the amino acid arginine for lysine at residue 510 in HSP70.

From the compare of the typical DHPLC peak and sequence results, the genotype of every individual in each SNPs site were confirmed. To those not sequenced individuals, the genotypes in each SNPs site were confirmed according to their DHPLC peak, same peak mean same genotype. Thus the relationship between genotypes and DHPLC peak was formed.

When consider the distribution frequency of genotype and gene in each SNPs site, significant difference ($P<0.05$) or highly significant difference ($P<0.01$) of mutation gene among different chicken breeds were found in eight sites (A258G, C276G, C507T, G1044A, T1476C, G1500A, A1529G, C1722T). But these frequency differences were not accordant with the difference of each chicken breed's geographic latitude. Generally speaking, in 3 to 4 sites, high frequency of mutation gene were found and significant difference ($P<0.05$) exist among breeds in 10 chicken breeds, they were Xinghua, Silkie, Taihe, Chongren Partridge, Yugan sooty, Qingyuan Partridge, Huiyang Bearded, Silkie, Guangdong, Taoyuan, Taibai and Henan game. In A1529G and C1722T, the mutation allele frequency in chicken breeds distributed in two southern provinces, Guangdong and Jiangxi, were significantly higher ($P<0.05$) or even highly significantly higher ($P<0.01$) than chicken breeds distributed in northern provinces. The average heterozygosity of every chicken breed in each SNPs site of the HSP70 gene were not high, the highest one was 0.2885 which exist in Huiyang Bearded and the smallest one was 0.1813 which exist in Lueyang Black. The gene divergence (H_T) of each SNPs site were different greatly, the highest H_T was 0.4971 that exist in C276G site. The H_T values in G1500A and A258G sites were also high; they were 0.4900 and 0.4387 respectively. The G_{ST} values of these sites were with the same trend. They indicated that high variation exist in these sites, not only within but also among breed groups. These three SNPs sites, C276G, G1500A and A258G, were suit to be used as marker sites to judge the genetic diversity of HSP70 gene in different chicken breeds.

56 haplotypes were inferred in the 10 SNPs sites of HSP70 gene exist in all of these 20 chicken breeds. 28 of them were truly existed in at least one individual. In these 28 truly existed haplotypes,

H14 H21 H5 H6 H7 H9 H10 H12 H

H15, H20, H25, H26, H31, H34 were the main types and distributed in most breed groups with relative high frequency. These 28 truly existed haplotypes hold 81% of all haplotypes frequency in this study; only 19% of haplotypes cannot be confirmed. In the truly existed haplotypes, 14 main types hold 85% of all haplotypes frequency, and the remaining 14 haplotypes hold 15% of all haplotypes and be considered as rare haplotypes. Great differences of the distribution frequency of different haplotypes among breed groups were found. In all of these haplotypes, only H14 and H21 were distributed in every breed. The distribution frequency diversity of haplotypes base on 10 SNPs in HSP70 gene were very abundant in the 20 chicken breeds.

Poster 2063

Title: Expression of genes associated to omega 3 fatty acid content in bovine skeletal muscle

Presenting Author: Isabel Tupac-Yupanqui. Dpt. Animal Production. Veterinary Faculty. UCM. Madrid Spain

Other authors (name only):

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Abstract:

Fatty acids are determinant of the sensorial quality of meat. Moreover, long chain unsaturated fatty acid (PUFA) are important for health purposes. We analyzed 120 bovine individuals for fatty acids profile and different candidate genes with the aim of associating the expression of particular genes to the amount of PUFA in skeletal muscle. Gene expression was measured through real time RTqPCR in samples from *Longissimus dorsi* of animals presenting different levels of long chain fatty acids omega 3 (LCFA ω 3), with the result of a significant higher expression of AANAT and UCP2 and down-regulation of AHA1 in those animals showing a high LCFA ω 3 phenotype. AANAT is an enzyme which catalyses the transformation of serotonin to N-acetylserotonin, the limiting step in melatonin synthesis. Both N-acetylserotonin and melatonin have been described as having protective effects against lipid peroxidation. UCP2 protein, placed on the inner mitochondrial membrane, has the ability to dissipate an excess of membrane potential produced by a strong proton gradient that induces the genesis of reactive oxygen species (ROS), rising the oxidative stress levels in the entire cell. During lipid peroxidation, fatty acids (mostly the PUFAs) lose

insaturations and suffer oxidative degradation. Up-regulation of genes AANAT and UCP2 seems to prevent against ROS aggressions avoiding LCFA ω 3 to be degraded. Moreover, the low expression of AHA1 (a molecular co-chaperone) in samples of high LCFA ω 3 phenotype, reflects a reduced cellular oxidative stress.

Poster 2064

Title: Y-chromosome haplotype diversity in cattle

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Abstract:

Cattle Y-chromosome diversity was investigated in Portuguese and Spanish native breeds, and related Creoles from the Americas. European and Zebu breeds which may have influenced Creole cattle were also analyzed. Haplotypes defined by five SNPs, one indel and 7 microsatellites in the non-recombining region of the chromosome were obtained for 748 males representing breeds from Argentina (2), Brazil (1), France (2), India (2), Mexico (2), Paraguay (1), Portugal (13), Spain (3), The Netherlands (1), United Kingdom (5) and United States of America (1). The 24 haplotypes identified were classified according to defined patriline Y1 and Y2 of *B. taurus* and Y3 of *B. indicus*. The highest diversity over all breeds was found within Creoles (0.484 \pm 0.090). Y3 haplotypes were present in high frequency in Creoles consistent with widespread male-mediated *B. indicus* introgression. Among European cattle, the Iberian breeds were the most diverse (0.234 \pm 0.259) showing 7 private haplotypes (H2Y1, H5Y1, H7Y2, H8Y2, H10Y2, H12Y2, H23Y1). The Y1 patriline found in Iberian cattle, and also reported in aurochs, could represent ancient local haplotypes. The presence of alleles *INRA189-90* in Caracu and *INRA189-104* in Portuguese, Canary Island and British White breeds suggested influence of African cattle in the genetic makeup of these breeds.

Poster 2065

Title: Application of 19 microsatellites DNA markers for paternity control in dogs.

Presenting Author: Anna Radko, National Research Institute of Animal Production, Immuno and Cytogenetics Department, Krakowska 1, 32-083 Balice, Poland

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1. Ewa Słota

Abstract:

Microsatellite DNA become the international standard for individual identification and parentage verification in canine breeds. Only one commercial kit prepared by Applied Biosystems company was available on market so far for canine genotypes. This set included 10 microsatellite markers, which was not always enough for confirmation of parentage. At present the assay of 19 microsatellite markers is available for canine parentage testing, offered by Finnzymes Diagnostics company. This kit contains the following 19 loci: AHTk211, CXX279, REN169O18, INU055, REN54P11, INRA21, AHT137, REN169D01, AHTTh260, AHTk253, INU005, INU030, FH2848, AHT121, FH2054, REN162C04, AHTTh171, REN247M23 and Amelogenin. These markers are included in the panel of loci recommended by the ISAG. The aim of this study was to analyse the polymorphism of 19 microsatellite markers and their usefulness for parentage verification in 28 individuals of Borzoi breed in Poland. Amplified PCR products were analysed by automated sequencer ABI PRISM 3130xl Genetic Analyser. The number of alleles per locus varied from 3 (INU005) to 8 (REN162 and AHT171) alleles. Except the locus INU005, which showed the lowest variation (PIC=0.396, H=0.438) the calculated PIC values exceeded 0.6 and H values ranged from 0.677 (INUQ55) to as much as 0,84 (REN162). Based on PE_C it was found that incorrect pedigree in this breed can be excluded with 99.9998% accuracy using DNA analysis.

Poster 2066

Title: POLYMORPHISM OF THE *OPN* GENE AND mRNA EXPRESSION PATTERNS OF THE *OPN* IN SOWS OVIDUCT

Presenting Author: ...Agnieszka Korwin-Kossakowska, Institute of Genetics and Animal Breeding, Polish Academy of Sciences, Jastrzębiec, Poland

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4. Grażyna Sender

Abstract:

The aim of our study was to find the relationship between the polymorphism of the *OPN* (osteopontin) gene, it's expression in female ovaries and oviduct and reproductive performance traits of sows.

Sixty sows after first mating was slaughtered and tissues samples of the ovaries and oviduct were taken. The total RNA was extracted from the frozen tissues according to the protocol of Chomczyński and then subjected to Real Time PCR analysis.

The polymorphisms within *OPN* gene for all sows were examined, simultaneously. The polymorphism in intron 6 of this gene was examined base on literature data (but with PCR fragments size 498 and 193 bp). The new polymorphism in promoter region was found. The fragment of 274 bp was chosen because of its contents of tree specific sites: type II collagen "silencer" sequence at -682 glucocortycoid response site at -658 and CAAT box -592. The results of first parity (number of piglets born alive and piglets weaned, litter weight at born and at weaning, age at farrowing) were also collected. The statistical analysis between the polymorphism of the *OPN* gene, the expression of this gene in ovaries and oviduct tissues and reproductive performance traits of the sows is under evaluation.

Poster 2067

Title: DNA Assignment for profitable production decision

Rogberg-Muñoz, A¹; JP Lirón¹; MV Ripoli¹; EI Francisco¹; EE Villegas-Castagnasso¹, DM Posik^{1,2}, P Peral-García¹; G Giovambattista¹.

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Abstract:

Extensive animal production can compromise relation between productivity and information; when both are necessary, technology can be useful. Wagyu breed is known for its marbling capacity and for improving carcass quality. Feedlot feeding system is more expensive than pasture, making economically important to decide which animal will be feedlot fattened. This study was done in a pasture extensive, commercial herd. 164 Angus and 26 half Wagyu/Angus dams were artificial inseminated with Wagyu, and mated after with Angus bulls. At 15 months, animals were moved for 300 days to a feedlot and Breed assignment was performed (there was no

more semen available for paternity). DNA was extracted from hair and eleven microsatellites were genotyped. Wagyu percentage was determined using Structure 2.2 software, Angus and Wagyu animals were included as control populations. From the 190 dams, 29 animals were Angus sired and 161 Wagyu sired. Ultrasound IMF is being scanned every two months, and all the animals Wagyu sired assigned (50% Wagyu or more) have satisfied expected grade and this was significantly higher; this will be confirmed at slaughter stage. This method appears to be adequate for select animals when breed assignment is economically important, i.e. for animal buyers for long term feedlots.

Poster 2068

Title: Whole genome association study of adaptive traits in tropical beef cattle

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Abstract:

Tropical adaptation in northern Australia is characterised by an animal's ability to survive stressors such as ecto-parasites (ticks and buffalo flies), endo-parasites (worms), heat, humidity and seasonal nutrition. The Cooperative Research Centre for Beef Genetic Technologies has developed a resource population and quantified several of these tropical adaptive measures in terms of tick scores, faecal egg counts, fly lesion scores, rectal temperatures under hot conditions, coat score, coat colour, and flight time as a measure of temperament. A sub set of this resource population constituting 582 Brahman females was genotyped using the Affymetrix 10K SNP panel. Genome wide association analyses were conducted using a variety of parametric and non-parametric methods. Significant chromosomal regions across the genome were identified for various adaptive traits under study. In this report, we present these broader chromosomal regions and highlight the possibility of developing genetic markers for implementation in the selection programmes.

Poster 2070

Title: Effect of a single nucleotide polymorphism within promoter region of the bovine myogenic factor 5 (*Myf5*) gene on the gene expression in bovine *m. longissimus dorsi*.

Authors; Presenting Author: Dagmara ROBAKOWSKA-HYŻOREK, Institute of Genetics and Animal Breeding, Polish Academy of Science (IGAB PAS), Jastrzębiec, 05-552 Wólka Kosowska, Poland

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Abstract:

Myogenic factor 5 (*Myf5*), belongs to the MRF family of transcription factors which control the progress of myogenesis and are candidates for molecular markers of meat production in farm animals.

We detected nine single nucleotide polymorphisms (SNPs) located in the promoter region of the bovine *Myf5* gene. The polymorphisms found are: G-1569A, A-823G, T-472G, G-882T, C-741G, T-511G, C-1185T, G-1466A (RFLP/*AvaII*), C-1128T (RFLP/*NsiI*). Using a Real Time PCR method we analyzed the possible influence of the promoter mutation G-882T (represented by all 3 genotypes - GG, GT, TT) on the expression levels of the *Myf5* gene in the muscle (*m. longissimus dorsi*) of 12-months-old bulls of different breeds. As a reference the expression of the *TBP* gene was used. Statistically significant differences (evaluated by ANOVA method) were found in the *Myf5* expression levels between GG and TT genotype bulls ($p < 0.001$), and between GT and TT genotype ($p < 0.01$) bulls. TESS software analysis have shown that the G→T substitution creates the *Hoxa5* transcription factor binding site, while in the G allele the LEF-1 site is created. These results suggest that the G-882T polymorphism may affect *Myf5* expression at the transcript level in the adult bovine muscle.

Poster 2071

Title: *UCP2*: Structural and expression studies in Casertana pigs

Presenting Author: MARIASILVIA D'ANDREA
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Abstract:

Casertana is an autochthonous pig breed characterized by massive accumulation of backfat and low growth rate. In this study Casertana pigs were compared with Large White focusing on genetic variation in *Uncoupling Protein 2 (UCP2)*. *UCP2* is an uncoupling protein that functions as an uncoupler of oxidative phosphorylation, thus dissipating energy as heat. The *UCP2* gene is expressed in adipose tissue and skeletal muscle. Several studies found that uncoupling proteins are associated with the biological traits of body weight, resting metabolic rate and energy conversion.

17 Casertana and 16 Large White, of the same age and raised outdoors in the same environmental conditions, were analysed.

Sequencing the whole gene in Casertana and Large White 3 new SNPs were found. Two PCR-RFLP methods and a SNaPSHOT method were used to genotype the population.

The Real-Time PCR was carried out by amplifying, in singleplex, the target (*UCP2*) and the reference (*TATABP*) genes. Results were analysed by using the relative standard curve method.

The difference between the two groups in *UCP2* mRNA levels was statistically ($p < 0.01$) supported in muscle and adipose tissues as well. These findings suggest that *UCP2* is a valid candidate gene for meat production traits.

Poster 2072**Title: GENETIC CHARACTERIZATION OF ORLOV TROTTER**

Presenting Author: LILIYA KALINKOVA, The All-Russian Research Institute for Horse Breeding, Laboratory of Genetics, Divovo, Ryazan region, Russia

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Abstract:

Orlov Trotter is the oldest Russian cultural breed of horses. The history of the breed goes back to 1776 when the stud at Khrenovoe was established by the count Alexei G. Orlov. Founder animals were imported from different countries of the world: Arabia, Denmark, the Netherlands, Italy, Spain, England, Germany, Poland, Persia and also from regions of the Don River, the Caucasus and Central Asia. In the XX century the unique breed suffered an extreme decrease in population size. In this study genetic variation of Orlov population ($n=803$) was evaluated in 8 blood polymorphic systems (EAA, EAC, EAD, EAK, Al, Es, Tf, Xk). In addition genetic

diversity within a group of leading stallions ($n=20$) was measured with the use of 17 microsatellite loci (StockMarks®). DNA was isolated from frozen semen stored in liquid nitrogen and hair roots. Allele frequencies, the number of alleles per locus and heterozygosities were determined. The results were compared to genetic variation observed in other horse populations of Russia (28 breeds). The level of genetic diversity in Orlov Trotter was relatively high. The panel of microsatellites evaluated in the group of Orlov stallions showed very high heterozygosity ($H_o=42,11$ to 100%, the mean $H_o=70,13\%$). Effective numbers of alleles were the highest at loci ASB17 (5,48), VHL20 (5,16), HMS2 (4,56), HMS3 (4,46), ASB23 (4,13). The substantial genetic variability of Orlov Trotter may be due to extraordinary various origins of historical founders of the breed.

Poster 2073**Title: Genetic characterization of the Jeju horse compared with other horse breeds reared in Korea by means of microsatellites**

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The Jeju horse (JH) is a native Korean breed selected for riding, racing and meat; its name comes from Jeju Island in South Korea. According to Jeju Province, the JH comprise about 850 herds. They have been separated into distinct areas, and have been used as a racing horse at the Jeju race park of the Korea Racing Authority. Genetic characterization of the JH was performed to construct a correct pedigree of the JH family. A total of 581 horses including 278 JH, 134 Thoroughbred-TB, 118 Jeju racehorse-JR, 31 Mongolian-MG, 12 Warmblood-WB and 8 Miniature-MN were genotyped using 20 microsatellite loci (AHT4, AHT5, ASB2, ASB17, ASB23, ASB25, CA425, HMS1, HMS2, HMS3, HMS5, HMS6, HMS7, HTG4, HTG6, HTG7, HTG10, LEX3, TKY297, VHL20). The CERVUS and DISPAN software were used to calculate allele frequencies, heterozygosity, PIC, PE, genetic distances and phylogenetic trees. All microsatellites were polymorphic in this study. The number of alleles varied from 5 to 13 (mean 8.45) in the JH. The observed heterozygosity and expected heterozygosity ranged from 0.4176 to 0.8647 and from 0.3731 to 0.8809, respectively. The polymorphic information contents (PIC) ranged from 0.3470 to 0.8670. AHT4, AHT5, ASB2, ASB17, ASB23, CA425, HMS2, HMS3, HMS6, HTG10, LEX3 and VHL20 loci had

relatively high PIC values (> 0.7). The total exclusion probability (PE) of the 20 microsatellite loci was 0.9999 in the JH. Phylogenetic trees constructed using Neighbour-joining method showed two clear separate clusters: the first includes the JH, MG and MN, the second contains the JR, WB and TB. These results provide basic information for developing an accurate pedigree and will be useful in making decisions regarding conservation of the JH.

Poster 2074

Title: Assignment of porcine full-length cDNA sequences and ESTs on the pig draft genome sequences

Presenting Author: Hirohide Uenishi, National Institute of Agrobiological Sciences
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Abstract:

We have determined more than 10,000 complete sequences of pig cDNA clones derived from full-length-enriched cDNA libraries. These sequences are useful for exploration of candidate genes for economically important traits in pigs, as well as functional analysis of pig genes. We estimated chromosomal locations of cDNA sequences that had been fully determined and contig sequences of ESTs used for development of SNPs, which had been localized on a linkage map. Meanwhile, pig genome sequencing is now performed by the international consortium using BAC clones, and draft sequences of nearly half of the genome have been open to the public. We used draft sequences of the BAC clones mapped on the pig chromosome for the alignment of the cDNA and EST contigs. We could determine the relationship of 5,739 cDNA clones with the BAC sequences among 10,147 clones. Furthermore, among 967 sequences used for development of SNPs that were mapped on chromosome, we could tag 542 to the BAC sequences, and the order of the assigned sequences on chromosomes well coincide with the BAC fingerprint map of the consortium. These data demonstrate usefulness of full-length cDNA sequences and ESTs for SNP development in particular regions or genes of interest.

Poster 2075

Title: Nutritional effects on epigenetic modification and their inheritance – F0 generation

Presenting Author: Martin H. Braunschweig, Institute of Genetics, University of Berne, Bremgartenstrasse 109A, CH-3001 Berne

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Abstract:

In a three generation pig feeding experiment we are investigating whether differential nutrition of F0 boars affects gene expression and carcass traits in the next but one F2 generation. It is hypothesized that the nutrition related transgenerational effects are epigenetic in nature and the transmission is through the male line. This implies that nutrition induced epimutations are not reprogrammed in gametogenesis or in the early embryo resulting in transgenerational epigenetic inheritance. Two groups of eight F0 boars received either a control diet or a methyl donor supplemented diet. Upon puberty the F0 boars were mated to sows to produce the F1 male generation, which was then later used to produce the final F2 generation. We measured *IGF2* and *IGF2R* gene expression in muscle, kidney and liver tissues of F0 boars. The imprinting status of the *IGF2* gene in these tissues was analysed by means of the microsatellite *SWC9*. DNA methylation patterns were determined in differentially methylated regions (DMR) in the *IGF2* gene. The expression levels of *IGF2* and *IGF2R* genes do not differ between the two groups of F0 boars. DNA methylation patterns around the *IGF2* locus in tissues of F0 boars receiving either the methyl donor supplemented diet or the control diet are presented.

Poster 2076

Title: Mitochondrial DNA variability and genetic relationships in four Italian donkey breeds (*Equus asinus*)

Presenting Author: M.C. Cozzi, Dipartimento di Scienze Animali - Sezione di Zootecnica Veterinaria - Università degli Studi di Milano, Via Celoria, 10, 20133 Milano – Italy

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Abstract:

The genetic variability of mitochondrial D-loop DNA (mtDNA) in 91 individuals from 4 donkey breeds reared in Italy (Asinara donkey ASD, Sardinian donkey SAD, Romagnolo donkey ROD and Ragusano donkey RAD) were analysed. A 478 bp fragment of the mtDNA was amplified and sequenced. Sequences obtained were aligned with the complete donkey mtDNA sequence (GenBank#X79547), using the CLUSTALX software. 20 polymorphic sites and 10 haplotypes were identified. Overall and per breed haplotype diversity (Hd), nucleotide diversity (π), average number of nucleotide differences (k) and Fu's *F_s* statistic were calculated using DnaSP 4.2 software. Hd ranged from 0,638 (ASD) to 0,867 (RAD), overall 0,822; π ranged from 0,005 (RAD) to 0,020 (ROD), overall 0,017; k ranged from 2,200 (RAD) to 9,750 (ROD) overall 8,315. These results indicated moderate haplotype diversity. Italian donkey mtDNA haplotypes were compared with about 400 worldwide donkey mtDNA sequences from on-line database. Genetic distances were estimated by the Kimura-two parameter method and a phylogenetic tree was constructed using the UPGMA algorithm by MEGA 4.0 software package. Median-joining network and mismatch analysis were calculated in order to investigate haplotypes relationship using NETWORK 4.5.0.1. The results were discussed to investigate origin of the Italian donkey breeds.

Poster 2077**Title: The effect of gene CAST polymorphism on productive traits in pigs**

Presenting Author: ...Paweł Urbański, Institute of Genetics and Animal Breeding, Polish Academy of Sciences, Jastrzębiec, Poland

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Abstract:

Calpastatin is an endogenous inhibitory protein acting on calpain, the calcium dependent cysteine proteases in animal cells and the system calpain-calpastatin is important in postnatal skeletal muscle cells. The porcine *CAST* gene has been mapped to chromosome 2.

The aim of this study was to evaluate the effect of two mutations localized in exon 14 porcine *CAST* gene, described by Ciobanu on growth rate and carcass traits. The study covered about 620 gilts of two breeds: Polish Large White (PLW) and Polish Landrace (PL) and synthetic line L990. The pigs were

fattened and slaughtered at the Pig Testing Station of the National Research Institute of Animal Production. In our studies we observed a significant relation between mutation identified with *PvuII* restriction endonuclease and different productive traits.

Respecting to second studied of mutation, recognized by *HpyI88I* enzyme we did not the polymorphism.

The animals which we analysed in our studies were monomorphic.

In our opinion the calpastatin gene may be have an important role in pig breeding and selection, but a further study should be continued on other pig breeds and lines.

Poster 2078**Title: Analysis of μ -Calpain and Calpastatin genes in Piemontese breed**

Presenting Author: Liliana DI STASIO, Dip. Scienze Zootecniche - Via L. da Vinci 44 - 10095 Grugliasco (Italy)

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Abstract:

Mutations at the μ -*Calpain* (*CAPNI*) and *Calpastatin* (*CAST*) genes have been shown to affect beef tenderness and the reported associations have been confirmed by validation studies. As no data exist on purebred Piemontese subjects, we analysed the variability at the two loci in the breed for evaluating the usefulness of integrating the markers into selection programmes. Fifty-three unrelated Piemontese subjects, representative of the breed, were tested for *CAPNI* g.5709G>C (AF252504), *CAPNI* g.6545C>T (AF248054), and *CAST* g.282C>G (AY008267) mutations, with PCR-RFLP procedures set up in our laboratory, and the allele frequencies were calculated. For a preliminary field evaluation of the marker effects, seventy meat samples from commercial Piemontese animals were genotyped for the three SNPs and tenderness was measured as shear force (WBs) at different days *post-mortem*. The frequencies of the *C* alleles, shown to be associated with improved tenderness, were 0.14 for *CAPNI* 5709, 0.52 for *CAPNI* 6545 and 0.63 for *CAST* 282. The observed genotypic distributions were in agreement with HW equilibrium. Significant associations with meat tenderness ($P=0.01$) were observed only for the *CAST* 282 marker, with a favourable effect of the *C* allele, consistent during the ageing (-1.997 kg at day 1 to -0.956 kg at day 11).

Poster 2079

Title: Localisation of the causative gene for reflex epilepsy in chicken

Presenting Author: Marine Douaud, UMR génétique Cellulaire, INRA Chemin de Borde-Rouge, Auzeville BP 52627, 31326 CASTANET-TOLOSAN CEDEX, FRANCE

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8. ...FrédériquePitel

Abstract:

In human, genetic reflex epilepsy is a type of idiopathic epilepsy in which a particular sensory stimulus evokes paroxysmal manifestations only in genetically predisposed subjects. The Fayoumi Fepi strain of chickens carries a recessive autosomal mutation causing generalized reflex epilepsy in homozygous animals after stroboscopic stimulation. The mutation was discovered in several animals in 1970 by Crawford who decided to build a synthetic strain of Fepi. The objective of this project is to identify the causative recessive mutation "*epi*", determining the epileptic phenotype. Two informative families were obtained by crossing two carrier sires with 5 or 6 affected homozygous dams each, giving a total of 209 offspring. Almost half of the offspring were affected. A genome scan was performed using informative molecular markers chosen from public databases or developed specifically. The marker showing significant linkage with the *epi* mutation mapped to a microchromosome where the genome sequence is incomplete. By developing additional informative microsatellite and SNP markers, mainly through comparative mapping with human, we were able to locate the *epi* locus in a 6cM region. Thus, insufficient coverage of the microchromosomes in the genome sequence may limit strongly the possibility to map genes of major effect or QTLs

Poster 2080

Title: Mining genomic DNA from defined regions of bovine chromosome 6 for novel transcripts

Presenting Author: Rosemarie Weikard, Forschungsinstitut für die Biologie landwirtschaftlicher Nutztiere (FBN), 19196 Dummerstorf, Germany

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2. Christa Kühn

Abstract:

The identification of the molecular background underlying the QTL for production, health, and conformation traits mapped on bovine chromosome 6 (BTA6) requires a well-annotated sequence map of the chromosomal regions comprising the QTL intervals. To complete the current BTA6 transcript map, we focused on targeted isolation of transcribed sequences from regions poorly covered with gene information. Using the method of exon trapping, 90 unique exon trapping sequences (ETS) were discovered from 14 BAC clones. Expression analysis showed that most of the ETS displayed a distinctive expression pattern in a multi-tissue panel indicating that they are functional constituents of the bovine transcriptome regulated tissue-specifically. Whereas 79% of the unique ETS were assigned to BTA6 sequence scaffolds, only seven of them displayed identity to known bovine transcript or gene sequences. The majority of identified ETS is located in intergenic regions and thus, represent novel noncoding transcripts. The results of our study demonstrate that exon trapping is very useful for targeted screening for novel transcripts located in chromosomal regions deficiently endowed with annotated gene information. The novel transcripts are valuable for functional studies, completion of the genome sequence assembly and transcription map of BTA6, and the identification of candidate genes underlying QTL.

Poster 2081

Title: Whole porcine genome linkage map based on the 7K porcine SNP Chip

Presenting Author: Rikke Kirstine Kaae Vingborg, Department of Genetics and Biotechnology, Faculty of Agricultural Sciences, University of Aarhus, Blichers Allé 20, P.O. box 50, DK-8830 Tjele, Denmark

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7. Christian Bendixen

Abstract:

Constructions of high-density linkage maps are required for identification of QTLs across genomes. The generation of porcine sequences has provided a solid base for SNPs (Single Nucleotide Polymorphisms). The 7K porcine Illumina Custom

Infinium® Bead SNP Chip was used to genotype a previously described family material, consisting of offspring from 12 different boar families, encompassing 236 sows and 5,195 piglets.

The data set was evaluated, and SNPs that deviated from Hardy-Weinberg equilibrium, contained extra alleles, demonstrated software or genotyping limitations or did not segregate were eliminated. Only SNPs genotyped in more than 90% of the animals were included, resulting in a data set comprising 4366 SNPs.

The calculations were performed on sow families, since all sows had litters with more than one boar. This established the possibility for generating genetic maps of all chromosomes, even though a close network of SNPs segregating in all families was missing.

The SNPs were physically assigned to all 20 porcine chromosomes. All sequences were blasted against the available porcine chromosomes assembly database, and through comparison with the human porcine comparative database. Here we present the preliminary evaluations of SNPs.

Poster 2082

Title: Association of SNPs from a QTL region for mastitis with proliferation of *S. aureus* in mammary epithelial cells and udder health phenotypes

Presenting Author: Hannes Joerg, Institute of Animal Sciences, ETH Zurich, Switzerland
Conference registration number of Presenting Author:

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Abstract:

Adherence to and invasion of epithelial cells in the bovine udder play a significant role in the pathogenesis of *Staphylococcus aureus* mastitis. In the present study, primary mammary epithelial cell cultures of 67 cows were incubated with *S. aureus* strains and the proliferation of *S. aureus* bacteria were categorized. The cows were daughters of 4 bulls having different QTL for mastitis on chromosome 2. The 67 cows were separated in two groups due to their paternal allele of 12 SNPs belonging to the QTL region. The highest differences of proliferation of bacteria were between the groups separated by the SNP +777 G/C of the CXCR2 gene. The group with paternal G-allele and C-allele had a mean proliferation of 1.80 ± 1.16 and 3.15 ± 1.18 , respectively. The same

groups did also show significant differences in udder health phenotypes. The least square mean of somatic cell score corrected for bull and lactation effects were 3.60 for the C-allele and 2.56 for the G-allele. The huge differences in somatic cell score is also due to the small number of cows in this project, but even if the effect will be reduced in the population it is still an interesting QTL for mastitis.

Poster 2083

Title: An alternative angiogenesis pathway seems to be activated in Canine Inflammatory mammary carcinoma

Presenting Author: Susana Dunner Dpt. Animal Production. Veterinary Faculty. UCM. Madrid Spain

Other authors (name only):

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Abstract:

Canine inflammatory mammary carcinoma (IMC) can be considered as a spontaneous animal model of human inflammatory breast carcinoma (IBC). It is the most malignant type of canine mammary cancer with extremely poor prognosis. In IMC, growth, proliferation and metastasis of breast cancer are not angiogenesis-dependent processes, as an alternative pathway called Vasculogenic Mimicry (VM) has been reported. In VM tumour cells form tube-like structures similar to micro-vascular channels in order to feed themselves. To throw light on the active IMC angiogenesis pathway, we used RNA samples from 23 bitches (11 IMC, 6 "non-inflammatory" mammary carcinoma with histological malignant grade III, and 6 healthy mammary tissues) and measured the expression through real time quantitative PCR of different candidate genes commonly associated with angiogenesis. Some of these genes are down-regulated in IMC when compared to the other samples, specially VGFD which expression is significantly low ($p < 0.008$), the receptor VGFR3 ($p < 0.01$) and also VGFA and TIE2 ($p < 0.03$). These results would indicate that a different pathway is activated in this special type of mammary cancer that allows metastasis to develop through VM in which proper tumoral cells are transformed into endothelial-like cells producing a rapid, efficient way to generate a vascular net that contributes to the high malignancy showed by IMC and IBC.

Poster 2084

Title: Insights into equine osteochondrosis based on genetic markers in Hanoverian warmblood horses

Presenting Author: Virginie Lampe, University of Veterinary Medicine Hannover, Institute for Animal Breeding and Genetics, Buenteweg 17p, 30559 Hannover, Germany

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Abstract:

The objective of this work is fine mapping of QTL regions for osteochondrosis (OC) and osteochondrosis dissecans (OCD) in horses using intragenic single nucleotide polymorphisms (SNPs) and microsatellites in order to identify genes involved in the pathogenesis of equine OC. We developed SNPs in positional candidate genes and microsatellites located in quantitative trait loci (QTL) regions identified through a whole genome scan in Hanoverian warmblood horses. The whole genome scan included 14 paternal half-sib families with 211 horses and more than 260 highly polymorphic microsatellites. Traits regarded were OC and OCD in fetlock and hock joints. Genome-wide significant QTL for OC were located on ECA2, 4, 5 and 16. Comparative human-equine maps were employed to select candidate genes in the QTL regions. SNP markers were developed using equine BAC end, EST, whole genome shotgun sequences and equine contigs. We were able to identify significantly associated SNP markers within these QTL regions for OC. The SNPs were significant in their genotypic effects on the fetlock OC and/or hock OC. Further SNPs will be tested using the Illumina Equine BeadChip. This work is an important step towards an equine SNP marker set to be employed in horse breeding.

Poster 2085

Title: Bovine placenta transcript analysis using 454 sequencing

Presenting Author: Frank Panitz, Dept. of Genetics and Biotechnology, University of Aarhus, Blichers Allé, 8830 Tjele, Denmark

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1. Pernille K. Andersen
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Abstract:

The recent introduction of new high-throughput sequencing technologies (e.g. 454 LifeSciences; Solexa) allows generation of high quality transcriptional profiles from large-scale sequencing of cDNA fragments. Here, we compared different strategies for cDNA synthesis with respect to transcript abundance, gene content and polymorphic

variation: By using either oligo-dT or random hexamer priming during cDNA synthesis two cow placenta cDNA libraries were obtained and sequenced in a single run on the GS20 sequencer with one region for each priming method. The sequences were clustered and compared to various databases in order to annotate the transcripts. Finally, single nucleotide polymorphisms (SNPs) were identified and mapped against the cattle genome to assess the genetic variation.

Poster 2086

Title: Identification of differentially expressed proteins during an *Actinobacillus pleuropneumoniae* infection by proteome analysis of BALF and lung tissue

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6. FUGATO-consortium IRAS

Abstract:

Acute respiratory diseases pose severe economical problems in pig breeding. With FUGATO (Functional Genome Analysis in the Animal Organism) and its subproject IRAS the German Federal Ministry of Education and Research (BMBF) intends to support the identification of genetic markers that facilitate breeding with regard to immune defence and resistance. To identify potential biomarkers associated with a reduced susceptibility to bacterial respiratory tract pathogens an aerosol infection model for *Actinobacillus pleuropneumoniae* was utilized. Lung tissues from infected animals were subjected to proteomics analysis using two different strategies: The first employed lectin affinity chromatography of bronchoalveolar lavage fluid (BALF) to compare glycoprotein profiles. The second examined the recruitment of proteins into lipid rafts during infection. Mass spectrometry of identified protein spots on 2-D gels revealed differential expression of seven glycosylated proteins (Fetuin A, α -1-acid glycoprotein, α -1-antichymotrypsin, hyaluronidase, surfactant protein D, hemoglobin, haptoglobin) and two raft-associated proteins (lactoferrin, protein similar to S100 A9). Fluorescence Difference Gel Electrophoresis (2-D DIGE) was performed to detect race varieties in the glycoprotein expression at defined point of times during the infection. These proteins are currently under further investigation regarding their

identity and their putative role as genetic resistance markers in the respiratory tract.

Poster 2088

Title "Positional cloning of the causative mutation for bovine dilated cardiomyopathy (BDCMP)"

Presenting Author: Marta Owczarek-Lipska, Institute of Genetics, Vetsuisse Faculty, University of Berne, Bremgartenstrasse 109a, 3001 Bern, Switzerland

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Abstract:

Bovine dilated cardiomyopathy (BDCMP) belongs to the group of progressive degenerative disorders of the myocardium. BDCMP is a severe and hereditary disease and was worldwide observed in cattle of Canadian Holstein origin. The main symptoms of BDCMP are edema of the brisket, congested jugular veins and tachycardia with gallop rhythm. Necropsy findings are cardiomegaly with hypertrophy and ventricle and atrial dilatation. The typical pathological sign is a progressive transmural myocardiofibrosis. The age at onset of BDCMP range usually from two to four years. Pedigree analyses performed in Canada, Japan and Switzerland led to the common ancestral Holstein-Friesian bull ABC Refection Sovereign. An autosomal recessive mode of inheritance was suggested and the BDCMP locus was mapped to the bovine chromosome 18 (BTA18). A BAC contig consisting of 10 INRA BACs and 24 CHORI BACs was constructed by PCR screening. The BAC contig spans the interval between microsatellites *MSBDCMP06* and *BMS2785*, corresponding to a physical distance of about 6.7 Mb. Recently, the region harboring the disease locus was fine mapped to an interval covered by four adjacent INRA BACs. Currently, we are focusing on the confirmation and on the further fine mapping of the interval containing the BDCMP locus.

Poster 2089

Title : Characterization of a deletion in the cattle Pro-opiomelanocortin gene.

Presenting Author: Heather Deobald, Department of Animal and Poultry Science, College of Agriculture and Bioresources, 51 Campus Drive Saskatoon, Sk, Canada S7N 5A8

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Abstract:

Pro-opiomelanocortin (*POMC*) is a complex gene with four gene products that play roles in the appetite, stress and pigmentation pathways. Previous data suggest that a SNP in *POMC* (*POMC c.288C>T*) is associated with shipping and hot carcass weight in cattle. While optimizing this variant using real time PCR an unexpected shift in melting temperatures was observed. Sequencing these animals revealed a 12 bp deletion (*POMC c.293_304delTTGGGGGCGCGG*) that results in four amino acids being removed (Val, Gly, Gly, Ala), and occurs five nucleotides downstream from the original SNP. The deletion starts at the second position of the 98th codon however it does not cause a frame shift. The allele frequency of the deletion was 2% in 386 steers and was only observed with the T allele. No homozygous deletion animals were found. A Mann Whitney U test showed an association with the presence or absence of the deletion and ultrasound rib-eye area at end of backgrounding (P= 0.036) and carcass rib-eye area (P=0.028).

Poster 2090

Title: A NOVEL HSP90AA1 PSEUDOGENE IN THE OVINE LATXA BREED

Presenting Author: Ane Marcos-Carcavilla, INIA (Dpto. Mejora Genética Animal), Ctra La Coruña Km 7.5, 28040 Madrid (Spain)

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Abstract:

While analysing the polymorphism of the ovine gene encoding the Hsp90a chaperone (*HSP90AA1*) in more than 600 animals pertaining to 23 different breeds, we found an *HSP90AA1* pseudogene (*HSP90AA1 Ψ*) which was presented only in the 10% of the Spanish Latxa breed sheep analysed. Thus, based on the functional gene mRNA sequence, four primer pairs were designed in order to infer the *HSP90AA1 Ψ* complete sequence. Additionally, the Universal Fast Walking (UFW) strategy developed by Myrick (2005) was employed to elucidate the pseudogene flanking regions with the aim of inferring its location within the ovine genome. As a result, more than 2,5 Kb of the *HSP90AA1 Ψ* sequence has been identified. For the moment, just one variation in the nucleotide sequence has been found between the functional gene and the pseudogene. Furthermore, by comparing the

sequences obtained with the UFW strategy with different databases, preliminary results of the location of this pseudogene have been obtained. Thus, the existence of an *HSP90AA1* pseudogene has been described for the first time on sheep as a recent event occurring exclusively within the Latxa breed. Nevertheless, further analysis should be performed in order to assess its biological significance.

Poster 2091

Title: The quantification of genetic components controlling observed variability to immunisation with a peptide derived from Foot-and-Mouth Disease Virus.

Presenting Author: R. J. Leach., The Roslin Institute, Royal (Dick) School of Veterinary Studies, University of Edinburgh, Roslin Bio centre, Midlothian, EH25 9PS, UK

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Abstract:

Identifying chromosomal regions which control immune responsiveness may suggest new approaches to disease control in livestock. A number of immune traits, including the response to immunisation with a foot-and-mouth disease virus (FMDV) peptide, have been measured in a Charolais-Holstein F2 and backcross cattle population (N = 195). The FMDV peptide comprises two FMDV derived VP1 capsid sequences, which represent the major neutralising antibody epitopes. Considerable variation was observed in the anti FMDV specific IgG1, IgG2 and T cell proliferative responses following immunisation. In order to identify potential genomic regions controlling these traits, a quantitative trait loci (QTL) mapping study was carried out using microsatellite markers covering the bovine genome. Thirty putative QTL were identified with more than half showing over-dominance (heterozygote > homozygote). The majority of the favourable alleles originated from the Charolais breed. This is the first evidence that the immune response to the FMDV peptide is under complex genetic control with multiple genes involved. The results may explain why the peptide only protects a proportion of cattle against viral challenge and why protection does not always correlate with high titres of neutralising antibody. Identifying genes underlying the QTL may help discover novel approaches to improve vaccine efficacy.

Poster 2092

Title: A Snapshot of Copy Number Variation in the Pig Genome

Presenting Author: João Fadista. University of Aarhus, Blichers Allé 20, P.O. Box 20, DK- 8830, Tjele, Denmark

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Abstract:

Recent studies of mammalian genomes have uncovered the extent of copy number variation (CNV) that contributes to phenotypic diversity, including health and disease status. Here we report the first glimpse of CNVs in the pig genome covering part of the chromosomes 4, 7, 14 and 17 already sequenced and assembled. We used a Nimblegen custom tiling oligonucleotide array with a median probe spacing of 409 bp to screen 12 unrelated Duroc boars that are founders of a large family material. After a strict CNV calling pipeline 40 copy number variable regions (CNVRs) covering all the four chromosomes were identified, with some of the CNVRs overlapping segmental duplications and pig unigenes. This CNV snapshot analysis is the first of its kind and constitutes the groundwork for a better understanding of porcine phenotypes and genotypes with the prospect of identifying important economic traits.

Poster 2093

Title: A WHOLE-GENOME ASSOCIATION STUDY OF MAJOR DETERMINANTS FOR PARASITIC INFECTION IN ANGUS BREED.

Marcos Vinicius Silva, Bovine Functional Genomics Laboratory, Agricultural Research Service, USDA Beltsville, MD, 20705, USA

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Abstract:

Fecal egg count (FEC) data recorded for 410 animals between 1992 and 2003 from an Angus herd were transformed using an extension of the Box-Cox transformation to approach normality. DNA for genetic analysis has been acquired for all animals from the resource population and over 70 sires in the

historic pedigree, in total of 595 animals. Genotyping was performed using Illumina's BovineSNP50. For GWA analysis, SNP with a call rate (<93%), departure from HW equilibrium (exact test $p < 0.01$), and minor allele frequency below 5 percent were excluded from the final analysis (26,158 markers retained). All the statistical analyses were carried out with scripts in the R environment and Fortran. Empirical p-values were corrected for genome-wide testing and maximization across genetic models, and a genome-wide significance level of 0.05 (two-sided) and Bonferroni corrected level of 0.01 were used. Five genetic models (codominant, dominant, recessive, overdominant and log-additive) were analyzed. The five most significant SNP were localized within a single linkage disequilibrium (LD) block. These findings provide the first evidence of biomarkers that contribute to early disease detection and primary prevention strategies for parasite infection in cattle and suggest new molecular targets for disease-modifying therapies.

Poster 2094

Title: Characterization of two bovine Y chromosome genes, DDX3Y and HSFY

Presenting Author: Wansheng Liu, Department of Dairy and Animal Science, College of Agricultural Sciences, The Pennsylvania State University, 305 Henning, University Park, PA, 16802, USA

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Abstract:

The *DEAD box polypeptide 3, Y-linked gene* (DDX3Y) encodes a putative ATP-dependent RNA helicase. This gene belongs to the DEAD box protein family, characterized by the conserved motif Asp-Glu-Ala-Asp (DEAD). The *heat shock transcription factor, Y linked gene* (HSFY) belongs to the HSF family, and is characterized by an HSF-type DNA-binding domain. Both DDX3Y and HSFY are candidates for *azoospermia factor* (AZF) and play an important role in spermatogenesis. Deletion of AZF region leads to infertility in otherwise healthy men. Here, we report the molecular characterization of the bovine (b) DDX3Y and HSFY genes. We found two transcripts for bDDX3Y, which correspond to the long and short transcripts of the human DDX3Y. The two bDDX3Y transcripts are identical except for a 3 bp insertion and an expanded 3'UTR in bDDX3Y-L. The bDDX3Y-S encodes a peptide of 660 amino acids (aa), while the bDDX3Y-L encodes a 661 aa. Both

bDDX3Y isoforms contain the conserved motifs of DEAD-box RNA helicases. We identified six transcripts of the bHSFY gene from an adult testis, classified into two groups. Group I contains 3 transcripts with a different size 3'UTR that encode a peptide of 207 aa. Group II contains the remaining 3 transcripts that encode a 417 aa. One of the group II transcripts shows a deletion of 9 bp, resulting in an isoform of 414 aa. All bHSFY isoforms contain the conserved HSF DNA-binding domain. Our preliminary data indicated that multi-copies of the bHSFY gene are present on the bovine Y chromosome. The transcripts of bDDX3Y and bHSFY are predominantly or exclusively expressed in the bovine testis. This project was supported by grants from USDA-CSREES-NRI to WSL (No. 2005-35205-18653).

Poster 2095

Title: Genetic control of swine responses to porcine reproductive and respiratory syndrome virus infection

Joan Lunney, USDA, ARS, BARC, APDL, Building 1040, Room 103, BARC-East, Beltsville, MD 20705 USA

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Abstract:

Our goal is use the Swine Protein-Annotated Oligonucleotide Microarray (www.pigoligoarray.org) to identify immune regulatory and protective pathways to uncover genetic components involved in early immune responses during porcine reproductive and respiratory syndrome virus (PRRSV) infection. Animals were divided into three groups: (1) pigs infected with a virulent PRRSV isolate MNW2B; (2) pigs vaccinated using a contemporary PRRS ATP vaccine (Ingelvac®); and (3) control pigs. Tissues [cranial lung, distal lung, tracheobronchial lymph nodes (TBLN)] were collected between days 3-6 post infection/vaccination. Total RNA was labeled using Alexa Fluor® 555 and 647 dyes (Invitrogen) and a microarray loop design applied to compare gene expression between individuals from all three groups. Validation of the pigoligoarrays was performed by analyzing transcriptional profiles for TBLN, Lung and longissimus dorsi muscle from normal or PRRSV infected pigs as confirmed using QPCR for candidate genes. Diagnostic analyses used designed array control features, negatives and perfect

match/mismatch oligonucleotide sets, to assess non-specific hybridization. Final statistical analyses of the PRRSV study are underway. These studies provide important support for our PRRS Host Genetic Consortium studies of neonatal pig resistance to infection that have just begun. Overall, these studies will reveal immune pathways and candidate genes involved in PRRSV resistance.

Poster 2096

Title: Arachnomelia in cattle: mode of inheritance and initial genetic mapping

Presenting Author: Dr. Johannes Buitkamp, Bavarian State Research Center for Agriculture, Institute of Animal Breeding 85580 Grub/Germany

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Abstract:

Arachnomelia is a congenital disease of cattle. It is characterized by malformed bones of the limbs, back and head. Sporadically, additional findings, like cerebral herniation or hydrocephalus externus and internus, are observed. It causes losses, not only since the calves are not viable, but, more importantly, because sires that carry the mutation are banned from breeding and cows are hurt during delivery. A rapid rise of arachnomelia cases was observed in the years 2005 and 2006 in German and Austrian Simmental cattle. A number of sires with high genetic merit were recognized carriers of the mutation. Therefore, appropriate control measures had to be rapidly developed to avoid further spread of the allele in the population. In a first step breeding management measures were applied. To avoid the final exclusion of important sire lines from breeding, we started the genetic mapping of the disease with the final goal to develop an indirect gene test. Data about the pedigree analysis and population frequency estimation of arachnomelia as well as the genetic mapping are presented.

Poster 2097

Title: Quantitative trait loci for milk-fat composition in Dutch Holstein Friesians

Presenting Author: Anke Schenninkg, Animal Breeding and Genomics Centre, Wageningen University, Wageningen, the Netherlands

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Abstract:

This study is part of the Dutch Milk Genomics Initiative and aims at the characterization of genes involved in milk-fat synthesis and milk-fat metabolism. Previous research has shown substantial genetic variation in milk-fat composition: heritabilities were high (0.42-0.71) for short- and medium-chain fatty acids (C4-C16) and moderate (0.22-0.42) for long-chain fatty acids (C18 and longer). To map QTL affecting milk-fat composition we conducted a whole-genome-scan. The mapping population consisted of 7 half-sib families containing 849 cows in a daughter design. A total of 1379 SNPs were typed covering all 29 autosomes. The phenotypes under study were 56 milk-fat composition traits, including saturated, mono-unsaturated, poly-unsaturated, and conjugated fatty acids, unsaturation indices and fat percentage. A regression interval mapping approach was used to estimate effects and positions of the QTL. QTL for short- and medium-chain fatty acids were detected on BTA6, 14, 19 and 26. QTL for long-chain fatty acids were detected on BTA14, 15, and 16. Our results will enable marker assisted differentiation and marker assisted selection, in order to optimize milk quality and to develop innovative dairy products. Fine mapping of the chromosomal locations is currently in progress.

Poster 2098

Title: Genomic signatures of artificial selection in U.S. Holstein cows

Presenting Author: Tad S. Sonstegard, USDA, ARS, Bovine Functional Genomics Laboratory
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Abstract:

Selection in Holstein cattle has achieved tremendous phenotypic changes over the past 40 years. However, it is unknown how selection has changed the Holstein genome and how those genome signatures of selection are associated with the phenotypic changes. To categorize genome regions either affected or unaffected by selection, we contrasted genome structure data between 148 contemporary Holsteins and 151 cows from an unselected line of Holsteins bred and maintained at the University of Minnesota since 1964. Marker genotypes from 46,231 SNP were analyzed for allele frequency differences and trait associations using Wright's F_{st} test and EPISNPmpi, respectively. Comparison of the most significant marker effects with F_{st} results revealed strong signatures of selection for production traits on chromosomes 1, 2, 3, 7, 8, 11, and 26. Chr 26 also had significant associations (16 markers; $p < 1.0E-12$) with udder traits that co-localize with strong signatures of selection. Of the 31 SNPs with the most significant effects on both milk and daughter pregnancy rate, 29 had opposite effects on these traits providing strong evidence for the antagonistic effect of selection for production on fertility. These findings identify where selection has affected the genome and assist in searching for genes of large effect.

Poster 2100**Title: Global gene expression for cattle selection lines with low and high feed efficiency**

Authors: Yizhou Chen¹, Cedric Gondro², Kim Quinn¹, Robert Herd¹

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Abstract:

Feed efficiency is an economically important trait in beef cattle and it is affected by several physiological systems. The net feed efficiency measured as net feed intake (NFI) is the difference between the actual feed intake over a test period and its expected feed intake based on its size and growth. The objective of this project was to identify differentially expressed genes between animals with high and low NFI and pathways which contribute to the phenotype by global gene expression profiling using a 24K bovine long oligonucleotide array designed by the Bovine Oligonucleotide Microarray Consortium.

Liver tissue biopsies were taken from the top and bottom 30 tested bulls of Angus cattle divergently selected for low or high NFI and 44 animals were chosen for the microarray experiment. The microarray experiment was designed by ranking the animals based on NFI phenotypes and then pairing the top with the bottom animals using dye-swap.

Standard QC measures were performed and arrays were individually normalized using print-tip loess normalization after background correction.

Differentially expressed genes were ranked based on the p-values of a moderated t-statistic after fitting a linear model. The differentially expressed genes were assessed with regard to gene ontology, biological function and known genetic pathways.

Poster 2101**Title: Comparative performance and efficiency of five methods used for genome wide selection in dairy cattle using high density SNP data**

Presenting Author: Gerhard Moser, Co-operative Research Centre for Innovative Dairy Products-CRC ID, Brisbane, Australia

Other authors (name only):

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Abstract:

Three commonly used methods (Least squares regression-LS, Kernel regression-SVM, Bayesian regression- BayesA) and two new approach (Partial least squares-PLS, PLS with SNP selection-PLSjack) were compared in a Genome Wide Selection (GWS) analysis for prediction of genetic merit in dairy cattle. The data included 1945 progeny tested Holstein Friesian sires and genome wide analysis with 7372 SNP. The accuracy (r) of predicting EBV based on SNP genotypes (MBV) was studied by cross-validation. When test subsets were randomly drawn from the data (mirror prediction), accuracies for Australian Profit Rank (APR), an index of 9 component traits, ranged from $r=0.69-0.80$ and $r=0.57-0.68$ for protein%. To assess accuracies of MBV for young animals (forward prediction), the data was partitioned in a training set of 1239 bulls born before 1998, and 5 single year cohorts (1998-2002) of young bull teams. For forward prediction a loss in accuracies of 45% to 75% for APR and 15% to 40% for prot% was observed as compared to mirror prediction. In mirror and forward prediction LS, and Bayes A performed worse than SVM or either PLS method. For GWS, PLS with SNP selection performed the best and most consistent across the 5 year cohorts, and was computationally highly efficient.

Poster 2102

Title: Associations of *Melanocortin 1 Receptor* with growth and carcass traits in beef cattle.

Presenting Author: Kim L. McLean, Department of Animal & Poultry Science, University of Saskatchewan, 51 Campus Drive, Saskatoon, Saskatchewan, Canada

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Abstract:

Melanocortin 1 Receptor (MC1R) is considered to be the main gene controlling production of eumelanin and pheomelanin resulting in black or red coat colour of cattle. The recessive red allele *e*, codes for a nonfunctional receptor which does not bind the agonist alpha-Melanocyte Stimulating Hormone (α -MSH), allowing for production of pheomelanin or red pigment, whereas the dominant E^D allele binds α -MSH leading to the production of eumelanin. We hypothesized that black cattle would have more α -MSH bound to MC1R leaving less circulating α -MSH to bind to the appetite suppressing receptor, Melanocortin 4 Receptor. We genotyped 328 crossbred steers of various colours that were purchased at weaning and fed until slaughter. Red cattle (*e/e*) were found to have a larger rib eye area, shipping weight and hot carcass weight. Black cattle (E^D/E^D and E^D/e) had increased back fat and required significantly fewer days on feed to reach a target fat level for slaughter than the red cattle. By sorting cattle on a coat colour basis, 21 days on feed were eliminated for black cattle ($P < 0.001$). As many as 33 days on feed were avoided by genotyping and sorting cattle based on the number of E^D alleles ($P = 0.001$).

Poster 2103

Title: Identification of polymorphisms influencing production and functional traits on bovine chromosomes 19 and 29 in Canadian Holstein bulls

Presenting Author: Aparna Prasad, Department of Agricultural, Food and Nutritional Science, University of Alberta, Edmonton, Alberta, Canada, T6G 2P5

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Abstract:

Detection of polymorphisms influencing economically important traits is a valuable tool for marker-assisted selection (MAS). The objective of the present study was to perform a chromosome-wide scan to detect polymorphisms associated with production and functional traits on bovine chromosomes 19 (BTA19) and 29 (BTA29) in Canadian Holstein bulls ($n=322$). In total, 505 and 220 single nucleotide polymorphism (SNP) markers on BTA19 and 29, respectively were genotyped. The average resolution of markers on BTA19 and 29 was 1 locus/172 Kb and 1 locus/228 Kb. Production traits studied were milk yield, fat yield, fat%, protein yield, and protein% and functional traits studied were herd life, daughter fertility, milking speed, milking temperament, calving ease and maternal caring ease. Single locus linkage disequilibrium regression model was used to test the association between SNPs and the different traits. In total, 133 SNPs were found to be significantly associated ($P < 0.01$) with these traits. Forty six of them were located in the intron, one in the untranslated region, and three in the coding-synonymous region of the gene, while eighty three of them were not located within any genes. Once confirmed in an independent cattle population, these associations can be used in MAS schemes.

Poster 2104

Title: Multiplex assay for simultaneous identification of paternity and molecular markers of economic interest in bovines

Presenting Author: KATIA TORRES DE SOUZA, katia@linhagen.com.br

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Abstract:

Following the world-wide trends, Brazil has invested in the development of more efficient mechanisms of lineage records and genetic improvement of bovines. In the last few decades, the lineage records of bovines have classified an individual animal and decided parentage questions only by using serological typing. This methodology presents limitations and recently has been replaced for DNA markers. LINHAGEN Biotecnologia has developed its own multiplex assay for bovine genetic identification, which includes, besides the 9 (nine) microsatellite markers recommended by ISAG (International Society of Animal Genetics), another 3 (three) extra markers which provides greater accuracy for the test itself.

Now we improved this assay even more by adding another markers to also analyse genetic characteristics of economic interest, such as quality and yield grade and/or the tendency for diseases, and use it for improvement genetic programs. The association of analysis between molecular markers and genetic identification in one test only is new in our country, will represent a significant advance and can provides: 1) bigger knowledge on the genetic characteristics of the national flock and dissemination of the use of molecular markers, hence that the paternity test tends to be mandatory and , consequently, will be carried through on a large scale; 2) A large scale economy for the laboratories and breeders, because this assay can provides several analysis simultaneously.

Poster 2105

Title: Prion gene haplotypes of Thai indigenous cattle

Presenting Author: Kalaya BOONYANUWAT¹
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Abstract:

Bovine spongiform encephalopathy (BSE) is a fatal neurological disorder characterized by abnormal deposits of a protease-resistant isoform of the prion protein. *PRNP* haplotype associated with BSE. The objective of this study was to identify polymorphisms and haplotypes of *PRNP* gene from octapeptides of the exon 3 region and promoter region through the 3'UTR in a sample of Thai indigenous cattle and compared with network haplotype to interplete BSE resistance and susceptibility. Four hundred and thirty-four individual DNA samples of Thai indigenous cattle were analysed for *PRNP* haplotype. Sequence analysed of octapeptide in exon 3 region identified 3 haplotypes of *PRNP* gene which had 1, 3, and 4 repeats of octapeptide. These polymorphisms define *PRNP* haplotypes that may influence BSE resistance in Thai indigenous cattle.

Poster 2106

Title: Genomic structure and genetic variants of leptin receptor (LEPR) gene in pig

Presenting Author: Kyung-Tai Lee / National Institute of Animal Science, RDA, Suwon, Korea..

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Abstract:

Leptin receptor (LEPR) gene is still considered as a positional candidate gene controlling QTL for fatness on chromosome 6 in pig due to its position and physiological role. Four BAC clones were screened by using LEPR-CA microsatellite marker and a segment of 324 kb was sequenced by shotgun strategy. We could define the genomic structure of LEPR gene in pig by comparing the genomic sequence of 324kb with LEPR transcripts found by RACE method. Porcine LEPR gene spanned 189,209bp and exon 1 and 2 were newly found and different from human and other species. Repetitive sequences were estimated to include 45.21% of the contig sequence, of which LINE and SINE were classified as 19.68 and 19.07%. The average G+C content and the number of CpG islands in the contig sequence were estimated to be 40.39% and 17, respectively. We investigated genetic variants such as SNPs within 5' upstream region including exon 1 of LEPR gene by using direct sequencing of 200 pigs from five. Association study for the genetic variants and methylation status on the CpG islands of regulatory region may produce a clue to understand relationship between the expression of LEPR gene and fatness in pig.

Poster 2107

Title: Sequencing-ready physical map of the horse Y chromosome

Presenting Author: ...**Terje Raudsepp**, Department of Veterinary Integrated Biosciences, College of Veterinary Medicine, Texas A & M University, College Station TX 77843, USA

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Abstract:

Horse Y chromosome (ECAY) project aims to identify all Y-linked genes and generate a detailed physical map for the chromosome. ECAY consists of a pseudoautosomal (PAR), male specific (MSY) and a heterochromatic (HC) region. We have constructed a BAC contig map spanning the entire PAR, MSY and a

part of HC. The map consists of 352 BAC clones which are arranged into 4 contigs by STS content analysis of 391 STSs and 52 genes/ESTs. The map demarcates the pseudoautosomal boundary and shows a segmental duplication between PAR and MSY. MSY is divided into single-copy and ampliconic regions. The latter contain multiple copies of known genes (*TSPY*, *CULABY*, *UBE1Y*) and several novel ESTs. Single-copy equine *SRY* is embedded in the middle of ampliconic sequences. Unique to ECAY is the discovery of ESTs in HC. The size of ECAY euchromatin is estimated to be ~13 Mb and the three gaps count for less than 2 Mb. The contig map of the ECAY euchromatin with 71 BACs forming the minimum tiling path is ready for sequencing. Complete information about the organization and function of ECAY is needed to understand the Y-linked component of stallion fertility and study sex chromosome evolution in mammals.

Poster 2108

Title: Fine mapping of QTL for left-sided displaced abomasum in German Holstein cattle

Presenting Author: Stefanie Mömke, Bünteweg 17P, 30559 Hannover

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1. Henner Scholz
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Abstract:

The displacement of the abomasum is a common disease in dairy cattle. Affected cows often have to be treated surgically and milk performance, as well as productive life are significantly reduced. For this study, samples of 360 animals belonging to 14 half-sib families were analysed. These 14 half-sib families comprised 328 LDA affected cows and 24 unaffected control cows. Five QTL on the bovine chromosomes (BTA) 1, 3, 21, 23, and 24 for left-sided displaced abomasum (LDA) in German Holstein cattle were fine-mapped, using a total of 92 already published or newly developed microsatellite and SNP markers. The peaks of these QTL were located at the markers *MNB152* (BTA1, 54.6 cM, Zmean: 3.18), *DIK4922* (BTA3, 5.9 cM, Zmean: 3.04), *BMS743* (BTA21, 75.3 cM, Zmean: 1.96), *IOBT528* (BTA23, 7.9 cM, Zmean: 2.84), and *DIK4984* (BTA24, 75.2 cM, Zmean: 1.94), respectively. The QTL on BTA1 and 3 reach genome-wide significance, while the QTL on BTA21, 23, and 24 are chromosome-wide significantly linked with LDA.

Further fine-mapping will be provided by a SNP chip analysis with following association studies.

Poster 2109

Title: Phylogenetic analysis and genetic variation of population of genus *Lepus* in Europe.

Authors:

Presenting Author: María J. Sanz-Martín. Genetics, Physical Anthropology and Animal Physiology Department. Faculty of Science and Technology. University of the Basque County.

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Abstract:

Three out of five hare species of genus *Lepus* in Europe coexist in the Iberian Peninsula, *L. europaeus*, *L. granatensis* and *L. castroviejoi*, and their genetic relationship is uncertain. This work assesses two aims: 1) The phylogenetic relationship of the Iberian hare in a European context 2) The development of an exhaustive genetic map of the Iberian *L. europaeus* with conservation purposes. To achieve these aims, 295 individuals from all five European species were analysed. A fragment of 400 bp from mtDNA's control region was sequenced and 17 microsatellite loci were genotyped. Results prove that *L. granatensis* is more closely related to the European lineage than to the African-Asian one. On the other hand, previous mtDNA studies have shown that the autochthonous populations of Iberian *L. europaeus*, contrary to the rest of *L. europaeus* in Europe, are characterized by an ancient introgression by *L. timidus*. Our results confirm this fact, although we also observed non-introgressed individuals in the northern Iberian Peninsula, likely due to non-controlled restocking from other European regions.

Poster 2110

Title: *DGATI*: a diet-regulated quantitative trait locus that affects hepatic lipid deposition

Presenting Author: Rosane Oliveira, Department of Animal Sciences, University of Illinois, 1201 W Gregory Dr, Urbana IL, 61821

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Abstract:

A lysine to alanine substitution at codon 232 in the Acyl-CoA:diacylglycerol acyltransferase1 gene (*DGATI* K232A) underpins a quantitative trait locus (QTL) responsible for differences in milk and fat yields in dairy cows. In the present study, dietary energy intake and *DGATI* genotype were evaluated for their effects on transcript levels of hepatic *DGATI* and accumulation of triacylglycerol (TAG) and total lipid in liver. Thirty-five cows fed energy *ad libitum* or restrictively during the prepartum period were genotyped for the *DGATI* K232A mutation. *DGATI* mRNA levels, TAG, and total lipid were determined in biopsied liver samples collected at -14, +1 and +14 days relative to parturition. TAG and lipid concentrations were significantly higher at day +14 in *ad libitum*-fed cows having one or two copies of the *DGATI*^K allele. *DGATI* genotype had a significant effect on *DGATI* mRNA levels, and an effect of diet on *DGATI* mRNA levels was found at one day postpartum. *DGATI* alleles appear to be responsible for liver TAG and lipid accumulation postpartum in animals overfed during the prepartum period. Nutritional management of cows having the *DGATI*^K allele can thus be used to reduce economic losses due to metabolic disorders occurring as a result of overfeeding energy prepartum.

Poster 2111

Title: Transcriptional profiling and biochemical pathways analysis of liver from two breeds of cattle.

Presenting Author: Pawel Lisowski, Department of Molecular Biology, Institute of Genetics and Animal Breeding, Polish Academy of Sciences (IGAB PAS), 05-552 Jastrzebiec, Poland

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Abstract:

We investigated the gene expression profile in the liver of cattle breeds distinctly different in meat and milk production capacity, carcass and meat quality. Studies are based on the assumption that animals differing in meat and milk performance must show different expression profiles of genes that determine these traits. The study was performed in liver tissues from bulls with two different genetic backgrounds, meat and dairy - Hereford and Holstein-Friesian. The animals were at 6 months of age during the intensive growth and muscle development. The 8-K bovine

oligo microarrays were used (BLO, Michigan State University). Microarray analyses identified 34 genes of which the expression levels differed at least 2-fold. These genes are involved in 97 biological processes. Hereford animals showed higher expression of L-FAB, GH receptor precursor, STAT1, and STAT3 genes, while in HF animals had higher expression of PKC and ATP synthase gamma chain genes. Combining the microarray results and biological databases information we choose the genes engaged in 24 biochemical pathways for further analysis. These pathways were related to cellular processes, energy metabolism and signal transduction. Our findings provide a description of molecular events accompanying individual development that may underlay the phenotype differences between two cattle breeds.

Poster 2112

Title: Genetic analysis of local Vietnamese chickens for conservation purposes

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Abstract:

The Vietnamese H'Mong chicken breed called the Black Chicken because of the black bones, skin and shank, had a strong cultural value. In order to characterise the H'mong chicken from the originating province, namely Ha-Giang, phenotypic and genetic analyses were performed. Thirty communes from the 11 districts of the province were surveyed. A total of 2487 animals were described for 29 phenotypic characters and analysed by Principal Component Analysis (PCA). The two principal components explained 53.6 % of the inertia. All variables describing black colour grouped together and allowed differentiation of three communes among the thirty. From the described animals set, 1082 animals were genotyped using 18 ISAG microsatellite markers. In the Ha-Giang population, H_{EXP} averaged 0.619 and F_{IS} averaged 0.121 and 3.7 % of the genetic diversity was due to differentiation between communes. Bayesian approach did not showed genetic subdivision into subpopulations. In summary, analysis of phenotypes revealed a geographic distribution of Black chickens while the bayesian admixture analysis within the

province did not showed genetic differentiation of the Black chicken within the province. Therefore, a reproductive pool of chicken considering only the black phenotype could be used if government wants to create a genetically distinct H'mong breed.

Poster 2113

Title: Individual assignments to dog breeds using simulated and real data

Presenting Author: Grégoire Leroy
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Abstract:

Dog breeds constitute clearly genetically differentiated populations. Thus, breed assignment may represent an interesting tool for clubs and breeders in order to investigate if a dog without genealogy can be assigned to a particular breed. Several Bayesian approaches have been developed during last years, using direct assignment tests, or simulating populations to exclude animals from the populations tested. In a first part, we tested the different approaches on dog breeds by using 21 microsatellite markers. The assignment of 1240 individuals belonging to 63 breeds was tested on a reference set of 2000 dogs of 9 breeds. Using simulation procedures, results showed that all individuals belonging to the same breeds of the reference set were correctly assigned. However, 14% of the animals of other breeds were incorrectly assigned in the 9 breeds at 1% threshold. In a second part, populations were simulated with a genetic structure more or less similar to dog breeds. Our results showed that, as expected, allelic richness constitutes a parameter that influences assignment effectiveness. However heterozygosity (*H*) by itself has some influence on the results: breeds with a high *H* level affect incorrectly a larger number of animals when population simulation procedures are used. Effects of other parameters, such as sample size were also tested.

Poster 2114

Title: Molecular phylogenetics of mtDNA D-loop sequences in Chinese buffalo: implication for origins of domestic buffalo (*Bubalus bubalis*)

Presenting Author: Yi Zhang, Department of Animal Genetics & Breeding, College of Animal Science and Technology, China Agricultural University, Beijing, China, 100094

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Abstract:

In this study, to characterize the genetic origin of Chinese swamp buffalo and their phylogenetic relationships with river buffalo and Southeast Asian swamp buffalo, we determined the whole mtDNA D-loop sequences of 100 domestic buffalo geographically covering their main distribution in China and analyzed these sequences in comparison with published data. Neighbor-joining phylogenetic tree revealed two highly divergent mtDNA lineages (A and B) in Chinese swamp buffalo, in which A was predominant (77%) and B could further subdivided into B1 (11%) and B2 (12%). These lineage groups all were demonstrated to have undergone recent population expansion. The divergence estimation between A and B, based on cytochrome *b* gene sequences, is approximately 650,000 years ago. Moreover, both A and B were found in all Chinese swamp populations with similar proportions (proportion of B = 0.16 ± 0.07). These findings suggest that Chinese buffalo might be derived from a single wild source population. In addition, a combined media-joining network analysis provided evidence that swamp and river buffalo originated from two independent domestication events in China and Indian subcontinent, respectively. The buffalo in South-east Asia, however, have both ancestries, indicating possible genetic mixture of two buffalo types in this region.

Poster 2115

Title: Investigation of the KIT gene in Nero Siciliano pigs with different coat colour phenotypes

Presenting Author: **Enrico D'Alessandro**,
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Abstract:

The Dominant White locus in pigs is caused by mutations in the *KIT* gene that have been described as

copy number variation and single nucleotide polymorphisms. Nero Siciliano (Black Sicilian) is a local Italian pig breed reared in the Nebrodi mountains of Sicily. The animals are usually completely black but a few show white portions mainly in the face or in the fore legs. We genotyped 97 Nero Siciliano pigs (25 with white portions) for the splice site mutation of the intron 17 of the *KIT* gene that is associated with the Dominant White *I* allele. These animals were also analysed for the presence of the duplication of the same gene. None of the pigs carried the splice site mutation while 7 out of 25 animals with white portions were positive for the duplication test. Then, 20 of the 21 exons of the *KIT* gene (including intronic regions) were sequenced in eight Nero Siciliano pigs with different colour patterns and in one Large White animal. Several new mutations were identified in Nero Siciliano pigs. The results suggest that the presence of white portions in Nero Siciliano pigs might be only in part due to mutations in the *KIT* gene.

Poster 2116

Title: Single Nucleotide Polymorphism (SNP) density and predicted mapping resolution of complex trait genes in the Thoroughbred horse

Presenting Author: S.C. Blott, Animal Health Trust, Lanwades Park, Kentford, Newmarket, Suffolk, CB8 7UU, UK

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Abstract:

The recent sequencing of the equine genome and associated single nucleotide polymorphism (SNP) discovery makes genome-wide association studies possible in horses. The required marker density for an effective whole genome scan depends on the extent of linkage disequilibrium across the genome. We have re-sequenced 52 SNPs across a 1 Mb region on ECA 26 in order to assess the extent of fine-scale linkage disequilibrium in the Thoroughbred horse. The median spacing between SNPs is 200 bp, maximum 80 kb and minimum 1 bp. The 52 SNPs have been re-sequenced in 24 unrelated Thoroughbreds and pairwise estimates of D' and r^2 between SNPs have been calculated. This allows the extent of linkage disequilibrium for genetic distances between 1 bp to 1 Mb to be assessed. Haplotype phases have been determined for each individual horse and the size of haplotype blocks and number of tagging SNPs per block evaluated. The precision and power of mapping, given the observed patterns of linkage disequilibrium, has been explored using simulation.

The information is useful in determining the SNP density required for whole genome association scans and for follow-up fine mapping in the Thoroughbred.

Poster 2117

Title: MUTATIONS IN TWO GENES ARE ASSOCIATED WITH INCREASED OVULATION RATE AND LITTER SIZE IN PROGENY OF LACAUNE MEAT SHEEP

Presenting Author: Drouilhet L. INRA Laboratoire de Génétique Cellulaire, 31326 Castanet-Tolosan, France

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Abstract:

A new autosomal major gene affecting ovulation has been evidenced by a statistical approach in the *Lacaune* population. A full genome scan localized this autosomal prolificacy gene (*FecL*) on sheep chromosome 11. Further investigations allowed the reduction of the localization interval between markers *BM17132* and *FAM117A*, corresponding to one synteny block of human chromosome 17 of 1.1 megabases, containing 20 genes. This fine mapping is still going on. The expression of those 20 genes was tested by qPCR and no difference was found in homozygous mutants versus wild type, in four tissues (granulosa and theca cells, hypothalamus and pituitary gland). A particular haplotype was associated with the *FecL* mutation, which allowed marker associated classification of animals as carriers or non-carriers of the mutation. In addition, ewes with extreme ovulation rate have been isolated in the *Lacaune* population and it was hypothesized that another mutation was segregating in the population. A new mutation has been evidenced in the *Bmp15* coding sequence (chromosome X). This mutation is a non conservative substitution (C53Y) that prevents the processing of the protein. Synergic action of the two mutations (in *FecL* and *Bmp15* genes) on both ovulation rate and litter size was demonstrated.

Poster 2118

Title: Mitochondrial DNA sequence variation in three Sicilian autochthonous horse breeds

Presenting Author: DONATA MARLETTA, DACPA - Sezione di Scienze delle Produzioni Animali. Università degli studi di Catania. Via Valdisavoia, 5 – 95123 Catania. Italy

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8. Giuseppe D'Urso.

Abstract:

Matrilineal genetic diversity in Sicilian horses was investigated sequencing the mitochondrial D-loop hypervariable region (397 bp) in Sanfratellano, Sicilian Indigenous and in Sicilian Oriental Purebred, 20 mares per breed. Twenty different haplotypes, with 30 polymorphic sites, were identified: 11 in Sanfratellano, 13 in Sicilian Indigenous, 1 in Sicilian Oriental Purebred.

Genetic distances between haplotypes were estimated using Kimura two-parameters (MEGA 4); phylogenetic tree was constructed using neighbour-joining method. Nucleotide diversity (μ), estimated by Arlequin 3.1, showed that Sicilian Indigenous was the most diverse population ($\mu = 0,018 \pm 0,009$) followed by Sanfratellano ($0,916 \pm 0,041$); haplotype diversity stated the same trend. BLAST search showed that all the sequences overlapped with many of the GeneBank database haplotypes, but for 3.

Phylogenetic analysis did not show monophyletic groups for the Sicilian breeds, confirming the multiple origins for the maternal lineages of the domestic horse breeds. The Sicilian Oriental Purebred haplotype, found in 5 Sanfratellano mares, denotes a mixture of these two genetic types. Some haplotypes, identified in Sanfratellano and Sicilian Indigenous, suggest a common origin between these two breeds; furthermore three haplotypes, not shared with other breeds, indicate a possible geographic segregation of these two autochthonous horses.

Supported by ISZS project 14, 03/05/2007

Poster 2119

Title Microarray analysis of skeletal muscle genes in pigs divergent for glycolytic potential.

Presenting Author: Roberta Davoli, Dipartimento di Protezione e Valorizzazione Agroalimentare (DIPROVAL), Sezione di Allevamenti Zootecnici, University of Bologna, 42100 Reggio Emilia, Italy

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Abstract:

Glycolytic potential (GP) is an indicator of the glycogen level in skeletal muscle and this parameter may have a marked effect on technological pork quality. The aim of this research was to identify pig genes involved in the regulation of GP in muscle by microarray analysis. Samples of semimembranosus muscle from eight pigs with extreme and divergent GP were selected among 277 sib-tested Italian Large White (ILW) animals and utilised for RNA extraction. A RNA pool with low GP (4 samples presenting GP < 65 μmol lactate equivalent g^{-1} muscle wet weight) and another RNA pool with high GP (4 samples with GP > 145 μmol) were obtained. The Cy3 and Cy5 labelled cDNAs were used to hybridise the 11K Pig Oligo set version 1.0 (Operon-Qiagen). Differentially expressed genes were identified by ANOVA using GLM procedure in SAS. The analyses showed that several genes up-regulated in the pool with low GP were involved in energy metabolism and only few genes coding for transcription factors, glycolytic pathway and cell metabolism were detected in the pool with high GP. Differential gene expression detected by microarray was confirmed for selected genes by quantitative real time on the individual RNA samples of the two pools.

Poster 2120

Title: New mutations in *BMP15* and *GDF9* genes are not associated with increased prolificacy in Rasa aragonesa sheep breed.

Presenting Author: Albert Martinez-Royo & Unidad de Tecnología en Producción Animal, CITA, Avda de Montaña 930, 50059 Zaragoza. Spain

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Abstract:

Recently, a new naturally occurring mutation in *BMP15* gene in the ovine Rasa aragonesa breed has been described (*FecX^R* allele). It has been associated with both increased prolificacy and sterility in heterozygous and homozygous ewes, respectively. Animals from controlled nucleus with high EBVs for prolificacy without the *FecX^R* allele have been analysed for *BMP15* and *GDF9* genes by SSCP technique. Both genes are members of the transforming growth factor beta (TGF β) superfamily

and play an essential role in mammalian fertility. Animals with different profiles were sequenced and the mutations were studied. A total number of 3 and 4 mutations have been found in *BMP15* and *GDF9*, respectively. The first of these is a leucine deletion in the predicted signal sequence of *BMP15* without any phenotypic effect. Four of the seven polymorphisms are nucleotide changes that do not result in an altered amino acid. The two remaining nucleotide changes occurs in *GDF9* and produce an amino acid change. Both occurs at a position before the furin cleavage site for the mature peptide, so are unlikely to affect the activity of the mature protein.

Poster 2121

Title: A genome scan for QTL affecting milk somatic cell count in the Italian Holstein Friesian cattle breed by selective milk DNA pooling in a daughter design

Presenting Author: **Luca Fontanesi**, DIPROVAL, Sezione di Allevamenti Zootecnici, University of Bologna, Via F.lli Rosselli 107, 42100 Reggio Emilia, Italy

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9. Vincenzo Russo

Abstract:

In dairy cattle, mastitis is the most important health problem causing substantial economic losses. Milk somatic cell count (SCC) is highly correlated with mastitis and hence is used as a surrogate trait for breeding objective. A selective milk DNA pooling strategy in a daughter design was used in the Italian Holstein-Friesian cattle breed to map QTL for milk SCC as a first step towards the application of MAS for this trait. Milk DNA pools from about 200 daughters with high and 200 with low estimated breeding value (EBV) for SCC were constructed for each of six sires with at least 3500 milking daughters each. Sires were genotyped for a panel of 133 dinucleotide microsatellites distributed over all bovine autosomes. DNA pools were genotyped for the markers that were heterozygotes in the sires. Shadow corrected estimates of sire allele frequencies in the pools were compared between high and low pools. Proportion of false positives (FPF) was applied to obtain experimentwise significant levels. Allele substitution effects were calculated for all significant sire-marker- trait

combinations. Several QTL were identified and their location was compared to the QTL already identified in the same population for milk yield and milk protein percentage.

Poster 2122

Title: Effects of 52 SNPs on growth rate, meat content and selection index in Large White and Landrace boars

Presenting Author: Stanislaw Kaminski, University of Warmia and Mazury, 10-718 Olsztyn, Poland, email: stachel@uwm.edu.pl.

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Abstract:

306 boars (108 Large White and 198 Landrace) were genotyped for 52 SNPs to find out which of them or their combinations influence growth rate (GR), meat content (MC) and selection index (SI). The effects of SNPs were estimated by using a mixed linear model including random animal effect, fixed effects of SNPs including additive, dominance and pairwise additive-by-additive epistases, year*season of birth, breed and RYR1 genotype. In order to estimate all possible SNP combinations without overparameterising a model a stochastic approach was adopted. 1900 repeats of the model were generated, each containing 5 randomly chosen SNPs. The final estimates of the fixed effects of the model equaled an average out of the replications. The hypothesis of a nonzero effect of SNP was tested by the t test. Among 4300 estimates calculated, many significant ($p < 0.01$) but minor effects (below 1% of a trait mean) were recorded. Outstanding effects (higher than 1 SD) were found in 17, 17 and 10 SNP pairs, for GR, MC and SI, respectively. Loci most frequently involved in epistasis were: H-FABP, MEF2D, LDLRRP1, LDHA, ESR2, GAD2, PRKAG3. The selected SNPs will be further investigated to find out which ones may be applied in MAS.

Poster 2123

Title: Copy number variation and missense mutations in the caprine agouti signaling protein (ASIP) gene are present in goat breeds with different coat colour

Presenting Author: **Stefania Dall'Olio**, DIPROVAL, Sezione di Allevamenti Zootecnici, University of Bologna, Via F.lli Rosselli, 107, 42100 Reggio Emilia, Italy

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7. Vincenzo Russo

Abstract:

The *Agouti* locus encodes the agouti signaling protein (ASIP) that plays a key role in the pigment-type switching from eumelanins to pheomelanins. In goats, the effect of the *Agouti* locus on coat colour and colour pattern distribution has been deduced analysing herd book records and segregation data in classical crossbreeding experiments. Here, we sequenced the coding region of the goat *ASIP* gene in six breeds (Camosciata, Derivata di Siria, Girgentana, Maltese, Murciano-Granadina and Saanen) showing different coat colours and patterns. Five single nucleotide polymorphisms were identified, three of which were missense mutations within exon 4, causing amino acid substitutions in conserved positions of the carboxy-terminal domain of the protein. Allele and genotype frequencies suggested that the missense mutations do not affect coat colour in the investigated goat breeds. Genotyping results and deviation from Hardy-Weinberg equilibrium, as well as allele copy number estimation from sequencing data indicated the presence of copy number variation (CNV) for this gene with high frequency in the Girgentana and Saanen breeds. The obtained results, together with the data already reported by others for the ovine *ASIP* gene, provide evidence for a recurrent interspecies CNV at this locus.

Poster 2124

Title: Determination of protein and the RNA expression levels of the commonly used housekeeping genes in a neurodegeneration mouse model

Presenting Author: Ana C. Calvo, LAGENBIO. University of Zaragoza. C/ Miguel Servet 177, 50013 Zaragoza, Spain

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Abstract:

The choice of housekeeping proteins or genes for internal standards should be made carefully, taking into account the cell and tissue type, the experimental conditions, and the disease state(s) under consideration. GAPDH, β -actin, and β -tubulin are generally considered housekeeping genes and are commonly used as internal standards in a broad range of studies examining levels of gene transcription. We are interested in studying changes in protein and gene expression that may shed light on a neurodegenerative disease mouse model, SOD1^{G93A} mice, that bear a G93A mutation in the human gene for superoxide dismutase 1 (SOD1) and therefore results in motor neuron pathologies. As such, we first wanted to assess whether β -actin, GAPDH and β -tubulin could be used as internal protein controls in spinal cord, brain, and skeletal muscle tissues. We next examined the variability of β -actin and GAPDH transcriptional levels, in the same experimental conditions. The results suggest the possibility that alterations in the integrity of microtubules may be early events of the neurodegenerative processes that may finally reflect later alterations in GAPDH metabolism, in terms of altered protein levels that potentially occur in skeletal muscle, one of the most affected tissues.

Poster 2125

Title: Polymorphism and Genomic Organization of the Equine MHC Class II Region

Presenting Author: Donald Miller, Baker Institute, Cornell University, Ithaca, NY, 14853 USA

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1. Douglas F. Antczak.

Abstract:

Studies of the organization and gene content of the Equine Major Histocompatibility Complex (MHC), also known as the Equine Leukocyte Antigen (ELA) region, have benefited in recent years from the construction of the equine Bacterial Artificial Chromosome (BAC) library, and the whole genome sequence (WGS) of the horse. These tools were derived from DNA of two closely related Thoroughbreds, each homozygous for the ELA-A3 haplotype, and members of the research herd at Cornell University. The BAC library provided a framework for the genomic organization of ELA, and provided sequence data that has enabled us to study gene content and polymorphism in other ELA haplotypes. The WGS has advanced these studies by confirming earlier sequence data and identifying gene loci that were previously undetected. In this study we examine nine DQ and DR genes located in the ELA Class II region, and compare the gene alleles of the ELA-A3 haplotype with those of the ELA-A2, -A5, -

A9, and –A10 haplotypes. Our findings reveal a higher level of polymorphism in the beta genes compared to the alpha genes, and three of the five beta gene loci studied have unique alleles for each of the five ELA haplotypes.

Poster 2126

Title: Parentage testing in alpacas (*Vicugna pacos*) using semi-automated fluorescent multiplex PCRs with 10 microsatellite markers

Presenting Author: Jose R. Espinoza. Laboratories for Research & Development (LID), Universidad Peruana Cayetano Heredia, Av. Honorio Delgado 430, Lima 31, Peru.

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Abstract:

A microsatellite multiplex system for parentage determination in alpacas was evaluated in a population of 329 unrelated alpacas from different geographical zones in Peru. All microsatellite markers (n=10), which amplified in two multiplex reactions, were highly polymorphic with a mean of 14.5 alleles per locus (six to 28 alleles per locus) and an average expected heterozygosity (HE) of 0.8185 (range of 0.698–0.946). The total parentage exclusion probability was 0.999456 for excluding a candidate parent from parentage of an arbitrary offspring, given only the genotype of the offspring, and 0.999991 for excluding a candidate parent from parentage of an arbitrary offspring, given the genotype of the offspring and the other parent. In a case test of parentage assignment, the microsatellite panel assigned 105 (from 105 cases) offspring parentage to 15 sires with LOD scores ranging from $2.19 \cdot 10^{+13}$ to $1.34 \cdot 10^{+15}$ and Δ values ranging from $2.80 \cdot 10^{+12}$ to $1.34 \cdot 10^{+15}$ with an estimated pedigree error rate of 13.2%. The performance of this multiplex panel of markers suggests that it will be useful in parentage testing of alpacas.

Poster 2127

Title: QTL for maternal ability traits in an Iberian x Meishan pig cross.

Presenting Author: Cristina Óvilo, Dpto Mejora Genética Animal, INIA, Ctra Coruña Km 7.5, 28040 Madrid, Spain

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10. Carmen Rodríguez

Abstract:

The aim of this study was to investigate chromosomal regions affecting maternal ability traits: litter weight at farrowing (LWF) and at weaning (LWW). For this purpose an F₂ experimental cross between Iberian and Meishan porcine breeds was used. The genomic scan was carried out using 134 markers (microsatellites and SNPs in several genes) covering the 18 porcine autosomes, with 670 and 615 records for LWF and LWW, respectively. Analyses were performed using a repeatability animal model to estimate QTL and additive polygenic effects. For LWF the results showed a genome-wide highly significant QTL (p<0.1%) located on SSC12 (45cM), not reported previously. This QTL present both additive (0.49±0.14) and dominant (0.56±0.20) effects, and coincides with the position of growth hormone gene (*GH*). A search of mutations was conducted on this gene for its evaluation as candidate for the QTL. Three missense polymorphisms were genotyped, resulting on three haplotypes segregating in the F₂ population. Results of a marker assisted association analysis showed significant association of *GH* polymorphisms, with a favorable effect of the Iberian allele on LWF (p=0.13x10⁻²). For LWW a genome-wide significant QTL (p<0.1%) was detected on SSC6, at 78 cM. This QTL shows additive effect (2.02±0.49).

Poster 2128

Title: Construction of chicken 50K SNP panel and its application in broiler breeding

Presenting Author: Tun-Ping Yu, DNA LandMarks Inc., St.-Jean-sur-Richelieu, Québec, Canada

Other authors (name only):

1. Ron Okimoto
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Abstract:

Cobb-Vantress, Inc. (Cobb) is the world leader in developing broiler breeding stock. Through collaboration with DNA LandMarks (DLM), the Center of Excellence for Genomics of BASF Plant Sciences, we have established a large scale chicken SNP panel for genomic research that enhances genetic progress in economically important traits. With custom-developed bioinformatic tools, a panel of over 60,000 SNPs was identified. Approximately 50,000 SNPs were converted to an Affymetrix custom panel. The concordance rate of SNP genotyping was over 99%. Among the 50K SNPs, 95% were confirmed to be true SNPs and showed segregation in Cobb lines. A highly informative SNP panel evenly covering the chicken genome for each of the Cobb pedigree lines was obtained through a stringent SNP filtering system according to scores of marker informativeness. Linkage Disequilibrium (LD) analysis was performed using Composite Linkage Disequilibrium analysis to gain better understanding of the genetic structure of Cobb pedigree lines. LD results provided key information for experimental design for a whole genome association analysis. Pedigree-based haplotype analysis and decremental search function were used to identify tagged-SNPs within the LD regions. The pedigree line-specific SNP panels are used to study production and animal welfare traits for application in marker assisted selection.

Poster 2129**Title: Gene expression-phenotype associations using eQTL based gene sets.**

Presenting Author: L. Janss, Aarhus University, Denmark.

Other authors (name only):

1. V. Obolonkin

Abstract:

In gene expression analysis, gene sets can be used to improve power, biological underpinning and consistency between experiments. Commonly, gene sets are based on bioinformatics data such as pathways or the Gene Ontology. In this study we present a gene expression analysis using gene sets based on eQTL locations for the gene expressions. This can be seen as a “data driven” gene set assignment which can fill gaps in existing knowledge about co-regulated genes. For this analysis, firstly, individual gene expressions were tested for association with the phenotype and a loose selection of primary candidates was made. Subsequently for all these candidates, an eQTL mapping was performed using a Bayesian multi-QTL mapping procedure and the selected expressions were grouped in gene sets according to common eQTL position. In the final stage the so determined gene sets

were tested again for association with the phenotype. This procedure identified 3 main gene sets each explaining ~8% of variation, and a handful of gene sets each explaining ~4-6% of variation in the phenotype. Being based on common eQTL position, a genomic location for the putative “master regulator gene” for each gene set is available as well.

Poster 2130**Title: CFA9 association study to identify potential modifier loci for progressive rod-cone degeneration (*prcd*) in the dog**

Presenting Author: Barbara Zangerl, University of Pennsylvania, School of Veterinary Medicine, 3900 Delancey Street, Philadelphia, PA, 19104

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Abstract:

A single mutation in the *PRCD* gene, causing progressive rod-cone degeneration (*prcd*) in the dog, was found to be shared among 27 breeds. The disorder causes retinal degeneration, always resulting in complete blindness; the time of onset and disease progression, although generally breed specific, varies between breeds. Previous breeding experiments established the stable segregation of these variants linked to the original disease. Thus, a modifier likely is segregating in genomic proximity to the *prcd* locus on CFA9. 11 and 13 animals affected by the fast and slow phenotype, respectively, were compared against each other on the canine Affymetrix SNP chip with respect to a control group comprised of 10 known carriers.

An initial 1,807 CFA9 markers in 29 individuals passed minimal quality requirements. PLINK, and t-tests for selected markers, placed the most likely signal for *prcd* between SNP 6,837,703 and 7,230,119, indeed including the mutation (7,186,710). Surprisingly, additional areas of interest are interspersed throughout CFA9. The strongest signal, 36 Mb from the mutation, suggests involvement of the neuronal amiloride-sensitive cation channel 1 (*ACCNI*) with disease phenotype, which is currently being investigated. Although preliminary, these data may provide first indicators for a potential *prcd* modifier.

Poster 2131

Title: A microarray-based approach for the identification of skeletal muscle genes related to stress in pigs.

Presenting Author: Zambonelli Paolo, DIPROVAL, Sezione Allevamenti Zootecnici, University of Bologna, Via Fratelli Rosselli107, 42100 Reggio Emilia, Italy.

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Abstract:

Stress reactions to slaughtering influence muscle metabolism and technological meat quality. The aim of this work was to identify genes involved in stress reactivity in pigs by using DNA microarray analysis. A total of 33 pigs of three breeds: Italian Large White (ILW), Italian Duroc (ID) and Pietrain (PI) were bred on the same farm and slaughtered in a single day on the same abattoir. All ILW and ID pigs had genotype 1843CC at RYR1 locus while PI pigs were CC and CT. At the abattoir the pigs were divided into two groups and only one was physically stressed by running along the raceway before slaughtering. After slaughter, samples of semimembranosus muscle were collected for RNA extraction. Labelled cDNAs were hybridized to 11K pig oligoset version 1.0 (Operon-Quigen) microarrays using a heart cDNA sample as common reference. The obtained data were analyzed in SAS using a mixed model. Using DAVID Bioinformatic Resources the ontology of differentially expressed genes among stressed and not-stressed pigs was produced for each breed. The results showed that the impact of preslaughter stress on metabolism determines different responses between breeds in term of differentially expressed genes. The differential expression of stress related genes will be validated by qRT-PCR.

Poster 2132

Title: The lavender plumage colour phenotype in Quail is caused by a large deletion in MLPH gene region

Presenting Author: BED'HOM Bertrand, INRA/AgroParisTech UMR1236 Génétique et Diversité Animales, Domaine de Vilvert, Bâtiment 211, F-78352 Jouy en Josas Cedex, France

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Abstract:

The lavender phenotype in Japanese Quail is recessive and autosomal. It produces a dilution of both eumelanin (from black to blue or light grey) and pheomelanin (from red/brown to buff or beige) in feathers. A similar phenotype is also known in several mammalian species (Mouse, Dog, Cat, Mink, Human) and in Chicken. All these phenotypes are caused by mutations in MLPH (Melanophilin) gene, which is involved in melanosomal transport in melanocytes. Homology between Japanese Quail and Chicken lavender loci has been demonstrated by hybridization between these animals (Minvielle *et al.*, 2002). We have previously established that the mutation in chicken MLPH is a single-point mutation in exon 1, implying amino-acid change in protein (Vaez *et al.*, 2008). By comparison of genomic organization in MLPH region between homozygous lavender and wild-type quails, we describe here that the mutation in Japanese Quail is surprisingly a large deletion encompassing the 3' end of MLPH gene (11 exons out of 17 are deleted), the entire PRLH (Prolactin Releasing Hormone) gene and the 3' end of RAB17 (member of RAS oncogene family) gene (5 exons out of 6 are deleted).

Poster 2133

Title: A genome scan for athletic performance in the thoroughbred

Presenting Author: Keith Durkin, Department of Veterinary Integrative BioSciences, Texas A & M University, College Station TX 77843, USA

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1. Terje Raudsepp
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Abstract:

The primary goal of the thoroughbred industry is to breed and train superior equine athletes capable of excelling on the racetrack. To date research into the genetic underpinnings of athletic ability has been limited in the horse. Advances in equine genomics and the genetics of athletic performance in humans spurred

us to begin investigations into this important trait. We initially mapped 47 candidate genes associated with human athletic performance in the equine genome by radiation hybrid (RH) and fluorescent in situ hybridization (FISH) mapping. Using RH data and later the draft equine sequence we identified microsatellites adjacent to these and other candidate genes, additional microsatellites were added to increase genome coverage producing a final panel of 205 markers. All potential markers were initially screened on a pool of 16 thoroughbreds to ensure they were polymorphic. The panel was genotyped on 161 thoroughbreds; Centimorgans (cM) between microsatellites were determined with CRI-MAP. The animal's athletic ability was estimated using career winnings which were log_e transformed to create a linear trait; unraced animals were treated as missing data. The data was analyzed in the program MERLIN using the non-parametric linkage analysis option. None of the genotyped microsatellites showed significant linkage with career earnings.

Poster 2134

Title: Novel mutations of bovine FASN gene, g.16024A>G and g.16039T>C, effects on the fatty acid composition of Japanese black beef.

Presenting Author: Tsuyoshi Abe/ 1, Odakurahara, Nishigo-Mura, Fukushima, 9618511, JAPAN

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Abstract:

We constructed the F2 reference family, derived from the cross between Japanese black and Limousin, and detected significant QTL for fatty acid composition of back fat, intermuscular fat, and intramuscular fat at 60 to 70cM region on BTA19. We then focused and analyzed bovine fatty acid synthase (FASN) gene as a candidate for this QTL. Eight novel and 4 known mutations were detected in the coding sequence of the FASN gene between two breeds that were used to produce our F2 population. Among these mutations, 6 were nonsynonymous, and in particular, g.16024A>G and g.16039T>C, detected in exon 34, which determine amino acid substitution of threonine (T) to alanine (A) and tryptophan (W) to arginine (R),

respectively, were clearly separated in the 2 parental breeds. Two haplotypes comprising these 2 mutations (TW and AR) were segregated in F2 individuals, and had a significant effect on the fatty acid composition of three adipose tissues above. The TW haplotype was associated with increasing C18:0 and C18:1 content and MUFA/SFA, and decreasing C14:0, C14:1, C16:0, and C16:1 contents, respectively. Furthermore, we observed a markedly higher frequency of the TW haplotype in Japanese Black (0.67) than in Holstein (0.17), Angus (0.02), and Hereford (0.07).

Poster 2135

Title: Comparative Study on Gene Expression of DNA Methyltransferases in *Gallus gallus* and *Mus Musculus*

Presenting Author: Taeko Isokane, Graduate School of Biosphere Science, Hiroshima University, 1-4-4, Kagamiyama, Higashi-Hiroshima, JAPAN; 739-8528

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Abstract:

In mammals, genomic imprinting leads to the monoallelic expression of specific genes. Commencing with genomic imprinting, DNA methylation is an essential process in the course of development, including X-chromosome inactivation. Five DNA methyltransferase genes (*Dnmts*) are involved in the methylation in mammalian species. However, avian species are reported to have no methylation in their genomes, which indicates that little transcription occurs from the genes orthologous to mammalian *Dnmts*. In the present study, the amounts of transcripts from 4 *Dnmts* (no ortholog of *Dnmt3L* in chicken) were determined in chicken 4 organs (Brain, Liver, Kidney, Testis) using real-time RT PCR through developmental stages, and compared with those of corresponding mouse organs. In the course of development, the amount of *Dnmt1* mRNA was consistently lower in chicken organs than in the corresponding mouse organs; those of *Dnmt2* and *Dnmt3b* mRNAs were low in chicken as well as in mouse organs; that of *Dnmt3a* mRNA was generally higher in chicken than in mouse. These findings indicate that the 4 *Dnmts* in chicken may contribute to the DNA methylation, though the methylation has not been reported so far; or that they have different roles other than methylation in chicken development.

Poster 2137

Title: Isolation and characterization of 27 novel microsatellites for individual identification and parentage testing in Humboldt penguin (*Spheniscus humboldti*)

Presenting Author: Kei Onga, Department of Life Science, Faculty of Agriculture, Meiji University, Higashimita, Tama-ku, Kawasaki-city, Kanagawa 214-8571 Japan

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Abstract:

The Humboldt penguin is one of the most endangered species in the wild. To conserve the species, captive breeding has become important at zoological organizations. High levels of genetic diversity are also essential for species survival. Therefore, appropriate management of individual pedigrees is required. Microsatellites are widely used in parentage testing and population genetic studies in various species. However, no microsatellites for parentage testing have been reported in the Humboldt penguin. We constructed a genomic library enriched with (CA)_n repeats. Thirty-one novel microsatellites were efficiently isolated from the library, and were genotyped in 24 Humboldt penguin individuals. Of these, 27 microsatellites were polymorphic. Heterozygosity ranged from 0.042 to 0.917, with an average of 0.514, while the number of alleles ranged from 2 to 13, with an average of 5.3. Power of discrimination (PD) and probability of exclusion (PE) ranged from 0.080 to 0.970 and 0.020 to 0.740, respectively, resulting in total PD and PE values of $1-1.80 \times 10^{-19}$ and 0.9999994, respectively. Application of these novel microsatellites will be a powerful tool for individual identification and completion of pedigree records in setting up captive breeding plans.

Poster 2138

Title: Allelic Analysis of Thoroughbred Horses Using Fourteen Microsatellite Markers

Presenting Author: SHIN-WOOK KANG & Korea Racing Authority, 685, Juam-dong, Gwacheon-si, Gyeonggi-do, South Korea, 427-711

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Abstract:

Microsatellite are repetitive DNA sequence that are randomly distributed throughout vertebrate genome. In this study, we experimented the total 1,660 thoroughbred horses. Genomic DNAs were prepared from hair roots samples. Genomic DNAs from Thoroughbred horses were extracted using MagExtractor System MFX-2000 according to the manufacturer's protocols. Multi-plex PCR was performed to respectively amplify alleles using horse DNA kit. We analyzed a number of alleles, allelic frequencies, observed(H_o) and expected(H_e) heterozygosity, probability of exclusion(PE), total PE for parentage test.

The number of alleles of the markers was varied between 4 and 10 with an average number of alleles of 6.6. The heterozygosity were ranged from 0.527 to 0.855 and mean expected heterozygosity was 0.706. Observed PIC was from 0.494 (CA425) to 0.817 (ASB2) and mean PIC was 0.659. The PE was observed from 0.314(CA425) to 0.674(ASB2) and total PE of all markers was 0.9999. These results verify that fourteen microsatellite markers used in this study are very powerful for parentage test and individual identification of thoroughbred horse in Korea.

Poster 2139

Title: Fine mapping of ovine microphthalmia

Presenting Author: Adrian Brunner, Institute of Genetics, University of Berne, Switzerland

Other authors (name only):

1. Jens Tetens
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Abstract:

The ovine microphthalmia (OMO) is characterized by bilateral abnormal small eyes in newborn lambs. This congenital anomaly is inherited as a monogenic autosomal recessive trait with complete penetrance and is predominantly found in the Texel breed. We assigned the OMO locus by linkage mapping to a 12.3 cM interval on OAR23. This chromosome region corresponds to segments of HSA18 and BTA24. We constructed a BAC contig covering the region of interest based on sheep-human BLASTN sequence alignments. For fine mapping purposes we scanned the bovine genome sequence for additional microsatellites and comparatively anchored ovine BAC end sequences (BES). PCR primers were designed from the anchored ovine BES and SNPs were developed in targeted regions by re-sequencing these PCR products. Eight new polymorphic

microsatellites and 206 SNPs were identified using this approach. These markers cover the entire critical interval with marker spacing between 5 and 200 kb. Genotyping of 117 affected lambs and 163 healthy relatives belonging to 60 sheep families lead to a reduction of the critical interval to approximately 8 Mb on the virtual OAR23 map.

Poster 2140

Title: Association of PPARGC1A with growth in a Charolais x German Holstein F₂ resource population

Presenting Author: Annett Eberlein, Research Institute for the Biology of Farm Animals (FBN), 18196 Dummerstorf, Germany

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3. Christa Kühn

Abstract:

The birth weight of calves is an important economic trait and strongly associated with dystocia, stillbirth and hypothermia, the most frequent causes for perinatal deaths. Birth weight is also used as an initial reference point for monitoring the development of an individual animal, because it represents the result of the most dynamic process of prenatal growth and development. Several studies reported overlapping QTL for birth weight, calving traits and stillbirths on bovine chromosome 6 (BTA6) in different breeds. On this account QTL detection for birth weight was performed in F₂ individuals generated by embryonic transfer as well as their P₀ and F₁ ancestors of a Charolais x German Holstein resource population. The analysis shows a genomewide significant QTL ($p < 0.05$) for birth weight on BTA6. This is in agreement with a previously reported QTL for stillbirth in the German Holstein population. Within this region the PPARGC1A gene (peroxysome proliferator-activated receptor γ coactivator 1 α) is located, a potential candidate gene, because of its function in energy, fat and glucose metabolism as a basis for growth and development. SNPs in the PPARGC1A and other candidate genes located in the targeted chromosomal region were tested for association with prenatal growth.

Poster 2141

Title: Bovine chromosome 18: Mapping of quantitative trait loci for somatic cell score and udder conformation traits

Presenting Author: Bodo Brand, Research Institute for the Biology of Farm Animals (FBN), 18196 Dummerstorf, Germany

Other authors (name only):

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Abstract:

To improve udder health and resistance against clinical mastitis (CM) many studies have attempted to identify chromosomal regions, genes and polymorphisms that exert influence on udder health in order to include this information in upcoming breeding strategies. So far QTL for CM or somatic cell score (QTL_{SCS}) have been detected on all autosomes. On bovine chromosome 18 (BTA18) several studies detected QTL_{SCS} in the telomeric region where also a QTL for mastitis incidence was found. To further characterise this QTL_{SCS}-region on BTA18 QTL detection in 6 large half-sib families in the German Holstein population was performed. Merging information from published linkage-maps, RH-maps, human and bovine sequence-assemblies as well as own linkage- and RH-mapping results enabled definition of a refined marker order and highlighted discrepancies in the bovine NCBI sequence-assembly Build_3.1. In order to gather information about the functional background of the QTL_{SCS} QTL analysis for SCS and functional traits with potential impact on SCS were performed. First results show that experimentwise significant QTL for SCS and udder conformation traits segregate in the telomeric region of BTA18. To check the value of the QTL_{SCS} information an exemplary marker assisted selection of animals with high and low susceptibility to mastitis was performed.

Poster 2142

Title: Hypotrichosis in a Charolais x Holstein cross is not associated with allelic variants in the bovine SILV gene

Presenting Author: Christa Kühn, Research Institute for the Biology of Farm Animals (FBN), 18196 Dummerstorf, Germany

Other authors (name only):

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Abstract:

A hypotrichosis phenotype, also known as rat-tail syndrome, is segregating in a Charolais x German Holstein F₂- resource population as well as in backcrosses from the F₂ individuals to a purebred German Holstein sire. The phenotype is characterised by curly, sparse hair in pigmented regions of the skin, whereas unpigmented hair is unaffected. Recently,

there had been reports that mutations in the bovine *SILV* gene that harbours polymorphisms associated with Charolais coat colour dilution (Dc) might also be the functional background of the hypotrichosis phenotype. Sequencing the entire coding region as well as the promoter of the *SILV* gene of seven individuals with different dilution, background coat colour and rat-tail-phenotypes did not reveal any polymorphism in total concordance with the expression of the hypotrichosis phenotype. For the bovine *SILV* gene, multiple splice variants have been described. However, sequencing *SILV* transcripts in skin from individuals with different dilution and hypotrichosis phenotypes did not reveal any indication on splice variation being responsible for the defect in hair structure. Thus, we conclude that segregation of hypotrichosis is independent from the *SILV* dilution locus. A refined model of inheritance of hypotrichosis in the Charolais x German Holstein F2 population will be presented.

Poster 2143

Title: Homozygosity mapping of arachnomelia in Brown cattle

Presenting Author: Tosso Leeb, Institute of Genetics, University of Berne, Switzerland

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Abstract:

Fifteen calves affected with arachnomelia were sampled in the Swiss and Italian Brown cattle population. Analysis of the pedigree data confirmed monogenic autosomal recessive inheritance and revealed that all affected individuals traced back to a single acknowledged carrier founder sire. Eight affected offspring and 10 available relatives were genotyped for a panel of 240 microsatellites spanning the 29 bovine autosomes. Homozygosity mapping showed homozygosity of all cases at two adjacent microsatellite markers on BTA5. For fine mapping purposes 14 additional BTA5 microsatellites were genotyped in all available cases and indicated shared homozygous haplotypes across 20 Mb among affected individuals. A second pedigree with 23 acknowledged carriers among 34 sires in total related to the founder were genotyped for these markers. Linkage and haplotype analysis showed that the arachnomelia mutation could be assigned to a 7.2 Mb candidate region (LOD score >6). The linked markers can be

used for indirect determination of the arachnomelia genotype in offspring of related carrier sires. This bovine chromosome segment corresponds to an ortholog segment of HSA12q containing more than 140 annotated genes and loci. The only obvious functional candidate gene of this region *GDF11* is probably not responsible for this fatal bovine disorder.

Poster 2144

Title: Genotyping methodology and frequencies of the myostatin F94L mutation in Australian cattle breeds.

Presenting Author: Dianne M Vankan, University of Queensland, Animal Genetics Laboratory School of Veterinary Science, St Lucia, Qld 4072 Australia

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Abstract:

The F94L SNP (g.433C>A) in the growth differentiation factor 8 (*GDF8*) gene, commonly known as the myostatin (*MSTN*) gene, has recently been shown to be functionally associated with increased muscle mass and carcass yield in cattle. Since the F94L mutation is not, like other *MSTN* mutations, associated with reduced fertility and dystocia, it is a candidate for introgression into other breeds to improve retail beef yield, and development of an accurate test for genotyping this mutation is warranted. Accurate genotyping of the F94L SNP by primer extension is not possible so genotyping has been achieved using PCR-RFLP. We found that variations in the efficiency of cleavage of PCR amplicons by *TaqI* compromised the accuracy of genotyping, resulting in an overestimation of the frequency of the mutant A allele. We developed two real-time PCR assays that both accurately genotype the F94L mutation in cattle based on allele-specific and high resolution melt analysis. The frequency of the F94L mutation was determined in 960 animals from 14 breeds of cattle. The mutation was present in Simmental (0.8%), Piedmontese (2%), Droughtmaster (4%) and Limousin (94.2%) but not found in Salers, Angus, Poll Hereford, Hereford, Gelbvieh, Charolais, Jersey, Brahman, Holstein and Maine Anjou.

Poster 2145

Title: Gene expression profiling in pig liver tissue following administration of Clenbuterol-HCL

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Abstract:

Clenbuterol-HCL can greatly reduce fat deposition and increase skeletal muscle growth at high dosages. In order to advance our understanding of the fundamental molecular mechanisms, Genechips, real-time PCR, two-dimensional electrophoresis and mass spectra were used to study the differential gene expression profiles of liver tissues among the pigs with/without Clenbuterol-HCL. Four Affymetrix Porcine Genome Genechips were used for global evaluation of the gene expression. Totally 692 probe sets presented on Microarrays were identified differentially expressed with 337 upregulated and 355 downregulated (False discovery rate adjusted p-values < 0.05). Gene ontology functional annotation analysis was applied to the above genes, and the most significantly overrepresented categories were deemed the organismal physiological process, enzymes of lipid metabolism, receptors of membrane, and mediators involved in positive regulation of signal transduction. Moreover, pathway mining analysis revealed that physiological pathways such as MAPK, Wnt, cell adhesion molecules as well as signaling pathway from G-protein families, etc have been regulated remarkably when Clenbuterol-HCL was feed. Results obtained in this study deepen our understanding in the function mechanism of β -adrenergic receptor agonists on animal models. Differentially expressed genes identified will be the candidates for further investigations on the molecular mechanisms involved in Clenbuterol-HCL of reducing adipose accumulation.

Poster 2146

Functional analysis of the equine kappa casein (CSN3) gene promoter

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Kappa casein gene (CSN3) is the fourth member of the casein gene cluster in mammals, which is evolutionary not related with the other three casein genes. However, the structure of its promoter allows the spatially and temporally coordinated expression with the other three casein genes. In our study we sequenced 2286bp of the equine CSN3 promoter and performed *in silico* search for potential transcription factor binding sites. Thirty two potential TFBSs were found by manual search within the 2286 bp region of the CSN3 gene promoter and additional ten were identified by bioinformatic tools. Fifteen SNPs were discovered by sequencing of the promoter and 12 of them were located within the identified TFBSs, potentially interfering with TF binding. We found SNPs in putative binding sites for NF1, C/EBP and TFIID, which are all presumed to be involved in expression of casein genes. Sequence alignment of the core CSN3 gene promoter (1234bp) of nine species (sheep, goat, cow, zebra, donkey, horse, human, chimp, macaque), revealed 22 completely conserved sequence stretches, six to fourteen bp in length and seven of them harbored potential transcription factor binding sites (TRFBs). Since many of these potential TFBSs may not be functional, it is possible that many of the discovered SNPs are not associated with TF binding. Electrophoretic mobility shift assay (EMSA) using synthetic DNA fragments representing potential equine CSN3 gene promoter elements has been performed. Furthermore, reporter gene constructs containing different parts of the equine CSN3 promoter were constructed and tested in the heterologous (bovine and murine) cell system.

Poster 2147

Title: Identification of the causative mutation of congenital pseudo-myotonia in Chianina cattle

Authors:

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Abstract:

Congenital pseudo-myotonia (PMT) in Chianina cattle is a muscle function disorder which is mainly characterized by an exercise-induced muscle contracture which prevents animals from performing muscular activities more intense than a simple walk at a slow pace. Mutations in the human *ATP2A1* gene, encoding a fast-twitch skeletal-muscle Ca^{2+} ATPase (SERCA1), cause Brody myopathy, a very similar rare autosomal recessive disorder characterized by exercise-induced muscle cramps and impaired muscle relaxation. The analysis of the collected Chianina pedigree data suggested monogenic autosomal recessive inheritance and revealed that all PMT affected individuals traced back to a single founder sire. A family with 16 PMT affected cattle was genotyped with two bovine *ATP2A1* gene flanking microsatellites. Linkage analysis within this family showed that the PMT mutation could be assigned to the *ATP2A1* gene region on BTA25 (LOD score >3). Subsequent DNA sequencing of the 16 PMT affected calves revealed a missense mutation (c.491G>A) leading to a p.Arg164His substitution in exon 6 of *ATP2A1*. Arg164 in bovine *ATP2A1* is located within the functional important N-terminal actuator domain and is a highly conserved residue. Genotyping 112 unaffected unrelated Chianina animals did not reveal a single individual homozygous for mutation and indicated a carrier frequency of >0.10.

Poster 2148

Title: Genetic Characterization of Korean Cattle (Hanwoo) using SNP Markers

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Korea government has introduced genetic testing to identify the origin of beef and to assure safety and quality of domestic “Hanwoo” beef products. To develop reliable and accurate markers, we used to single nucleotide polymorphisms (SNP) based on the information of NCBI bovine SNP database. The SNP sequence data developed at the BCM-HGSC were used for DNA sequencing of Hanwoo, Hereford and Limousin. One hundred four SNP sequences spanning 27.5 kb were randomly selected and analyzed by

direct sequencing of 24 individuals for each breed. A total of 270 SNPs were identified from the sequence comparisons, among which 22 SNPs were further genotyped for the 960 animals with Hanwoo or other foreign breed origin. Thirteen SNP markers showed significant genotypic differences between Hanwoo and other beef breeds, which may enable designing of robust and accurate high-throughput genotyping assays for Hanwoo meat identification.

Poster 2149

Title : Identification of new pig cSNPs using polymorphisms between Korean native pig and Genbank registered sequences.

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7. Kunho Suh
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Abstract

Sequences from the clones of full-length enriched cDNA libraries serve as valuable resources for functional genomic studies. We have analyzed 1,970 high quality chromatograms (Phred value ≥ 30) that were obtained from sequencing the 5' ends of brainstem, liver, neocortex and spleen clones derived from full-length enriched cDNA libraries from Korean native pigs. In addition, 50,000 pig EST sequence trace files were obtained from Genbank and combined with our sequencing information to facilitate SNP identification in silico. The process generated 8,118 contigs containing 678 putative cSNPs. Of these, 33 putative cSNPs were randomly selected for confirmatory analysis and validated using 20 pigs from four different breeds (Duroc, Landrace, Yorkshire, Korean native pig). 20 of the 33 putative cSNPs were confirmed with a confirmation percentage of 61%, which was similar to that reported by other studies. We also identified 15 new cSNPs from the validation process, which were not detected by our in silico analysis. Our study shows that analyzing genetically diverse breeds pig breeds such as the Korean native pig could serve as a useful strategy for generating a large number of cSNPs.

Poster 2150

Title: Regulatory SNPs and fat related traits in cattle. Implications of the bovine g.763G>C *FASN* SNP on milk fat content.

Presenting Author: Laura Ordovas, Laboratorio de Genética Bioquímica, Facultad de Veterinaria, Miguel Servet 177, 50013 Zaragoza, Spain

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7. Clementina Rodellar

Abstract:

Recent surveys have estimated that between 22% and 35% of SNPs located in regulatory regions (rSNPs) have functional consequences. The accumulation of several rSNPs could constitute the basis for the different gene expression profiles that define traits in a species-, breed- or individual specific manner. The aim of our research is to identify and functionally characterize rSNPs in candidate genes for fat related traits in cattle with the ultimate goal of obtaining genetic profiles that could define them. As result, we previously reported the identification of a Fatty Acid Synthase (*FASN*) SNP, g.763G>C, that was significantly associated with milk-fat content in dairy cattle. The SNP is included in a GC-rich region that may constitute a *cis* element for Sp transcription factors (TFs). In this work, the functional characterization of the g.763G>C SNP is presented. Reporter assays in 3T3L1, HepG2 and MCF-7 cells showed that the SNP affected the bovine *FASN* promoter activity. Supershift assays (EMSA) demonstrated that the Sp1/Sp3 TF-binding ability of the sequence was also altered by the SNP in these cellular lines. In bovine lactating mammary gland, EMSA assays showed that the specific binding of Sp3 may account for the association of the g.763G>C SNP with milk-fat content.

Poster 2151

Title: "Discarded SNPs to be used for identification even showing intermediate frequencies"

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Abstract:

SNPs are being implemented as an essential tool in traceability and identification studies. In this sense, a set of SNPs has been selected from two previously reported panels to be included in a bovine meat traceability Project. One of these SNPs was the transversion g.329C>T (AJ496781) on cytochrome P450 17A1 gene at the seventh intron, which maps on bovine chromosome 26. It was tested by minisequencing in a total of 702 samples belonging to eight Spanish cattle breeds. The observed heterocigosity for this marker was 0,98, which implies a high deviation from Hardy-Weinberg equilibrium ($P < 0.001$) as a consequence of a heterozygote excess). A BLAST search performed between the described sequence containing the SNP (AJ496781) showed two identity with two with two different regions in the bovine contig NW_001503883.1 Each region showed a fixed base C or T in the SNP position. We carried out sequencing analysis that supported the hypothesis of two different fragments exist. As numerous gene duplication within superfamily genes had been reported and because of its key role on metabolism we supported the hypothesis of duplication of gene *CYP17A1* in *Bos Taurus* genome. Therefore we strongly recommend to discard this transversion g.329C>T (AJ496781) or any SNP located in this region to be used in identification, traceability or disease association studies

Poster 2152

Title: High exclusion probability in ovine genealogical control through 18plex and 20plex PCR

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Abstract:

Genealogical control is one of the weaknesses in the Selection and Breeding Programs in sheep breed mainly due to the reproduction systems, based in the use of large number of females with several males more than artificial insemination. It is very important to develop a system allowing certify animals genealogy as well as the unequivocally identification of them throughout their life. Central Veterinary Laboratory Algete (LCV) in collaboration with TRAGSEGA has developed a high-exclusion

capacity, *multiplex* PCR system which amplify, in two reactions 18 (BM1818, CSR247, ETH152(D5S1), HSC, ILST87, INRA005, INRA006, INRA023, INRA049, INRA063, INRA172, MAF65, McM42, McM527, OarCP49, OarFCB20, SPS113 y SPS0115) and 20 (BMS1967, BP28, BP33, CD5, CSR2111B, CSR2115B, DU330122, ILSTS05, ILSTS39A, ILSTS62A, INRA081, MAF209, MMP9, OarAE119, OarFCB128, OarJMP11, TCRVB6, TGLA53, THRA, UMJM30) microsatellite markers, respectively. All of them have been selected from the list proposed by the International Society for Animal Genetics (ISAG) for the 2007-08 Ovine Comparison Test and used for studies in this specie. Primers were adapted to achieve a final configuration that allowed all markers to be analysed together. Robotic procedures were implemented to minimize the risk of genotyping errors and parentage verification mistakes. The proposed system has been designed to process a very large number of samples in a short space of time ensuring a perfect reliability and traceability of the obtained results. The main set of markers is used routinely for ovine parentage verification at LCV and reached a exclusion probability of 99,9999930%. It further provides for a panel composed of 20 additional markers to be used in cases where there is a need for greater exclusion capacity.

Poster 2153

Title: Effect of a SNP in the promoter region of the lactoferrin gene upon ovine milk somatic cell counts

Presenting Author: Maria Rosário Marques, Escola Superior de Biotecnologia, Universidade Católica Portuguesa, Rua Dr. António Bernardino de Almeida, P-4200-072 Porto, Portugal

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Abstract:

Lactoferrin (LF) is an iron-binding glycoprotein, which is naturally found in milk. Its concentration in milk increases dramatically in response to udder infection, which may be an indication of its role upon prevention of/fight against mastitis. The occurrence of subclinical mastitis has been related to increased levels of somatic cell counts (SCC) in milk. The objective of this effort was to find single nucleotide polymorphisms (SNP) in the promoter region of the LF gene, and ascertain their effects upon SCC in ewes' milk.

Polymerase chain reaction – single strand conformation polymorphism analysis (PCR-SSCP)

was carried out in a DNA fragment of the promoter of the LF gene. Three SSCP patterns were detected and sequenced, and a SNP (A/G) was found at position 73 (GenBank sequence n° AF091651). Seventy-six *Lacaune* crossbred sheep were genotyped. The frequencies of the three SSCP patterns, which correspond to genotypes AA, AG and GG, were 71.1%, 27.6% and 1.3%, respectively. Mixed model data analysis revealed that AA ewes tended to have significantly lower SCC in milk than AG ewes ($P < 0.10$), but no differences were observed on milk production. The aforementioned SNP disrupts a putative transcription factor binding site in the promoter of the LF gene – hence suggesting its involvement in the regulation of LF expression levels.

Poster 2154

Title: Expression profile of the MRF4 gene in muscle during postnatal growth of pigs.

Presenting Author: Mariusz Pierzchala, IGAB PAS, 05-552 Jastrzebiec. Poland

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Abstract:

The MRF4 (herculin) is a member of the basic-helix-loop-helix family of proteins. The basic Helix-Loop-Helix (bHLH) proteins are transcription factors that play important roles during the cells development of various tissues. The myogenic factor MRF4 gene is expressed in a unique pattern defined by period and place within the skeletal muscle. The goal of our study was to analyse the MRF4 gene expression profile during postnatal growth pigs. As the study material the 5 different pig breeds were chosen: Polish Large White, Polish Landrace, Duroc, Pietrain and Pulawska (polish native pig breeds). Two muscle probes - m. longissimus dorsi and m. semimembranosus were collected from pigs at slaughter at the age of 60, 120 and 180 days. Quantification of transcripts were performed by Real-time PCR technique. The MRF4 gene transcript level was standardised to three reference genes such as: B-ACT, GAPDH, and B2M. The results show significant variation in the level of MRF4, related to breed and the age of slaughter. The highest difference were observed between Duroc, Pietrain and Polish Landrace or Pulawska. This confirm such presumption that selection of pig breeds would result in variation of metabolic pathways of myogenic factor (MRF4) regulation.

Poster 2155

Title: Polymorphism of the *ADIPORI* gene and its transcription level in two skeletal muscles during postnatal development in five pig breeds.

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¹ Department of Genetics and Animal Breeding, University of Life Sciences in Poznan, Poland; ² Chair of Animal Breeding, Technical University of Munich, Freising, Germany

Abstract:

The adiponectin receptor 1 gene (*ADIOPORI*) can be considered as a candidate for the pig fatness and growth. The aim of the study was searching for its polymorphism as well as transcription level in two skeletal muscles (*m. longissimus dorsi*, *m. semimembranus*). Five breeds were analysed: Polish Large White, Polish Landrace, Duroc, Pietrain and Pulawska. Tissue samples were collected *post mortem* at the age of: 60, 90, 120, 150, 180 and 210 days. Transcription start site and a nucleotide sequence of the 5'UTR region was established. The transcript of the porcine *ADIPORI* gene contains 190 bp of the 5'-UTR sequence (GenBank, EU556496) and its sequence is similar to a previously reported *ADIPORI* transcript in human with the identity of 85%. In this fragment the Kozak consensus sequence occurs at the position 188–194 bp. Altogether, eleven SNPs were detected in exon 5, intron 6 and 3'UTR. Genotyping performed for a silent SNP in exon 5 (759 G>A) and two SNPs in 3' UTR (+ 129 A>C and + 536 A>G) revealed interbreed variability in terms of allele frequencies. Differences between breeds in the transcript level occurred mainly at early stages of the postnatal development (60, 90 and 120 days).

Poster 2156

Title: Single Nucleotide Polymorphism (SNP) discovery from the horse Y chromosome (ECA Y)

Presenting Author: Maria do Mar Oom, University of Lisbon, Faculty of Sciences, DBA/CBA C2-3° Piso, Campo Grande, 1749-016 LISBOA, PORTUGAL

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Abstract:

Mammalian Y chromosome is male specific, constitutively haploid and inherited exclusively through patrilineages. Polymorphic markers derived

from male specific region of Y (MSY) are, thus, ideal tools to trace recent evolutionary events in the populations of horses and other equids. Previous attempts to identify SNP or microsatellite polymorphism from ECA Y have not been successful. Taking advantage of the recently developed BAC contig map of the entire ECA Y we conducted a systematic search for SNPs both from MSY and the pseudoautosomal region (PAR). BAC end sequences were used to generate 54 STSs (sequence-tagged sites) which collectively covered ~ 30 kb. All STSs were sequenced from: i) a DNA pool of 39 male horses and ponies representing 11 diverse breeds; ii) Bravo –the DNA donor for CHORI BAC library; iii) 1 tuva horse; iv) 1 Mongolian horse; v) 2 Equus przewalskiis, and, vi) 1 donkey. Preliminary sequence analysis has already detected a few SNPs from the ampliconic region of MSY and from PAR.

Poster 2157

Title: MHC typing in swine: SLA class I and class II-allele distribution in Austrian Large White, Landrace, and Pietrain breeding stocks

Presenting Author: Sabine E. Hammer, Institute of Immunology, University of Veterinary Medicine Vienna, Veterinärplatz 1, A-1210 Vienna, Austria

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Abstract:

MHC (major histocompatibility complex) genes encode cell surface glycoproteins presenting antigenic peptides to T cells. They are highly polymorphic suggesting that diversity in MHC genes is a good measure of population fitness. Resource herds of swine leukocyte antigen (SLA)-characterized pigs are valuable large animal models for biomedical research in terms of immune responses, disease resistance, and production traits. Furthermore detailed knowledge about the distribution of MHC genes in a population enabled a more target-orientated design of vaccines and the development of specific reagents for studying correlates of protection like tetramers. This study represents the initial characterization of founder haplotypes of purebred Large White, Landrace, and Pietrain populations. The respective founder SLA-haplotypes were detected by a reverse transcription-polymerase chain reaction (RT-PCR)-based SLA typing method to clone and DNA sequence the putative alleles at four SLA class Ia loci, designated as SLA-1, SLA-2, SLA-3, and SLA-6 and four SLA

class II loci, SLA-DQA1, SLA-DQB1, SLA-DRA1, and SLA-DRB1. The obtained sequences are used to establish rapid SLA typing assays based on sequence-specific PCR primers (PCR-SSP) to customize allele-specific PCR screening assays for large-scale MHC typing of Austrian pig populations and on a next level European pig herds.

Poster 2159

Title: Genetic variation in pro-melanin-concentrating hormone affects carcass tenderness and fat levels in *Bos taurus* cattle.

Authors: Sarah C. Helgeson & Sheila M. Schmutz
Department of Animal and Poultry Science,
University of Saskatchewan, Saskatoon, Canada

Abstract:

Pro-melanin-concentrating hormone (PMCH) had previously been shown to affect appetite and metabolism in rodent species. *PMCH* was mapped to BTA5 (Stone et al. 2002) and QTL to backfat were reported for this chromosomal region (Casas et al. 2000, Li et al. 2004). The objective of this research was to examine whether there were similar effects in cattle. Sequencing of *Bos taurus PMCH* from several prevalent North American beef breeds revealed an adenosine-to-thymine single nucleotide polymorphism (SNP) upstream of the transcriptional start site. This SNP was subsequently shown to significantly affect both average fat and grade fat levels in two populations of beef cattle. One of these populations had tenderness data. Consumer taste panel evaluations indicated that the thymine allele contributed to decreased steak tenderness, as measured by a consumer taste panel and mechanical shear force, in the one population where these data had been measured. *In silico* analysis of the promoter region indicated that the presence of the thymine allele introduces a binding site for a transcriptional repressor, E4BP4. It is believed that the introduction of the thymine allele leads to downregulation of *PMCH*, which would increase physiological metabolic rates, resulting in tougher steaks and decreased fat production.

Poster 2160

Title: Using a 1.5K ovine SNP array to expand the sheep linkage map

Jillian F. Maddox¹, Allan M. Crawford², Roxann Ingersoll³, James Kijas⁴, Brian P. Dalrymple⁴, Ian R.W. Evans⁵, Frank W. Nicholas⁶, John McEwan², Tracey van Stijn², Herman Raadsma⁶ and the International Sheep Genomics Consortium⁷.

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Abstract

The sheep linkage reference map has been developed by genotyping a 3-generation, full-sibling pedigree, the International Mapping Flock (IMF), produced by AgResearch that consists of 127 sheep in 9 families. Version 4.7 of the map, released in December 2006, comprised 1,425 markers representing 1,381 loci and spanned approximately 3,600 cM. The majority of mapped markers (87%) were microsatellites, and 51% were derived from markers initially developed for cattle. The International Sheep Genomics Consortium (ISGC) recently identified 6,022 ovine SNPs from a targeted BAC end resequencing project and public ESTs. Of these 1,536 SNPs were selected for genotyping, based on their predicted chromosome positions, with Illumina's BeadArray platform. SNPs were selected to represent every ovine chromosome (OAR) with an average of 56 SNPs per chromosome ranging from one SNP on OARY to 151 SNPs on OAR1. The 1.5K array was used to genotype the IMF so that the SNPs could be integrated into the linkage map. The CRI-MAP and MultiMap programs were modified so that they could better handle a larger volume of genotyping data. Construction of version 5 of the linkage map is currently underway and the new map will contain approximately 2,530 markers including 1,113 SNPs from the array.

Poster 2161

Title: Expression of the chicken $\delta 2$ -crystallin gene: Occurrence of sense and antisense RNAs in testis.

Presenting Author: Takeshi SHIMOGIRI, Korimoto 1-21-24, Kagoshima, 890-0065, JAPAN

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Abstract:

δ -Crystallin, the major eye lens protein in birds, is homologous to the urea cycle enzyme argininosuccinate lyase (ASL). In chicken, there are two δ -crystallins, denoted $\delta 1$ and $\delta 2$. Of them, $\delta 2$ -crystallin gene (*CRYD2*) is the chicken ASL ortholog because only $\delta 2$ has retained the ASL activity. Birds are uricotelic in terms of nitrogen excretion and apparently do not require the urea cycle for nitrogen excretion. Here, to examine how and why the urea cycle genes have been maintained in chicken, we examined the *CRYD2* expression in chicken tissues by RT-PCR. We detected the *CRYD2* fragment in various chicken tissues using cDNA prepared with random hexamers and PCR. These results revealed the presence of the *CRYD2* mRNA in all tissues and two additional transcripts in testis. Chicken BLAT search indicated that exon/intron structure was different amongst the *CRYD2* mRNA and two testis-specific transcripts. Additionally, we found that two testis-specific transcripts were natural antisense RNAs of the *CRYD2* gene: the testis 1st-strand cDNA prepared with gene-specific forward primer produced the two PCR products corresponding to the testis-specific transcripts by the subsequent PCR reaction, while the testis 1st-strand cDNA prepared with gene-specific reverse primer produced the PCR products from the *CRYD2* mRNA.

Poster 2162

Title: Linkage disequilibrium mapping of QTL on chromosome 7 affecting female fertility and twinning rate in Israeli Holstein cattle

Presenting Author: Ron M., The Volcani Research Center, P.O.B. 6, Bet-Dagan 50-250, Israel

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Abstract:

Two segregating quantitative trait loci (QTL) affecting female fertility, defined as: 100/(number of inseminations to conception), were detected on BTA7 in the Israeli Holstein population by a daughter design analysis. One QTL was located near the centromere, and the other in the vicinity of 40 Mbp from the centromere. In order to confirm and fine map these QTL, linkage disequilibrium between SNPs and the putative QTL was analyzed by the regression of the genetic evaluations of 650 bulls on their marker genotypes. DNA samples were genotyped for 206 SNPs located within 43 Mbp from the centromere, using IPLEX technology. Two markers, located near position 42 Mbp had significant effects on female

fertility (comparison-wise $p < 10^{-4}$), and a marker near position 40 Mbp had a significant effect on twinning rate (comparison-wise $p < 10^{-4}$). Thus the QTL near position 40 Mbp was confirmed, but not the QTL near the centromere. The allelic substitution effects on female fertility and twinning rate were 1.05% and 0.71%, respectively. Previous results have shown a negative genetic correlation between twinning rate and fertility, and this was confirmed for all eight SNPs with nominally significant effects on both traits.

Poster 2163

Title: Fine mapping of QTL affecting Non Return Rate in French dairy cattle

Presenting Author: Slim Ben Jemaa: INRA, UR339 Laboratoire de Génétique Biochimique et Cytogénétique, F-78352 Jouy-en- Josas

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Abstract:

Our aim was to map finely three quantitative trait loci (QTL) influencing female fertility (FF) in the French dairy cattle breeds Prim'Holstein, Normande and Montbeliarde which had been previously detected, by linkage analysis (LA) on bovine chromosomes (BTA) 1, 2 and 3 on a sample of 78 half-sib families from the three breeds. The mapping pedigree was composed of 41 families from the three breeds. Non return rates (NRR) within 56, 90 and 281 days after artificial insemination (AI) were used as FF phenotypes. The three QTL previously detected, were confirmed ($P < 0.05$) only in Prim'Hostein. Their localisation interval remained quite large. 437 SNP mapping to BTA03 were then genotyped on a sample of 17 Holstein families in order to narrow down the localisation interval of the QTL on this chromosome. LDLA analysis, refined the QTL position to a set of narrow peaks. To find the causal mutations, the regions including exons of six positional candidate genes: Paqr6, Spna1, Mtx1, Shc1, Adar1 and Tpm3 were sequenced on 4 individuals' pools (2 pools +/- and 2 -/-) belonging to heterozygous sires. Nine SNP having a compatible profile with the a priori status of the individuals were identified.

Poster 2164

Title: Evaluation of the genetic structure of the Brazilian Fila dog with microsatellites

Presenting Author:

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5. Denise Aparecida Andrade Oliveira

Abstract:

The Fila dog is the most important molosser breed in Brazil for its guard abilities, loyalty and its historical origin associated to the development of the Brazilian society and culture. But there is a lack of studies about its genetic structure. Using the ISAG recommended markers of 21 microsatellites, 26 non-related dogs were genotyped. All microsatellites were polymorphic, with a number of alleles ranging around 3 (AHTk253, INU030, REN162C04) and 8 (AHT121). Allele frequencies, inbreeding index (F), effective number of alleles (Ne), observed (Ho) and expected heterozygosity (He), polymorphic information content (PIC), parentage exclusion probability when both parents are typed (PE-2) and matching probability of two unrelated individuals (MPR), as well as Hardy-Weinberg genetic equilibrium were estimated. Mean Ho was 0.598 (ranging from 0.385 at AHTk211 and AHTH130 to 0.808 at REN105L03). Two loci (AHTH171 and INU005) were out of HW equilibrium ($p < 0.01$). The PIC value for the set of 21 microsatellites was 0.56. The PE-2 reached 0.9999. This study contributed to elucidate the genetic structure of the Brazilian Fila dog breed and to orientate conserving programs, in order to avoid high levels of endogamy, specially when associated to reproductive biotechnologies. Thanks: *GenLab/UFMG Veterinary School and CAFIB Fila Brasileiro breeders.*

Poster 2165

Title: Characterization of PrP gene polymorphism in South Croatian sheep populations

Presenting Author: Vlatka Cubric Curik, Faculty of Agriculture, University of Zagreb, Department of Animal Science, Svetosimunska 25, 10000 Zagreb.

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Abstract:

Polymorphism at codons 136, 154 and 171 in the ovine prion gene (PrP) influence the progress and incidence of scrapie and bovine spongiform encephalopathy (BSE). The European Union legislation promotes breeding programs that favour genetic resistance to scrapie. In Croatia, EU Candidate country, the genetic polymorphism in sheep populations has never been studied. Here, we present first results of the polymorphism at codons 136, 154 and 171 in six Croatian sheep breeds/populations (Cres Island sheep, Rab Island sheep, Pag Island sheep, Brac Island sheep, Dalmatian Pramenka and Dubrovnik Ruda), each sampled over more than 50 individuals. The preliminary results (estimates) have shown that three haplotypes; ARQ (> 50%), ARR and AHQ were the most common haplotypes in the Croatian meta-population, although, variability among breeds/populations have been observed. The estimated frequency of VRQ haplotype has been very low. Still, at some breeds/populations the frequency of “non-resistant” genotypes (ARQ/VRQ) was not negligible (> 5%) indicating that genetic resistance for polymorphism at codons 136, 154 and 171 is probably not the reason why scrapie has never been reported in Croatia.

Poster 2166

Title: Gene expression profiles in CNS of naturally scrapie-affected sheep

Presenting Author: Inmaculada Martín-Burriel, Laboratorio de Genética Bioquímica. Universidad de Zaragoza. Miguel Servet 177. 50013 Zaragoza (Spain)

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Abstract:

Transmissible spongiform encephalopathies (TSEs) are fatal neurodegenerative disorders caused by prions. Naturally scrapie-infected sheep can be considered as a natural animal model of these diseases. The underlying mechanisms of scrapie pathogenesis are still poorly understood. The identification of genes with differential expression in

CNS (central nervous system) of infected animals might provide clues to clarify the molecular mechanisms that lead to neuronal loss and to identify molecular biomarkers that might be the basis for new diagnostic tests. We present here the expression profiles of 18 genes involved in the regulation of apoptosis and in the response of cells to stress (HSPs) using Real Time RT-PCR. This study has been carried out in four CNS regions that display different lesion degrees. We have detected the up-regulation of pro-apoptotic genes and down-regulation of anti-apoptotic genes in brain stem (the most affected area). However, we have also observed the induction of antiapoptotic genes suggesting that neuroprotective mechanisms could counteract the effect of pro-apoptotic stimuli. Moreover, we report here an initial study on the transcriptional differences in cerebellum using cDNA microarray hybridizations. Small sequences of *GMPS*, *RPL32*, *ACTG1* and *ATP6AP2* genes align with up-expressed clones. By contrast, *GNB2L1*, *HSPA8*, *RPS3* and *FNI* genes have similarity with down-regulated sequences.

Poster 2167

Title: The pig as a biomedical model for melanoma: identification of MITF as a potential predisposing gene

Presenting Author: Emmanuelle BOURNEUF^{1,2}

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10. Silvia VINCENT-NAULLEAU

Abstract:

The MeLiM strain is a porcine model of cutaneous melanoma characterized by the spontaneous development of tumours *in utero* or during the first weeks of life. Clinical and histopathological observations have shown a high similarity between porcine and human lesions. A cross between MeLiM and healthy pigs was used to perform a QTL analysis, that mapped several susceptibility regions to swine chromosomes 1 (SSC1), SSC2, SSC13, SSC15 and SSC17. The most significant QTL ($p < 0.01$), located

on SSC13, is partly orthologous to HSA 3p14, and harbours the MITF gene, a master regulator of melanocyte biology. MITF amplification has been shown in human melanoma, and thus represents a particularly interesting candidate gene for melanoma predisposition in our model. We used BAC-end sequences to detect new microsatellites within the SSC13 QTL, and genotyped 15 on our backcross pedigree. The results show a refinement of the QTL peak above MITF, and an association between a close marker and melanoma development. We are currently analysing a panel of SNPs spread over 1Mbp around the gene to confirm this association. Ongoing functional studies have already shown a subtle regulation of MITF isoforms in melanocytes and tumors.

Poster 2168

Title: Mapping and expression studies of the DMRT1 gene in domestic pig

Presenting Author: Ewa Słota, Department of Immuno-and Cytogenetics of Animals, National Research Institute of Animal Production, Krakowska 1, 32-083 Balice n. Kraków, Poland

Other authors (name only):

1. Anna Bratuś

Abstract:

Doublesex and *mab-3* related transcription factor 1 (*DMRT1*) acts during animal sexual development and -in the majority of vertebrates- its function is limited to the upstream or downstream testis regulators. Porcine radiation hybrid panel (IMpRH) was used for physical mapping. PCR was carried out using two primer pairs located in 5'-flanking (AF426435) and 5'-UTR region (AF216651) resulting in PCR fragments of 399 and 122 bp respectively. PCR signals of hybrid clones were scored, evaluated with the IMpRH mapping tools (<http://imprh.toulouse.inra.fr>). For both examined sequences the closest linked marker is SW1621 (LOD score 5.66) on SSC1 (RH Map 4442 centiRay). Expression of swine *DMRT1* was analyzed by RT-PCR using dT-primers for RT and PCR-primers located on different exons (AF216651). Total RNA was isolated from various tissues. As a positive control all the cDNA samples were amplified with beta-actin primers (AJ312193). The highest *DMRT1* expression was detected in testis cDNA and followed unexpectedly by ovaries, kidney, heart and muscle. No amplicons were detectable in brain, liver, lung and spleen.

The present study of porcine *DMRT1* gene confirms its autosomal status and its chromosomal homology to humans, but reveals differences in expression pattern in adult tissues which is not limited only to testis.

Poster 2169

Title: The Ile-442-Met substitution in *NCAPG* as a positional candidate for bovine carcass weight QTL (*CW-2*) on chromosome 6

Presenting Author: Akiko Takasuga, Shirakawa Inst. Anim. Genet.

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Abstract:

We previously mapped carcass and body weight QTL (*CW-2*) on BTA 6 using Japanese Black Sire A's half-sib family. The *CW-2* QTL was replicated in Sire B that harbored the same *Q* haplotype as Sire A. We also mapped a body or carcass weight QTL in Sire C that harbored a different *Q* haplotype, and Japanese Brown Sires D and E that shared another *Q* haplotype, in the overlapped regions with *CW-2*. Assuming that these QTL were the same, we searched for a shared *Q* region using 39 microsatellite markers spanning the *CW-2* region (38-55 cM), and performed linkage disequilibrium mapping using maternal alleles of Sire C and Sire D offspring. An 810-kb region was shared among the three *Q* haplotypes and associated with body and carcass weight. By adding 42 SNP markers, the region was refined to 660 kb containing 4 genes. The SNP changing Ile-442 to Met in *NCAPG* (chromosome condensation protein G) was heterozygous in all the 5 sires and highly associated with carcass weight ($p < 1.2 \times 10^{-11}$) in a large Japanese Black population (187 steers selected from the 4.7% extremes of 7990 steers). This SNP provides a *CW-2* marker as a positional candidate QTN.

Poster 2170

Title: Biomarker development for pork quality using SELDI-TOF proteomics technology

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Abstract:

Development of meat quality relates to rate and status of the degradation of the muscle proteome after slaughter. Therefore, protein breakdown quantified using SELDI-TOF might identify good predictors of meat quality. SELDI-TOF technology, with its high throughput capability, allows for faster analysis of many samples permitting its use in explorative research of biomarkers. Knowledge regarding the progress of proteolysis on the entire proteome of muscle tissue is limited and breed-specific differences have received little attention. The objectives therefore were to investigate breakdown profiles of the m. longissimus dorsi proteome and the repeatability of the proteomic measurements. Meat quality and proteome profiles (in duplicate) of 5 Yorkshire and 5 Duroc pigs and proteome profiles were determined at slaughter and after 1, 2, 3, 7, and 10 days of ageing. The proteome showed four degradation profiles for different peptides. Association analysis suggested several biomarkers for drip loss and shearforce, but none for cooking loss. The repeatability between duplicated SELDI-TOF measurements ranged from 0.38 to 0.82 depending on the matrix and on the relative amount of the protein in the sample. For biomarkers development, samples should be taken at least in duplicate to minimize chance of obtaining false positive markers.

Poster 2171

Title: Selecting cattle for host resistance to tick (*Rhipicephalus microplus*) infestation – natural and human-directed selection

Presenting Author: Nicholas N Jonsson, University of Queensland, School of Veterinary Science, St Lucia, Qld 4072 Australia

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Abstract (199 words):

Two taurine populations of cattle with high resistance to tick infestation exist in Australia: the Adaptaur, selected by CSIRO from Hereford and Shorthorn cattle for resistance to ticks, and the Kimberley Shorthorn, a nearly feral descendent of original cattle introduced into Australia over 100 years ago and selected naturally to survive with ticks and without insecticide. We genotyped 58 Adaptaur and 94 Kimberley Shorthorns from tick-infested locations, 67 Shorthorn, 65 Hereford from non-infested areas, and 60 Brahman cattle, using 9 microsatellite markers from our parentage testing panel and 9 markers for

known genes within the MHC. Mean allele number in the MHC differed: Adaptours 5.6 ± 0.34 , Herefords 7.3 ± 0.91 , Shorthorn 9.2 ± 0.12 , Brahman 11.1 ± 1.3 and Kimberley Shorthorn 12.6 ± 1.6 . Linkage disequilibrium was high in the MHC in all breeds, particularly for CYP21 and DRB3. PIC values for both these loci in the Kimberley Shorthorn were 0.90 compared with overall mean PIC of 0.68. Our results suggest that directed selection for high host resistance led to a population with low diversity and likely high susceptibility to other diseases (Adaptours), while natural selection and gene flow from other breeds led to cattle with high resistance and high levels of variability in the MHC (Kimberley Shorthorn).

Poster 2172

Title: Characterization of ORF and LTRs of Porcine Endogenous Retroviruses in pigs.

Presenting Author: Kie-Chul Jung, Division of Animal Science and Resources, Chungnam National University, Daejeon 305-764, Korea

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Abstract:

Porcine endogenous retroviruses are one of serious problem regarding xenotransplantation because PERVs can infect human cells in vitro. Previously, PERVs have 30 to 50 copies in the pig genome and are classified as PERV-A, -B and -C based on the receptor-binding domain of the ENV gene. The PERV positive BAC clones were screened by PCR from the Korean native pig BAC library and 11 non-redundant BAC clones were selected for full-length PERV sequencing. Also, sequences from PERV flanking regions and chromosomal locations were investigated. The ORF analysis showed that only one PERV has complete ORF structure and the others have premature stop codons and INDEL mutations in gag, pol and env genes, indicating they are non-functional. Phylogenetic analysis of PERV LTRs was also performed and the preliminary results indicated that some of the LTR sequences are distinct from others. The results presented here can be a valuable information for generating PERV negative pigs which can be used as the xenotransplantation donor.

Poster 2173

Title: Polymorphisms in MYOD Gene Family Affect Carcass Traits in Cattle

Presenting Author: Mohammad Shamsul Alam Bhuiyan, Division of Animal Science and Resources, Chungnam National University, Daejeon 305-764, Korea;

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Abstract:

Myogenic differentiation genes (*MYOD*) are skeletal muscle specific transcription factors that encode highly conserved basic helix-loop-helix proteins. The *MYOD* family consists of four related genes; *MYOD1* (*MYF3*), *MYOG* (*MYF4*, myogenin), *MYF5* and *MYF6*. These genes play a key role in growth and muscle development in farm animals and are therefore considered as candidate genes for meat production and quality related traits. Polymorphisms were detected and confirmed by PCR using 17 primer pairs, sequencing and restriction fragment length polymorphism (RFLP) method. Fourteen SNPs (4 from *MYOD1*, 4 from *MYOG*, 3 from *MYF5* and 3 from *MYF6*) were identified both in intron and exon of these genes, whereas 2 of them were missense mutations. Allele frequencies for g.783G>A and g.1274A>G mutations in *MYOD1*; g.511G>C and g.1111C>G in *MYOG*; g.1911A>G in *MYF5* and g.183T>C in *MYF6* varied significantly among the 7 different cattle breeds. The g.783G>A genotype of *MYOD1* gene was significantly associated with carcass weight ($P < 0.05$) and eye muscle area ($P < 0.001$) in Korean cattle population. The g.1111C>G and g.1911A>G SNPs in *MYOG* and *MYF5* genes, respectively, had significant association with carcass weight ($P < 0.01$) and lean weight ($P < 0.01$). Moreover, the g.1111C>G polymorphism in *MYOG* gene was associated with back fat thickness ($P < 0.001$). In conclusion, the SNPs g.783G>A, g.1111C>G and g.1911A>G of *MYOD* gene family may be useful for marker assisted selection on carcass and meat quality traits in cattle.

Poster 2174

Title: Distinctive patterns of IGF2 and IGF2R expression in Bos indicus, Bos taurus and hybrid fetuses

Presenting Author: Carolyn Fitzsimmons, University of Adelaide, School of Agriculture, Food, & Wine. Roseworthy Campus, Roseworthy, SA 5371 Australia

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Abstract:

We have investigated transcript-specific *IGF2* and *IGF2R* expression in brain, cotyledon, liver and semitendinosus muscle of *B. indicus*, *B. taurus*, and reciprocal cross fetuses (n=40). Real-time qPCR data were analysed (GLM, SPSS) including fixed effects dam breed, sire breed, fetal sex, and covariate dam weight (i.e. four factors) and 3 housekeeping genes. Significant ($P<0.05$) effects on *IGF2* transcripts were similar within each fetal tissue. Muscle *IGF2* exons 2, 6, and 10, were affected by dam weight, and exon 10 by fetus sex. Liver *IGF2* exon 10 was affected by the interaction of sire breed \times dam weight. Cotyledon *IGF2* exon 6 was affected by the interaction of dam breed \times dam weight \times fetus sex; exon 10 by an interaction between all four factors. No effects were significant on exon 10 expression in brain. The expression of *IGF2R* in brain, liver, and cotyledon, but not muscle, displayed dissimilar patterns than *IGF2*. Interactions between sire breed \times fetal sex, and dam breed \times dam weight, were detected in brain. None of the factors affected *IGF2R* expression levels in cotyledon or liver. Dam weight again affected *IGF2R* in muscle. These data demonstrate the complexity of imprinted gene expression in fetal development.

Poster 2175

Title: Partial duplication of the *PRLR* and *SPEF2* genes at the late feathering locus in chicken

Presenting Author: Martin G. Elferink, P.O. Box 338, Wageningen, The Netherlands

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Abstract:

One of the loci responsible of feather development in chickens is called K. The K allele is associated with the late feathering phenotype that causes a retard in emergence of primary and secondary flight feathers. The k+ allele is associated with the early feathering phenotype resulting in the earliest emergence of feathers.

The K locus is located on the Z chromosome and can be used to produce phenotypes that allow distinction between the sexes of chicks at hatch. This sexing method based on differences in rate of feather growth provides a convenient and inexpensive approach.

Previous studies on the organization of the K allele concluded the integration of endogenous retrovirus 21 into one of two large homologous segments located on the Z chromosome of late feathering chickens. In this study, the K locus was investigated with quantitative PCR by examining copy number variations in a total of fourteen markers surrounding the ev21 integration site. The results showed a duplication at the K allele and sequence analysis of the breakpoint junction indicated a tandem duplication of 176,324 basepairs. The tandem duplication of this region results in the partial duplication of two genes; the prolactin receptor and the gene encoding sperm flagellar protein 2.

Poster 2176

Using image analysis in a Jersey – Limousin double backcross to identify candidate genes for fat deposition traits in cattle

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Internal, intramuscular and subcutaneous fat deposition traits were mapped previously in a Limousin-Jersey double backcross cattle herd and several quantitative trait loci (QTL) identified. However, in order to quantify seam fat as a measure of intermuscular fat and to better describe intramuscular or marbling fat, a method was developed to analyse photographs of the *L. dorsi* muscle from the QTL mapping progeny. Using Adobe Photoshop[®] and Matlab[®] software designed for the photographic image analysis, seam fat areas were quantified and new marbling parameters were determined. QTL were then genetically mapped from the image analysis data. Two significant QTL of large effect were found on BTA2 and BTA19 for seam fat. *Myostatin* was an obvious candidate gene for the seam fat QTL on BTA2. Subsequent genotype analyses of the *myostatin* gene indicated that the Limousin F94L *myostatin* DNA variant was responsible for this QTL. For the seam fat QTL on BTA19, several candidate genes have been selected for sequencing to identify polymorphisms for association studies. In addition, the seam fat and marbling parameter measurements are being incorporated into fat deposition models to provide more informative and specific QTL for adipose partitioning.

Poster 2177

Title: Sequence of mitochondrial gene 12SrRNA in *Emys orbicularis orbicularis* population from Poland

Presenting Author: Beata Prusak, Institute of Genetics and Animal Breeding, Polish Academy of Sciences, Jastrzębiec, Poland

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Abstract:

From the last morph metric observations conducted in Europe it results that Polish population of pond turtle is the last one relatively plentiful group of nominative subspecies of *Emys orbicularis orbicularis* (Linnaeus 1758) with presumably two ecological lines existing ("eastern" and "western") differing morphologically and genetically. According the fact that pond turtle is morphologically varied, *Emys orbicularis* is probably the most divided vertebrate species in North Pale arctic with several genetic lines existing in Europe. Aim of the research is to determine the genetic diversity of pond turtle within and between populations from different regions of Poland, to determine reference sequences of mtDNA for populations existing in Poland and to verify hypothesis about distinctiveness of domestic eastern and western *E. orbicularis* populations. From the research, it results that pond turtles from western Poland are genetically closer related to Spanish population than to Polish eastern one. The material covered buccal swabs possessed from turtles from four main regions of the population living-grounds in Poland: Włodawa Lake District, Radom environs, Mazurian Lake District and Wielkopolska-Kujavian Lowland. Polymorphism of mitochondrial 12SrRNA gene was analyzed. Results presented are first step of wider research project covering coding and non-coding regions of pond turtle' mitochondrial genome.

Poster 2178

Title: Disease gene identification in dogs: use of SNP arrays and candidate genes

Presenting Author: Alan N Wilton, School of Biotechnology and Biomolecular Sciences, Ramaciotti Centre for Gene Function Analysis, University of New South Wales, 2052 Sydney, Australia

Other authors (name only):

1. Jeremy R Shearman

Abstract:

We have identified mutations for a storage disease (NCL) which causes nerve degeneration and a neutrophil dysfunction (TNS) that results in high susceptibility to infections. TNS (Trapped Neutrophil Syndrome) is common in border collies on all continents. Both diseases have homologous human diseases. High throughput SNP arrays now make it easier to locate disease genes in inbred dog populations. Whole genome analysis on affected dogs and controls (around 10 of each) could be used in association studies and homozygosity analysis. Diseases where the mutations are identical-by-descent will be homozygous in the region of the disease gene in affecteds. The method is effective in cases where we have already identified the mutation. The application to cerebellar abiotrophy that causes ataxia in kelpies. seems simple in theory but is proving difficult in practice. Reliability of genotype calls and errors in assigning disease phenotypes are possible reasons for the lack of significant associations. A combination of homozygosity analysis and comparative genomics could also be used to identify disease genes from a single affected dog by eliminating large regions of the genome as candidate regions. We are applying this approach to a rare second type of NCL in broder collies.

Poster 2179

Title: Polymorphism and mapping of the porcine inhibitor of differentiation genes, *ID1*, *ID2*, *ID3* and *ID4*

Antonín Stratil¹, Jitka Filkuková¹, Pavel Horák¹, Mario Van Poucke², Heinz Bartenschlager³, Luc J. Peelman², Hermann Geldermann³

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Abstract:

ID proteins are important members of signaling pathways involved in development, cell cycle and tumorigenesis. They are considered negative regulators of MRFs and the responsible genes are therefore candidates for carcass and meat quality traits. Specific PCR primers for the four porcine *ID* genes (*ID1*, *ID2*, *ID3* and *ID4*) were designed and the PCR fragments were comparatively sequenced (Meishan and Pietrain) to verify the genes and identify polymorphisms. The genes were mapped using

radiation hybrid (IMpRH) panel and by linkage analysis in the Hohenheim Meishan x Pietrain F₂ family. The results of RH mapping were: *ID1* – SSC17, closest marker *ENDO3* (21 cR; LOD=11.11); *ID2* – SSC3, closest marker *SW2532* (20 cR; LOD=13.88); *ID3* – SSC6, closest marker *SW709* (8.0 cR; LOD=16.10); *ID4* – SSC7, closest marker *SW1369* (65 cR; LOD=4.64). By linkage analysis only *ID3* and *ID4* genes were mapped (in *ID1* and *ID2* no suitable polymorphism was found). *ID3* was mapped to SSC6 (...*FABP3* – 3.5 cM – *ID3* – 14.2 cM – *S0146*...) and *ID4* to SSC7 (...*S0064* – 27.0 cM – *ID4* – 12.1 cM – *CYP21A2*...). The *ID4* gene maps to chromosome region with numerous QTL for carcass and meat quality traits. For this locus and for *ID3* significant associations between genotypes and a series of traits were found (GLM procedure). (Supported by the Czech Science Foundation 523/06/1302)

Poster 2180

Title: GENETIC RELATIONSHIP BETWEEN ANDALUSIAN AND AMERICAN HORSE BREEDS

Presenting Author: Alexandra Gómez Tarazona
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Abstract:

Microsatellite polymorphisms were used to assess both the genetic diversity and the relationship between Spanish, South American and American Paso Fino horse breeds. Allelic characterization was made with 35 molecular microsatellite markers in order to do further population genetics studies. An amount of 387 animals unrelated registered representing each breed; Andalusian, Colombian horse, Peruvian and Paso Fino American horse breeds were analyzed. American horse populations displayed a relatively high level of genetic variation as estimated by allelic diversity and heterozygosity, whereas the Spanish breed showed reduced levels of genetic diversity. Only six percent of the total genetic variability could be attributed to differences among breeds ($F_{ST}=0.06$). American breeds clearly formed a separate cluster from the Andalusian breed. In addition, the molecular variance analysis indicated that 5.4% of variance could be explained by differences between Andalusian and New world breeds.

Poster 2181

Title: Single Nucleotide Polymorphisms in the ovine MHC class II region

Presenting Author: Chee Yang Lee, Curtin University, School of Biomedical Sciences, GPO Box U1987, Perth, Australia 6845

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Abstract:

There is a lack of detailed knowledge on the structure and sequence variation of the ovine Major Histocompatibility Complex (MHC). The ovine class II region is divided into two distinct regions (class IIa and IIb) separated by a region of non-MHC genes and was formed by chromosomal inversion. This structural rearrangement appears to be unique to ruminants. The existence of other chromosomal inversions within the ovine MHC region has yet to be determined, although there is evidence of small chromosomal rearrangements within the class III region. Six Bacterial Artificial Chromosomes from the CHORI 243 sheep BAC library have been identified to contain MHC class IIa and IIb DNA. Shotgun-cloning was performed to identify distinct plasmid isolates. From these sequences, PCR primers were designed to amplify selected regions across the MHC class IIa and IIb in animals from two family groups of Merino sheep. Several single nucleotide polymorphisms (SNP) were identified. A map of the positions of these SNPs in the class IIa and IIb regions is presented. This information can then be used to develop haplotype maps of the sheep MHC. A Hap-Map of the sheep MHC will assist in identifying correlations between parasite resistance traits and MHC.

Poster 2182

Title: Quantitative Trait Loci for Short and medium even chain fatty acids *

Presenting Author: W.M. Stoop, Animal Breeding and Genomics Centre, Wageningen University, P.O. box 338, Wageningen, The Netherlands

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Abstract:

A genome scan was performed to identify quantitative trait loci (QTL) for short and medium chain FA*. Seven half-sib families with in total 849 cows and their 7 sires were genotyped for 1,379 SNP. Phenotypes of the cows were pre-corrected for systematic effects. The daughter design was used for a QTL program with multi-marker interval mapping in a weighted across-family QTL regression. Thresholds and confidence intervals were set using 10,000 permutations and 1,000 bootstraps. The genetic map measured 2,831 cM and had an average information content of 0.83. The results showed significant QTL ($P_{\text{genome}} < 0.05$) for 15 traits at 4 distinct chromosomal regions. Two of these QTL regions mapped to the approximate locations of the DGAT1 and the SCD1 gene. Correcting phenotypes for these two major genes resulted in complete disappearance of the linkage previously found in the two regions. Furthermore, suggestive QTL ($P_{\text{chromosome}} < 0.05$) were found for 28 traits. The identified QTL may be a first step in understanding physiology of milk fat synthesis, as well as providing the basis for the development of a genomic selection tool to decrease the proportion of the unfavorable fatty acids c14:0 and c16:0. *c4:0, c6:0, c8:0, c10:0, c12:0, c14:0, c16:0, c10:1, c12:1, c14:1, c16:1, c10index, c12index, c14index, c16index

Poster 2183**Title: Identification of differentially expressed genes in adipocyte differentiation using Affymetrix Bovine Genome Array**

Presenting Author: Seong-Lan Yu, Department of Animal Science, College of Agriculture and Life Science, Chungnam national university, 220, Gung Dong, Yuseong Gu, Daejeon 305-764, South Korea.

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Abstract:

Adipocytes are differentiated from preadipocytes and have large capacity for storing fats inside cells. In cattle, intramuscular fat (IMF) content is one of the major determinants for meat quality and also highly affects market prices, especially in Japan and Korea. In order to profiling differentially expressed genes between intramuscular fibroblast-like cells (preadipocyte cells) and their differentiated adipocytes, we have been established intramuscular fibroblast-like cells from *M. longissimus thoracis* in

Korean cattle (Hanwoo). The differentially expressed genes were selected by comparing these two cells using commercially available 23k Affymetrix Bovine Genome Array. The data analyses indicated that 206 array elements were up or down regulated. Of these, 38 and 57 known genes were up and down regulated, respectively, in adipocytes using both 2-fold difference and Welch's T test as cut-off points. The differentially expressed genes identified in this study can be used as good candidate markers for improving meat quality traits, especially IMF content, in cattle.

Poster 2184**Title: Fine-mapping of a pea comb locus on chicken chromosome 1**

Presenting Author: Shuji Sato, National Livestock Breeding Center, Nishigo, Fukushima, 961-8511, Japan

Other authors (name only):

1. Shinichi Sato
2. Tsuyoshi Otake
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5. Eiji Kobayashi

Abstract:

Although the Japanese game fowl (Shamo) is characterized by the pea comb (*P/P* or *P/p*), chickens with the single comb (*p/p*) often appear due to the presence of carrier (*P/p*). Our objective of this study was to narrow the genome region affecting the comb type to facilitate candidate gene identification. We used two populations for this purpose. First, an F2 resource population was generated by crossing Shamo and White Plymouth Rock (single comb, *p/p*). The number of F2 offspring was 265 from F1 intercross between eight males and 57 females. The comb type was segregating with the ratio of pea : single = 3:1 in F2 generation and we mapped a pea comb locus to a chromosomal region on *Gallus gallus* chromosome 1, flanked by microsatellite markers *MCW0019*, *MCW0112* and *ABR521*. The second population was five generations population derived from cross between Shamo and Rhode Island Red (single comb, *p/p*), which was genotyped for additional polymorphic SNP and microsatellite markers within this region developed from Chicken draft sequences. To close some gaps in Chicken draft sequences, we constructed a BAC contig and sequenced it by the shot-gun sequencing. Chickens selected from some pedigree in these populations are grouped according to the inheritance of P or p haplotype at this locus constructed by additional markers. Finally, this locus was fine-mapped to roughly 50kb based on the association of the haplotypes and comb types. Chicken draft sequences suggest that *sex determining region Y*

-*box 5 (SOX5)* gene is the most likely candidate gene for the pea comb locus.

Poster 2185

Title: Functional analysis of the *c.1892+19 T>C* SNP of the *PPARGC1A* gene which is associated with variation in milk fat yield

Presenting Author: Rainer Fürbass, Research Institute for the Biology of Farm Animals (FBN), 18196 Dummerstorf, Germany

Other authors (name only):

1. Christa Kühn
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Abstract:

Recently, we identified the bovine peroxysome proliferator-activated receptor- γ coactivator-1 α (*PPARGC1A*) gene as a plausible positional and functional candidate gene for a previously described QTL for milk fat yield on BTA6. A significant association between a SNP within intron 9 of *PPARGC1A*, *c.1892+19 T>C*, and milk fat yield was observed in a major dairy cattle population, indicating that the *PPARGC1A* gene could be involved in genetic variation underlying the QTL for milk fat synthesis on BTA6. A possible functional role of this SNP for *PPARGC1A* expression was investigated by electrophoretic mobility shift assays (EMSA) using allele-specific probes and nuclear protein extracts from various embryonic and adult bovine tissues. Probes including the C-allele of the SNP yielded a specific binding complex with all protein extracts. In contrast, no complex was observed with T-allele-specific probes. Hence, the C-allele represents a new binding site for a transcription factor. This finding supports the hypothesis, that the trait-associated SNP may have a functional effect. By competition and supershift experiments it could be demonstrated that the sequence motif is a target of the transcription factor Sp1. Currently, possible effects of the SNP on gene expression are being investigated by reporter gene analyses in various cell culture models.

Poster 2186

Title: Expression of antimicrobial peptides in different localisations of the bovine udder

Presenting Author: Jens Tetens, Institute of Animal Breeding and Husbandry, Olshausenstr. 40, D-24098 Kiel

Other authors (name only):

1. Judith Friedrich
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Abstract:

Antimicrobial peptides (AMP) are key effectors of innate immunity and play an important role in local host defence. In order to enhance the knowledge about the role of bovine AMPs in the protection against udder infection we analysed the gene expression level of the six bovine AMPs *LAP*, *TAP*, *DEFB1 (EBD)*, *BNBD4/10*, and *S100A7* (psoriasin) in five localisations of the bovine udder, namely the streak canal, the rosette of Fürstenberg, the cisterna, the udder lymph node, and udder parenchyma. We analysed samples from nine healthy cows applying a quantitative real-time PCR approach. Consistent with the role of AMPs in local host defence, predominantly taking place at the sites of invasion, we found no or marginal expression of all genes in parenchyma. The highest expression levels for *LAP* and *TAP* were detected in the lymph node with remarkably high values for *LAP*. The neutrophil β -defensins *BNBD4/10* were predominantly expressed in the lymph node with *BNBD10* also showing remarkably expression in the streak canal and cisterna. The *S100A7* (psoriasin) gene which has been shown to be active against *E. coli* was found highly expressed in the streak canal and the cisterna, indicating a major role in the defence against coliform mastitis.

Poster 2187

Title: Characterization of BLV (Bovine leukemia virus) envelope gene in Holstein from Korea

Presenting Author: H. J. Jeong, Department of Animal Science, College of Agriculture and Life Science, Chungnam National University, 220, Gung Dong, Yuseong Gu, Daejeon 305-764, South Korea.

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Abstract:

Bovine leukemia virus (BLV) is an oncogenic virus predominantly present in cattle. It belongs to *Delta-retrovirus* in the family Retroviridae. The genomic structure is similar to human T-cell leukemia virus. BLV has envelope proteins which contain functional domains that determine its infectivity. We conducted a nested polymerase chain reaction (PCR) for amplification of the envelope gene which can be act for the serological status in the host. To examine the distribution of BLV variants, sequencing of the PCR product was performed. The nucleotide sequences were compared with other previously characterized BLV sequence from different serological areas. We

compared sequences of Korea Holstein population and BLV env sequences available at GenBank. The phylogenetic tree showed three different clusters and most samples from Korea population formed one cluster similar in USA Holstein sequences. Further research about BLV give some clues in order to make general strategy for improving the disease.

Poster 2188

Title: Associations between polymorphisms in the cholecystokinin B receptor gene and behavioural traits in beef cattle

Presenting Author: K. Glenske, Department of Animal Breeding and Genetics, Justus Liebig University, Giessen, Germany

Other authors (name only):

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Abstract:

The importance of temperament and docility in successful management of beef cattle is already recognized as well as its genetic background. One candidate gene is the cholecystokinin B receptor (*CCKBR*), which plays a role in regulation of anxiety. The molecular analysis of two regions of bovine *CCKBR*, including exon III - exon IV and the 3'UTR, resulted in two SNPs. The SNP in intron 3 (C/T), which belongs to a binding site for a transcription factor, was genotyped by a PCR-RFLP with *Bse*NI; the SNP (C/T) in the 3'UTR by a tetra-primer ARMS-PCR.

Behavioural traits related to temperament were scored in 962 calves (born in 1998-2001) from progeny groups of German Simmental (n = 8) and German Angus beef cattle (n = 6) in three standardized tests at different ages (tethering test, 3 weeks; weighing test, 3 month; separation- and restraint-test, 7-8 month). Behaviour scores were given to the animals. The association analysis was done for each SNP with a mixed linear model within breeds, including the four years and genotype as fixed effects, age at test as covariate and sire as random effect. No significant associations between the SNPs and the behavioural scores could be demonstrated.

Poster 2189

Title: QTL mapping of Japanese Black cattle using a family structure combining seven paternal half-sib families.

Presenting Author: TOSHIO WATANABE, Shirakawa Institute of Animal Genetics, 1 Odakurahara Odakura Nishigo Fukushima, Japan

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Abstract:

For QTL analyses of livestock, it often takes long time to collect enough number of half-sib offspring to achieve reasonable statistical power. In this study, we examined plausibility to map QTL of a grandsire by combining several small families to construct a large three-generation family of Japanese Black cattle.

We combined genotype data of seven half-sib families, where all the sires are sons of grandsire A, containing 44-103 offspring (426 in total) and genotyped with 185-436 microsatellites. Transmission of grandsire A's haplotype to a sire was traced using 263 microsatellites. Three QTL-models were assumed: superior QTL allele is present (1) on the first haplotype of the grandsire, (2) on the second haplotype, and (3) on both. Probabilities to succeed the grandsire's haplotype were linearly regressed on phenotypic values of 3rd generation offspring. Sixteen QTL were detected for beef marbling standard score, carcass weight, longissimus muscle area, and rib thickness ($P < 0.05$, chromosome-wise significance level). Of sixteen, seven were newly mapped and nine were overlapped with the regions previously mapped in Japanese Black. We ensured that marbling QTL on BTA19 segregated within A's 400 offspring. Combining small families will provide more opportunities to map novel QTL in livestock.

Poster 2190

Title: Two novel single nucleotide polymorphism markers of *MC1R* gene in Thai Native White Lamphun cattle

SUPAMIT MEKCHAY^{1,2}

¹Department of Animal Science, Faculty of Agriculture, Chiang Mai University, Chiang Mai, 50200 Thailand, ²Center of Agricultural Biotechnology, Faculty of Agriculture, Chiang Mai University, Chiang Mai, 50200 Thailand

Abstract

Melanocortin receptor 1 (*MC1R*) gene is an important gene in the regulation of pigment synthesis and responsible for coat color in mammals. The polymorphism of *MC1R* gene in *Bos taurus* has been characterized as four alleles (E^D , E^+ , E^1 and e). In this study was carried out to identify single nucleotide polymorphism (SNP) of *MC1R* gene in Thai native

cattle (*Bos indicus*). The coding region of *MC1R* gene in Thai native cattle namely White Lamphun cattle (white coat color) was sequenced and compared with Holstein and Charolais breeds. The *MC1R* sequence of White Lamphun cattle has high homology with the *Bos taurus* (99% identity). Four polymorphic sites were found in *MC1R* gene of the White Lamphun at position 296, 416, 663 and 725 bp of open reading frame. Out of these, three SNPs were identified as missense mutation, consisting of (1) a single base substitution (T296C) resulting in an amino acid change from leucine to proline (E⁺ allele) (2) a single base substitution (C416T) leading to an amino acid change from alanine to valine and (3) a single base substitution (A725C) leading to an amino acid change from asparagine to threonine. Moreover, a non-synonymous mutation was located at position A663C of bovine *MC1R* coding region. Based on this observation, two novel SNPs at position 416 and 663) were found only White Lamphun cattle breed. This result indicated that the *MC1R* gene of White Lamphun cattle breed was E⁺ allele and these two novel SNPs may be used as allele specific markers for the White Lamphun cattle breed.

Poster 2191

Title: Expression of Non-Classical MHC Class I Genes by Bovine Embryos

CJ Davies¹, A Wilhelm¹, BR Sessions¹, P Parasar¹, Y Li², EE Schmidt², KL White¹

¹Department of Animal, Dairy and Veterinary Sciences & Center for Integrated BioSystems, Utah State University, Logan, UT, USA; ²Veterinary Molecular Biology, Montana State University, Bozeman, MT, USA.

Abstract:

In human pregnancy trophoblast cells express the non-classical MHC class I (MHC-I) proteins HLA-G and HLA-E, which are important immunoregulatory factors. Previously, we showed that during the third trimester bovine trophoblast cells expressed four non-classical MHC-I genes, BoLA-NC1 through BoLA-NC4 (Davies et al. AJRI 55:188-200, 2006). Thus far, no one has studied expression of bovine non-classical MHC-I genes early in pregnancy. We have cultured bovine *in vitro* fertilized embryos to gestational age 30-37 days in either DMEM/F12 or bES medium plus 15% serum and used these embryos for analysis of MHC-I gene expression. Following RNA extraction from individual or pooled embryos with TRIzol Plus (Invitrogen), RNA was reverse transcribed with Superscript III (Invitrogen), cDNA amplified by PCR with MHC-I primers that amplify alleles encoded at all classical and non-classical MHC-I loci, cloned in the pCR4 vector (Invitrogen), and 40-48 clones

sequenced by cycle sequencing using BigDye V3.1 and an ABI PRISM 3730 DNA Analyzer (Applied Biosystems). Embryos expressed both classical and non-classical MHC-I genes. Expression of non-classical MHC-I genes suggests that bovine trophoblast cells express these genes around the time of placental attachment, which occurs about day 30 of pregnancy. Expression of classical MHC-I genes could either be by trophoblast cells, which are the predominant cell type in these embryos, or by endothelial or other cells. For one pool of four day 37 embryos 33% of the pCR4 clones contained BoLA-NC1 sequences. BoLA-NC1 transcripts exhibit differential splicing involving intron 4 and exon 5, which encodes the transmembrane domain. The splicing patterns are similar to patterns seen with HLA-G. Consequently, BoLA-NC1 may be a functional homolog of HLA-G.

Poster 2192

Title: Predicting genetic merit for production and health traits in dairy cattle using genome wide selection and high density SNP screens

Presenting Author: Herman W. RAADSMA, Reprogen, The University of Sydney, Australia

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Abstract:

A genome wide selection (GWS) platform was developed for prediction of genetic merit in dairy cattle. The critical components of the GWS platform included a genome wide SNP analysis assay representing between 15,000 and 70,000 SNP's and 1945 progeny tested Holstein Friesian sires with EBV for 42 lactation performance traits. Two methods for complexity reduction and feature selection were used - Partial Least Squares (PLS) and regression using a genetic algorithm (GAR), to find optimal solutions of EBVs against SNP information. Extensive internal cross validation was used to find the best predictive models followed by external validation without direct use of pedigree information or SNP location. Derived Molecular Breeding Values (MBV) using either all or a fraction of the available SNP information were shown to have high predictive value for genetic merit ($r=0.65-0.87$ with EBV) when test subsets were randomly drawn from the data (mirror prediction). A general loss in accuracy was seen when genetic merit

was predicted in test co-horts of young bulls which were not part of the training set. However MBV was more accurate in predicting genetic merit than pedigree based EBV alone at time of birth for young bulls. PLS was computationally more efficient than GAR.

Poster 2193

Title: Positional cloning of the canine hairless mutation reveals an essential role for the *FOXI3* gene in ectodermal development

Presenting Author: Cord Drögemüller, Institute of Genetics, University of Berne, Switzerland

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Abstract:

Chinese Crested, Mexican and Peruvian hairless dogs are characterized by a very sparse hair coat and deficient and abnormally shaped teeth, a phenotype summarized under the classification canine ectodermal dysplasia (CED). CED is inherited as a monogenic, autosomal, semi-dominant trait. Heterozygous dogs exhibit the CED phenotype and homozygous mutant animals die during embryonic development. We applied a whole-genome SNP association mapping approach using DNA from 20 hairless and 20 coated Chinese Crested dogs and the canine Affymetrix v2 SNP genotyping microarray. This analysis identified a single region of strong association on CFA17. This region contains a three SNP haplotype with complete phenotype concordance. Subsequent linkage disequilibrium fine mapping across three breeds narrowed the critical interval to 102 kb. This interval contains only one gene, a previously uncharacterized member of the forkhead box transcription factor family (*FOXI3*). Whole mount *in situ* hybridization of mouse embryos demonstrated a specific expression of *Foxi3* in hair and whisker follicles and in developing teeth. Mutation analysis revealed a frameshift mutation within the *FOXI3* coding sequence that co-segregates with the CED phenotype in all three investigated dog breeds. Thus, using a two-stage, genome-wide mapping approach, we have efficiently identified a novel gene involved in ectodermal development.

Poster 2194

Title: High resolution chicken radiation hybrid maps using the Illumina™ technology

Presenting Author: Alain Vignal, UMR génétique Cellulaire, INRA Chemin de Borde-Rouge, Auzeville BP 52627, 31326 CASTANET-TOLOSAN CEDEX, FRANCE

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Abstract:

The chicken genome sequence assembly is composed of more than 1 Gb of sequence covering almost all chromosomes, with the exception of a few microchromosomes. To confirm the existing assembly and to study the missing microchromosomes, we are developing high density radiation hybrid (RH) maps. Markers from the ChickRH database and 9212 markers genotyped on an Illumina platform were used and the construction of framework maps was done using the Carthagene software. Alignment of the maps to the sequence indicates a good agreement with only a few minor discrepancies, such as small inversions of a very limited number of markers, which can suggest an ordering error either in the sequence or in the RH maps. The resolution of the framework maps is 26 kb/cR with an average spacing of 410 kb between markers. Additional linkage groups could correspond to microchromosomes with very little genomic sequence information. These results indicate a good quality of the chicken sequence and confirm the possibility of using the Illumina™ technology for RH mapping, despite the fact that it was primarily designed for SNP genotyping.

Poster 2195

Title *PGK1* endogenous gene shows expression differences in porcine tissues.

Presenting Author: Ana Isabel Fernández. Dpto. Mejora Genética Animal. INIA. Ctra. Coruña Km7.5 28040 Madrid. Spain

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Abstract:

Gene expression is quantified by normalization to the level of one or more reference genes. However different conditions might influence their expression, which could lead to errors in the interpretation of results. The *Phosphoglycerate kinase 1 (PGK1)* gene codes for a protein that participates in glucose metabolism and it is usually considered as reference gene. We conducted a gene expression analysis to identify hepatic gene expression patterns in male and female Iberian pigs fed with high and low feeding levels using microarrays. This study showed that *PGK1* expression was conditional on feeding level. Here we have performed the validation of this result by qPCR in a larger number of liver samples (n=17) using as endogenous *GADPH* and *BM2* genes and geNorm software. Statistical analysis model of the expression differences by qPCR included the sex, feeding level and their interaction effects. Statistical results show that *PGK1* liver gene expression depends on sex (p= 0.0003; females=1.5x males) and feeding level (p=0.0251; high= 1.23x low), and the interaction sex x feeding level is also significant (p=0.0215). In addition, we have detected similar effects in porcine *psaos major* muscle. These results would invalidate *PGK1* as reference gene for these tissues in our pig population.

Poster 2196

Title: Detection of signature of selection among 14 cattle European and African populations in genomic regions associated to trypanotolerance

Presenting Author: Guiguigbaza Kossigan DAYO, CIRDES, BP454 Bobo Dioulasso, Burkina Faso

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Abstract:

Breeding of indigenous African taurine cattle tolerant to African trypanosomoses remains a straightforward alternative to control these diseases. Previous studies have identified QTL regions underlying trypanotolerance in experimental cross. In order to validate and refine these QTL in different cattle populations, we looked for signature of selection in surrounding regions using a panel of 92 microsatellite markers genotyped on 20-50 individuals belonging to 14 different cattle breeds. Breeds were chosen according to their history and their known status

regarding tolerance to trypanosomoses. Hence, 4 West African trypanotolerant taurine breeds and 10 European and African trypanosusceptible cattle breeds were chosen. Two F_{ST} based tests and tests based on heterozygosity, were considered to analyse the data. Among 92 microsatellite loci, two were significantly less variable and lied out in heterozygosity based tests when comparing tolerant and susceptible populations. These results suggest that these loci are likely under selection or linked to local adaptative genes. Results of the two F_{ST} based tests were also presented and we discussed differences obtained with all methods. From a practical perspective, the functional testing of the polymorphisms of genes mapped in outliers regions may be used to determine how their genetic variation modulates susceptibility to trypanosomoses.

Poster 2197

Title: TG and DGAT have no influence on intramuscular fat content in Bavarian Simmental bulls

Presenting Author: Susanne Kämmerer, Bavarian State Research Center for Agriculture, Department Quality Assurance and Analysis, 85586 Grub/ Germany

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Abstract:

Research has demonstrated that single nucleotide polymorphisms in the thyroglobulin gene (TG) and in the gene encoding diacylglycerol O-acyltransferase (DGAT 1) are associated with intramuscular fat content (IMF) or rather with the assessed trait marbling in cattle. IMF being an important beef quality trait, genotyping of breeding bulls is widely used. Nevertheless, it is unknown whether these associations are useful for typical breeds and production conditions in Germany. In the present study the associations of an SNP in the 5' promotor region of TG and a lysine/ alanine polymorphism of DGAT1 with the fat content of *m. long. dorsi* in 100 Bavarian Simmental bulls was investigated. Bulls were chosen from 2.110 animals raised under standardized conditions of the Bavarian progeny test on station. Variance components and breeding values for IMF were estimated with DMU and PEST, respectively. Two groups were selected, each consisting of 50 bulls with the highest (Ø 0,92%) and lowest (Ø -0,60%) IMF breeding values, respectively. Neither of the markers showed significant differences in allele- or genotype-frequencies in both groups.

These results suggest that the TG and DGAT1 markers are no efficient predictors of IMF content in Bavarian Simmental bulls.

Poster 2198

Title: Maternal dietary methionine supplementation modulates foetal hepatic expression profiles

Presenting Author: Klaus Wimmers, Research Institute for the Biology of Farm Animals (FBN), Research Unit 'Molecular Biology', 18196 Dummerstorf, Germany

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Abstract:

Impact of maternal nutrition during pregnancy on gene expression profiles and phenotypes of offspring was shown experimentally and in epidemiological studies in model animals and humans. The aim of the study is demonstrating of foetal programming in pigs, listing of genes sensitive to foetal programming and evidencing and quantifying of the role of DNA-methylation in this phenomenon. Therefore, 36 sows were fed methionine supplemented ('MET') vs. control ('CON') gestation diets. Liver samples of fetuses of three developmental stages (d35, d63, d91 p.c.) were subsequently monitored for diet-dependent differential gene expression by using Affymetrix micorarrays. Statistical and bioinformatics evaluation of the expression profiles revealed largest effect of gestation diets at late foetal stages (91 dpc) when more than 1600 transcripts were differentially regulated. Functional annotation clustering highlighted the GO terms biological quality, cellular and anatomical structure morphogenesis, and negative regulation of metabolic processes. Interestingly, members of the IGF axis were found being diet-dependent regulated that play a key role in prenatal growth and are sensitive to regulation by DNA-methylation. Quantification of the DNA methylation at these loci will shed light on the role of this epigenetic mechanism as the molecular basis of genotype-environment (diet) interactions.

Poster 2199

Title: Frequency of *IGF2* mutation in Polish breeds of pigs.

Presenting Author: Barbara Rejduch, National Research Institute of Animal Production Department

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Abstract:

IGF2 is one of the genes that control muscle development. Using assay based on Real Time PCR for *IGF2* genotyping, we evaluated frequency of *IGF2* mutation in Polish breeds and assessed the usefulness of selection for this marker in Polish pig industry.

In total 400 gilts and 35 wild boars (WB) were genotyped. Pigs represented seven breeds: Puławska (Pł), Landrace (L), Large White (LW), Duroc (D), Hampshire (H), Pietrain (P) and Line 990 (L.990).

Previous reports shown that breeds, subjected to strong selection for lean meat have low frequency of G allele. Our study, performed on much higher number of animals, are in a great agreement with earlier findings. No G allele was observed in Duroc. In L. 990, Hampshire and Pietrain frequency of G allele did not exceed 11%. Higher frequency (approximately 25%) of G allele was noted in Landrace and Large White. The highest frequency of G allele (36%) was observed in Puławska. Similarly to previous studies, no A allele was observed in wild boar.

Poster 2200

Title: Characterization of the ovine multicopy ribosomal protein SA gene family

Presenting Author: Mario Van Poucke Heidestraat 19 B-9820 Merelbeke Belgium

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- 2 Ane Marcos-Carcavilla
- 3 Karine Hugot
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- 5 Luc Peelman

Abstract:

Transmissible spongiform encephalopathies (TSEs) are fatal, neurodegenerative diseases occurring in man (e.g. Creutzfeldt-Jakob) and animals (e.g. BSE and scrapie). They are caused by the accumulation of PrP^{Sc}, the alternatively folded isoform of the cellular protein PrP^C, which is encoded by the *PRNP* gene. Several other genes are thought to play a role in TSE pathogenesis. This is the case for the ribosomal protein SA gene (*RPSA*), previously named 37-kDa laminin receptor precursor (LRP)/67-kDa laminin receptor (LR), which not only acts as a receptor for

both PrP^C and Prp^{Sc}, but is also involved in the propagation of prion diseases. *RPSA* is a member of a complex multicopy gene family, generally presenting several pseudogenes, which has been described in several species. We studied the complex multicopy *RPSA* gene family in sheep by screening an ovine BAC library with different primers designed on the ovine *RPSA* mRNA sequence. Fifty-one BAC clones were positive. After content mapping and construction of mini-contigs, preliminary results suggest at least 8 different genes in the ovine *RPSA* gene family.

Poster 2201

Title: The role of MHC class II genes in nematode infection of lambs.

Presenting Author: Stear MJ University of Glasgow, Bearsden Road, Glasgow G61 1QH
Conference registration number of Presenting Author: ...0107

1. Murphy L.

Abstract:

Current methods for controlling nematodes are threatened by the evolution of drug resistance. Selective breeding of nematode-resistant animals represents an attractive alternative. In the UK, essentially all grazing sheep are exposed to a mixed, predominantly *Teladorsagia circumcincta* infection. Faecal nematode egg counts are widely used to classify lambs but the use of genetic markers would simplify the identification of superior animals. One genetic marker is the *DRB1* locus in the class II region of the major histocompatibility complex. The G2 allele has been found to be associated with resistance in both Scottish Blackface and Suffolk lambs. The mechanism of gene action appears to be the IgE-mediated prevention of nematode establishment and possibly survival. Targets of the IgE response have been identified by 2-dimensional gel electrophoresis followed by tandem mass spectrometry. Individuals with different MHC alleles respond differentially to specific parasite molecules.

Poster 2202

Title: Association in heavy pigs of CTSE and CAST polymorphisms with productive and meat quality traits

Presenting Author: Estefânia Alves, INIA, Dept^o Mejora Genética Animal, 28040 Madrid - Spain

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Abstract:

Cathepsin F (CTSF) is a lysosomal enzyme involved in intracellular nonselective protein degradation and Calpastatin (CAST) is the endogenous inhibitor of Ca²⁺-dependent proteases, μ - and m-calpain, which seem to play an important role in the breakdown of muscle structural proteins. Besides its task in muscle protein degradation, Calpastatin is also implicated in muscle development and it has been reported to inhibit fusion of rat and mouse myoblasts into myotubes. Effects of *CTSF* and *CAST* genes on several productive and meat quality traits were previously reported, and new association analysis have been performed in the present study. A missense mutation (Glu>Asp) on *CTSF* exon 9 and two mutations on *CAST*, one silent and one missense (Arg>Lys), were genotyped on 447 animals of a hybrid commercial line, by two pyrosequencing protocols. The joint analysis of both genes showed significant associations between *CTSF* and live weight, backfat thickness, loin weight as well as water content and Minolta colour parameters. The two *CAST* polymorphisms resulted completely linked and showed significant associations with backfat thickness (with additive and dominance effects), shoulder weight and the percentage of stearic fatty-acid in subcutaneous fat. Analysis revealed no statistical interaction between both genes.

Poster 2203

Title: The porcine ACTC1 gene – structure, polymorphism, mapping and expression

Pavel Horák¹, Antonín Stratil¹, Aleš Knoll², Karel Bílek², Mario Van Poucke³, Luc J. Peelman³

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³Faculty of Veterinary Medicine, Ghent University, Merelbeke, Belgium.

Abstract:

The *ACTC1* gene (actin alpha cardiac muscle 1) belongs to the alpha actin gene family. Alpha actins are a major constituent of the contractile apparatus and are involved in various types of cell motility. A partial cDNA (~1200 bp) of the porcine *ACTC1* gene was identified in the subtracted foetal hind limb muscle cDNA library (44 days of gestation; using adult biceps femoris cDNA as the driver). Specific PCR-primers were designed for amplification and comparative sequencing (Meishan and Pietrain breeds) of corresponding segments of the gene. The whole gene

structure was completed using primer-walking approach applied on a porcine genomic BAC clone isolated with the *ACTC1* primers. The gene is approx. 8 kb long and consists of 7 exons spanning 4163 bp, 1134 bp of which is the coding sequence. A G>A substitution was identified in intron 1. The gene was mapped using radiation hybrid panel (IMpRH) on chromosome 1 with *SW65* as closest marker (41 cR; LOD = 7.73). By real-time RT-PCR, the gene was proven to be significantly overexpressed in porcine foetal skeletal muscles compared to adult ones. The *ACTC1* gene is expected to be involved in muscle development and growth in pigs. (Supported by the Czech Science Foundation 523/06/1302)

Poster 2204

Title: Determination of *IFNG*, *IL2*, *IL4*, and *GATA3* single nucleotide polymorphisms in New Hampshire and White Leghorn chicken and analysis of association with worm burden after artificial infection with *Heterakis gallinarum*

Presenting Author: G. Lühken, Department of Animal Breeding and Genetics, Justus Liebig University, Giessen, Germany

Other authors (name only):

1. M. Gaily
2. S. Weigend
3. H. Brandt
4. G. Erhardt

Abstract:

There is much evidence in chicken, similarly to mammalian species, that antihelminthic worm reactions are driven by an increase of type 2 cytokines and a decline of type 1 cytokines.

The 5'-flanking region of the type 1 cytokine genes *IFNG* and *IL2*, the type 2 cytokine gene *IL4* and the type 2 immune response directing transcription factor gene *GATA3* were analysed for the occurrence of polymorphisms in the two layer lines New Hampshire and White Leghorn. Novel single nucleotide polymorphisms (SNPs) were identified in *IL4* and *GATA3*. For each of the four genes, a SNP was genotyped by PCR-RFLP analysis in samples of New Hampshire (n = 146) and White Leghorn (n = 263) chicken where the worm burden had been determined eight weeks after artificial infection with 100 embryonated *Heterakis gallinarum* eggs. White Leghorn showed a significantly higher number of worms when compared with New Hampshire. Genotyping results of *IFNG* and *GATA3* in New Hampshire were not used for statistical analysis because the two SNPs were nearly monomorphic in this line. Restricted maximum likelihood analysis with fixed effects of line, sex and genotype revealed no

association of any of the tested SNP with the worm burden.

Poster 2205

Title: Genetic characterization of the Bracco Italiano (Italian Hound) breed: first results on 22 STRs from the ISAG Canine Comparison Test

ROBERTA CIAMPOLINI¹, FRANCESCA CECCH¹, ASSUNTA BRAMANTE², FABIO CASETTI³, BRUNO DE SANCTIS⁴, SILVANO PRESCIUTTINI¹

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Abstract:

The Società Amatori Bracco Italiano is financing a project aimed at investigating the demographic, genetic and genealogical structure of this old breed. We show an assessment of the genetic variability for 22 STRs typed in a sample of 33 unrelated Italian hounds ("Bracchi") and a sample of 43 dogs from other breeds ("Other dogs"). Individual codes of all subjects were traced in the breed database (20,499 animals), so that the coefficient of relationship was < 0.2 with any other animal.

Three multiplexes were worked out, which allowed analyzing 22 STR markers from the panels recommended for the 2006 and 2008 ISAG canine comparison test. Allele size in bp was determined using the comparison-test reference samples as anchor values. Data were analyzed using ARLEQUIN 3.11. The number of different alleles ranged 3 to 8 (mean 5.2) in the "Bracchi", compared with 6-13 (mean 9.5) in the "Other dogs", whereas the expected heterozygosity ranged 0.41-0.81 (mean 0.65) compared with 0.67-0.89 (mean 0.82). Interestingly, the difference between the observed and expected heterozygosities was lower in the "Bracchi" than in the "Other dogs" (0.65 - 0.58 = 0.07 vs. 0.82 - 0.62 = 0.20), despite the absolute values were lower in the "Bracchi".

Poster 2206

Title: MicroRNA profiles of the fetal pig during skeletal muscle development

Presenting Author: Tara G. McDaneld, USDA/ARS Meat Animal Research Center, Clay Center, NE, 68901, USA

Other authors:

1. Ralph T. Wiedmann
2. Jeremy R Miles
3. Tim P.L. Smith

Abstract:

MicroRNA (miR) are a class of small RNAs that regulate gene expression. To evaluate the role of miR in skeletal muscle growth of the fetal pig, global miR expression was measured in skeletal muscle at three stages of growth including: primary fiber development (d 60), secondary fiber development (d 90), and parturition (d 105). Adult muscle was also evaluated to compare muscle at a mature stage of growth (adult) to rapidly growing muscle (fetal). MicroRNA clone libraries identified a total of 95 miR matching sequences in the database with 3 sequences representing novel miR. Muscle-specific miR-206 was the highest expressed miR at all time points evaluated. Additionally, muscle-specific miR-1 was moderately expressed throughout fetal development with highest expression at d 105. Differential expression of miR at different stages of fetal development was also observed. MiR-432 was moderately expressed during early fetal development at d 60 compared to d 90 and d 105, while miR-424 and miR-126 expression increased during d 90 and d 105, respectively. These data are the first to evaluate miR expression at specific stages of fetal skeletal muscle growth in swine and identify novel miR that may play a vital role in skeletal muscle development and growth.

Poster 2207

Title: The porcine arthrogryposis multiplex congenita (AMC) candidate gene *CNTN1*

Presenting Author: Monika Haubitz, Institute of Animal Sciences, ETH Zurich, 8092 Zurich, Switzerland

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1. Benjamin Bucher
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Abstract:

Arthrogryposis multiplex congenita (AMC) is a malformation in mammals. Newborn piglets showing AMC symptoms suffer from persistent flexion of the limbs, deformation of the spinal column and do not survive. AMC can be caused by extrinsic and genetic factors. A genetic caused AMC was identified in Swiss Large White pigs. It is autosomal recessively inherited and the mutation was mapped to a 5 cM region between the two microsatellite markers *SW152* and *SW904* on porcine chromosome 5, close to

marker *S0018*. Functional and positional candidate genes were partially sequenced in order to locate new markers and to find the causative *AMC* mutation. A candidate gene located close to *S0018* is the contactin 1 gene *CNTN1*. It was investigated by sequencing several of its exons and introns and subsequent sequence alignments. In eleven exons and the surrounding intron sequences 16 SNPs, three deletions and one insertion were found. One SNP is in linkage disequilibrium with *AMC*. The SNP was studied in 82 animals from our own breed and in 38 animals from commercial farms. This SNP may be useful to discover *AMC* carriers and therefore, to reduce the incidence of *AMC* in pig farming.

Poster 2208

Title: Nutrigenomics studies in sheep using a custom microarray platform

Presenting Author: Sgorlon Sandy, Via delle Scienze, 208 – 33100 Udine, ITALY

Other authors (name only):

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3. Dreos Renè
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Abstract:

The aim of the research was to investigate the modifications of gene expression profiles in blood cells, after dietary supplementation with *Echinacea angustifolia* (EA) or *Andrographis panicolata* (AP) (INDENA, Milan) to sheep under ACTH challenge. Experimental protocol was approved by local laws and regulations. Twenty four sheep at maintenance were allotted to 4 groups: CTR- (negative control, without ACTH and supplementation), CTR+ (positive control, with ACTH and without supplementation), EA (ACTH and 3 mg/kg liveweight of EA) and AP (ACTH and 1 mg/kg liveweight of AP). ACTH was injected for 3 subsequent days and blood was sampled before (T0) and after 3 (T3) and 51 (T51) hours from the first injection. RNA extracted samples were pooled together within group and time of sampling. A custom oligoarray was synthesized using 24384 35-40mer probes designed from 12194 UniGenes (NCBI) on a CombiMatrix 90K platform. Cy5 labelled samples were hybridized on the chip. Data analysis of microarray allowed to identify a set of genes which were up or down regulated as a consequence of ACTH treatment. Functional classification showed that most of the genes behave to immune response and translation categories. EA and AP administration also showed an activity on these pathways regulation.

Poster 2209

Title: Linkage disequilibrium in a Korean beef cattle (Hanwoo) population

Presenting Author: Jong-Joo Kim, School of Biotechnology, Yeungnam University, Gyeongsan, 412449, Korea

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Abstract:

The purpose of this study was to evaluate linkage disequilibrium (LD) in a Korean beef cattle (Hanwoo) population. Forty eight steers under paternal half-sib families and eight sires, each with six steers, were selected and sampled for DNA extraction in the population of Jeong-Ub Hanwoo association. Using the Illumina chips with 55K SNPs, 38,023 SNPs were determined as useful SNPs in Hanwoo, which was based on minor allele frequency of 0.05 and call rate of 90%. The SNPs were distributed across autosomal chromosomes with maximum (minimum) number of 2372 (659) on BTA1 (BTA28). Linkage phases or haplotypes were determined using fastPHASE (v 1.2.) program, and LDs between syntenic markers using only maternal chromosomes (haplotypes) were measured with the r^2 method. When the two markers were within 10 kbp, the average r^2 value was 0.52, decreased down to 0.2 for the pair of markers within 40 kbp, and down to 0.1 within 200 kbp. When the pair of markers was located longer than 5000 kbp (5cM), the average r^2 value was in equilibrium at 0.025. These results will provide useful information in implementing fine-mapping QTL for production traits in Hanwoo.

Poster 2210

Title: Frequency of the Red Factor (MC1R) in the Polish Primitive Horse in Poland

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Abstract:

The Polish Primitive Horse is a breed developed in Poland beginning in 1923. The aim of the development of the breed was the regeneration of the

extinct Forest Tarpan by way of natural selection in semi-wild conditions. The horses used in constructing the breed were descended from the wild Tarpan which had existed in Poland until late in the XIX century. The founding population of horses was small but today the total number of animals is about 2000. To match the appearance of the Tarpan, the preferred color of Polish Primitive Horses is grullo or mouse. However, some chestnut colored horses are born each year. As this is an undesirable color to breeders we initiated a study to determine the frequency of the recessive missense mutation in the gene for melanocyte-stimulating hormone receptor (MC1R) associated with the chestnut color in horses. 186 individuals that were not closely related were genotyped using an RFLP method (120 to 200 more animals are being typed). The frequency of the red factor mutation was 0.167. Identification of carriers of the red factor can be used as an aide to breeders in making breeding decisions to eventually eliminate this mutation from the population with minimal impact on genetic diversity within the breed.

Poster 2211

Title: Molecular genetic dissection of bone mineral traits and osteochondrosis in porcine limbs

Presenting Author: Chirawath Phatsara, Institute of Animal Science, University of Bonn, Endenicher Allee 15, Bonn GERMANY

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Abstract:

Osteochondrosis (OC) is regarded as the main cause of leg weakness in pigs. In this study, the OC lesion at articular cartilage was observed using a histological scoring method in 310 F₂ Duroc X Pietrain crossbred population. As well as bone mineral content and density (BMC and BMD, respectively) were also observed using a dual-energy x-ray absorptiometry technique. The histological scoring results revealed the most prevalent location was the knee joint (distal femur). There were no correlation between bone mineral traits (BMD and BMC) and performance traits, except slaughter weight. Five functional candidate genes (*MGP*, *MMP3*, *TGFβ1*, *COL2A1*, and *COL10A1*) for Osteochondrosis and bone mineral traits were screened for polymorphisms and further analysed for an association to the traits. Ten SNPs

were identified in the candidate genes and genotyped by multiplex SNPs base extension method. An association analysis revealed polymorphisms of *TGFβ1* and *MGP* were significantly associated with OC lesion at knee and hip joints, respectively. Polymorphisms of *MGP*, *MMP3* and *COL2A1* were found associated with BMD, while the association between *MGP*, *MMP3* and *TGFβ1* polymorphisms and BMC were also detected. Haplotype analysis of five candidate genes showed no association to the traits of interest.

Poster 2212

Title: Genetic diversity and admixture among Chinese cashmere goats

Ran DI¹

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Abstract: Cashmere goats occur in northern China and Mongolia and are used to produce high quality natural fibre for the textile industry. In this study, 14 ISAG/FAO goat microsatellite markers were used to assess genetic diversity and admixture among nine Chinese cashmere goat breeds. Two Iranian and one West African goat populations were genotyped for comparative purposes. The results showed that the Chinese cashmere goats were much more genetically diverse (Nei's unbiased gene diversity = 0.540-0.6520; allelic richness = 3.904-5.956) than the West African population (0.599 and 4.901), but lesser than the Iranian populations (0.6535-0.699 and 6.340-6.566); backing the hypothesis that goats were domesticated in the Middle East, from where they dispersed to Asia and Africa. Bayesian clustering approach grouped the Chinese cashmere goats into three separate genetic entities that were consistent with their geographic distribution and breeding history. The Tibetan Hegu goats were genetically distinct and formed the first cluster. The Liaoning cashmere goats from northeastern China together with the Xinjiang and Shanbei cashmere goats formed the second, while the remaining five breeds (Alashan, Erlangshan, Wuzhumuqin, Caidamu and Hexi) found in northwestern China formed the third cluster. These results could form the basis for genetic improvement of cashmere trait.

Poster 2213

Title: The first genetic linkage map for the saltwater crocodile (*Crocodylus porosus*)

LEE MILES¹, SALLY ISBERG^{1,2}, PAULINE DALZELL³, TRAVIS GLENN^{4,5}, PETER THOMSON¹, DANIEL PETERSON⁶, XUEYAN SHAN⁶ AND CHRIS MORAN¹

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Abstract:

Genetic maps currently exist for many commercially as well as evolutionary important species. In this study we presents the first genetic-linkage map for the Saltwater Crocodile (*Crocodylus porosus*), and indeed the first for the Class Reptilia. We constructed male, female and sex-averaged linkage maps for *C. porosus* using a total of 189 microsatellite DNA markers typed for between 83 and 480 individuals from between five and ten families obtained from Darwin Crocodile Farm, NT Australia. We identified 12 linkage groups (LG) with LG sizes ranging from two to 53 loci. The overall map consists of 161 loci, while 28 markers still remain unlinked. LG are currently being physically anchored to chromosomes (2n=34) using Fluorescent *In Situ* Hybridisation (FISH) methods. The current linkage map confirms that female crocodiles have extraordinarily higher rates of recombination than males, with overall map lengths of 1636.5 cM and 275.2 cM respectively. Next to the Atlantic salmon, this uncommonly large difference in recombination rates is the greatest reported for a vertebrate species. This first genetic map lays the framework for further mapping analyses, with QTL mapping of economically and evolutionarily important traits currently underway in the saltwater crocodile.

Poster 2214

Title: Intra-specific sequence variability of tumor necrosis factor-alpha gene (TNFα) in domestic horse breeds

Francisco EI^{1,3}, Kalemkerian PB^{1,3}, Liron JP¹, Giovambattista G¹, Echeverría MG², Peral García P¹, Diaz S¹

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Abstract:

Tumour Necrosis Factor alpha (TNF α) is a cytokine that plays a fundamental role in inflammation and immune responses. In horses the coding sequence of TNF α has been mapped within the MHC Class III region in chromosome 20, and SNPs in the promoter region have been previously reported in equine breeds and species. In order to preliminarily investigate the genetic variation within the TNF α gene in domestic horses, we analyse the genomic full-length sequences in fourteen animals from four horse breeds (Argentine Creole, Arab, Spanish Pure Breed and Argentine Silla). Oligonucleotide primers were designed based on the reported TNF horse genomic sequence, to amplify by PCR four overlapping fragments comprising ~2.6 kb within the gene. Each PCR product was sequenced, analyzed and the TNF full-length sequence assembled on each animal by using the algorithms of the Vector NTI and DNAMAN programs. A total of nine SNPs were observed, seven of them located in non-coding regions and two in coding regions. In addition, inter-specific variability was observed when compared to TNF reported sequences from other mammalian species; this variability corresponds to the presence of horse specific amino-acid motifs. Further studies are needed in order to investigate the functional specificity of those motifs and the significance of the observed intra- and inter-specific polymorphisms both in gene expression levels as in susceptibility to various horse diseases.

Poster 2215

Title: Association Study between DLA polymorphism and susceptibility to chronic superficial keratitis in German Shepherd dogs.

It, Verónica¹; Laura S Barrientos¹; Gustavo Zapata²; Silvina Díaz¹, Guillermo Giovambattista¹.

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Abstract:

Chronic Superficial Keratitis (CSK) is a progressive, inflammatory and potentially blinding disease of the canine bilateral cornea. The German shepherd is the most commonly affected breed. CSK have been described as an immune-mediated disease. The increased MHC class II expression may play a part in

perpetuating the corneal inflammation seen in the disease. Given the previously reported association between the CSK and up-regulation of major histocompatibility class II antigen expression, the primary goal of this work is to determine the association between the Dog Leukocyte Antigen system (DLA) and the disease in German Shepherd dogs. Blood samples were collected from twelve dogs with pannus and twelve control dogs. The polymorphism of DLA-DQA, -DQB and -DRB promoters, and four DLA linked microsatellites was genotyped. Genetic diversity was calculated for each locus, and association between genetic markers and presence/absence of pannus was evaluated. Preliminary results showed that: (i) the allele DLA-DQA1-DQA*p1a is fixed in both subpopulations; (ii) two alleles of the DLA-DRB1 promoter (DLA-DRB1*p1 and DLA-DRB1*p2) were detected with similar gene frequencies in both groups; (iii) we have only detect one (DLA-DQB*7) out of four DLA-DQB alleles reported in German Shepherd, as well as, the allele DLA-DQA*p1; (iv) three putative new DLA-DQB promoter variants were detected in our sample; (v) not significant differences were observed between illness and control group for the microsatellites FH2200 and FH2202; (vi) significant differences between pannus and control for the loci FH2054 and FH2975 (pFH2054 = 0.00744; pFH2975 = 0.003214) were observed; (vii) a relative risk of 6.25 and 2.75 for the alleles FH2054*152 and FH2579*320, respectively, were calculated; and (viii) an odds ratio of 9 and 3.4 for these alleles, respectively, were estimated.

Poster 2216

Title: Genetic variability in Hunting dog breeds using microsatellite markers and mitochondrial DNA polymorphisms.

Presenting Author: Giulia Pertica, Dipartimento di Scienze Animali - Sezione di Zootecnica Veterinaria - Università degli Studi di Milano, 20133 Milano – Italy

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Abstract:

In order to define genetic relationships of Hunting dog breeds, 258 unrelated dogs of 13 breeds (pointing, retrievers and spaniels) were analyzed using ten microsatellite markers. Allele frequencies, average

number of alleles per locus, Hardy-Weinberg equilibrium were determined by GENEPOP. Observed proportion of heterozygotes (H_o), gene diversity within sample (H_s), overall gene diversity (H_t) were calculated using FSTAT. Factorial Correspondence Analysis (FCA) was carried out using the GENETIX. Individual genetic distances (D_{DAS}) were estimated and individual dendrogram was constructed using POPULATION. Individual breed assignment was carried out by STRUCTURE. Genetic variations at the 694 bp fragment of the mtDNA D-loop were evaluated. We analysed 5 samples for each breeds. Sequences obtained were aligned with the complete canine mtDNA sequence (GenBank#NC_002008) using the CLUSTALX. Haplotype diversity (H_d), nucleotide diversity (π), average number of nucleotide differences (k) and Fu's F_s statistic were calculated using DnaSP. Genetic distances were estimated by the Kimura-two parameter method and a phylogenetic tree was constructed by UPGMA algorithm. Median-joining network and mismatch analysis were calculated in order to investigate haplotypes relationship using NETWORK. The proportion of allele sharing combined with mtDNA analyses resulted in a clear clustering of the analyzed breeds.

Poster 2217

Title: Horse morphological judgment follow natural selection criteria?

Presenting Author: Jose Luis Vega-Pla. Laboratorio de Investigación Aplicada. Aparatado Oficial Suc. 2, 14071-Cordoba (Spain)

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Abstract:

The heterozygosity theory attempts to identify the essence of genetic quality. It leads to the following general prediction: Traits of all kinds that are used by females when judging males reach their extreme expression in males with the greatest average heterozygosity. (Brown, 1997). More specifically, if the heterozygosity theory is correct, male symmetry expression, therefore male mating success in a large population, will positively correlate with degree of individual heterozygosity. The present study checks on the hypothesis that judge criteria in morphological competitions of Spanish Pure Breed horses (Andalusian) are linked to the heterozygosity theory. The inbreeding coefficient over the entire pedigree of each animal was estimated in two hundred champion stallions and eighty-four mares that became one of the three first positions in different morphological

competitions. Also Heterozygosity and Fis statistic was calculated from twelve microsatellites typing. Results were compared with those obtained from a random sample population.

Poster 2218

Title: MicroRNA expression in differentiating porcine myoblast cells

Presenting Author: Mathilde Nielsen, University of Aarhus, Department of Genetics and Biotechnology, Denmark

Other authors (name only):

1. Bo Thomsen

Abstract:

MicroRNA contributes to regulation of gene expression by binding to complementary sites in protein-coding mRNA, thereby modulating the translation process. MicroRNAs control many biological processes such as myogenesis, and several muscle-specific microRNAs are known to be involved in diverse aspects of muscle function such as myoblast proliferation, differentiation, contractility and stress responsiveness. We are interested in identifying microRNAs that regulate muscle growth and differentiation. Thus, small RNA libraries were constructed from pig skeletal muscles and sequencing analysis identified several microRNAs including miR-133 and miR-206. The expression pattern of miR-133 and miR-206 were examined during differentiation of cultured myoblast cells into myotubes using stem-loop primers for reverse transcription followed by TaqMan PCR analysis. The data showed that the levels of mature miR-133 and miR-206 were low in the undifferentiated proliferating cells and that induction of myogenic differentiation pathway led to abruptly increased expression levels. This result is in agreement with the observation that miR-133 and miR-206 are among the most abundant microRNAs in skeletal muscle tissue and suggests a critical role of these microRNAs in expression of muscle-specific proteins.

Poster 2219

Title : Method of accurately detecting copy number variations in porcine *KIT* copies by oligonucleotide ligation assay

Presenting Author: Chae-Kyoung, Yoo, Division of Applied Life Science, Gyeongsang National University, Jinju 660-701, Korea

Other authors

1. Bo-Yeong, Seo

2. In-Cheol, Cho
3. Jung-Gyu, Lee
4. Jin-Tae, Jeon

Abstract

Previous studies have used pyrosequencing, minisequencing, real-time PCR, invader assays and other techniques to detect copy number variations (CNVs). However, the higher a genome's copy number, the more difficult it is to resolve the copies; so an accurate method of measuring CNVs and assigning genotypes is needed. PCR followed by a quantitative oligonucleotide ligation assay (qOLA) was developed for quantifying CNVs. The assay's accuracy and precision were evaluated for porcine *KIT*, which was selected as a model locus. Using a method that combined qOLA and another pyrosequencing to quantitatively analyze *KIT* copies with spliced forms, we confirmed the segregation of *KIT* alleles in 145 F₁ animals with pedigree information; and verified the correct assignment of genotypes. In a diagnostic test on 100 randomly sampled commercial pigs, the genotypes obtained by grouping observations on a scatter plot agreed perfectly with those obtained by clustering using the nearest centroid sorting method implemented in PROC FASTCLUS of the SAS package. A test on 159 large white pigs noted only two discrepancies between genotypes assigned by the two clustering methods (98.7% agreement), confirming that the quantitative ligation assay established here enables genotyping through the accurate measurement of high *KIT* copy numbers (>4 per diploid genome).

Poster 2220

Title : Reconfirming and minimizing a QTL for intramuscular fat content by polymorphism of pig *PDE4B* in an Iberian x Landrace population

Presenting Author: Jae-Hwan, Kim, Address : Division of Applied Life Science, Gyeongsang National University, Jinju 660-701, Korea

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2. Eung-Woo, Park
3. Almudena, Fernandez
4. Jun-Heon, Lee
5. Jung-Gyu, Lee
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Abstract

The phosphodiesterase 4B (*PDE4B*) gene belongs to the PDE4 family, which specifically hydrolyzes intracellular cAMP. In this study, three isoforms of pig *PDE4B* were cloned and classified as two forms: *PDE4B1* and *PDE4B3*, which contain both upstream conserved regions (UCR1 and UCR2); and *PDE4B2*,

which contains only UCR2. Each isoform's amino acid sequences showed strong conservation with human and rat genes. Thirteen different tissues express *PDE4B2* in a wide range of tissues, but the expression of *PDE4B1* and *PDE4B3* varies among tissues. Using an informative polymorphism (which needs to be specified exactly) for the Iberian x Landrace intercross detected from intron 12, a linkage map was constructed. The location of *PDE4B* was estimated at 123.6 cM outside the quantitative trait loci (QTL)-confidence interval (CI) (124-128 cM) for intramuscular fat content (IMF). However, QTL-CI for IMF was reconfirmed with high significance, and its position was narrowed down to an interval of 4 cM (the region defined by markers *PDE4B* and *SW1881*). Using radiation hybrid mapping, *LEPR*, *LEPROT*, *DNAJC6*, *AK3L1* and *AK3L2* were selected as positional and/or functional candidates related to QTL.

Poster 2221

Title : Molecular cloning and characterization of six transcription factors in cat embryonic stem-like cells

Presenting Author: Eun-Ji, Jung, Division of Applied Life Science, Gyeongsang National University, Jinju 660-701, Korea

Other authors

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4. Jung-Gyu Lee
5. Jin-Tae, Jeon

Abstract

Six transcription factors (*POU5F1*, *NANOG*, *SOX2*, *LIN28*, *KLF4* and *MYC*) play important roles in the maintenance of embryonic stem cell pluripotency. We recently isolated cat embryonic stem (ES)-like cells from cat blastocysts generated *in vivo*. To identify genetic markers for characterizing cat ES-like cells, we have determined these six genes' coding sequences (CDSs). The sequence identity of these genes with orthologs of humans and mice was 76-100% and 58-99%, respectively, at the amino acid level. Among these, the expression of *POU5F1* and *NANOG* mRNA was investigated in ES-like cells, blastocysts, fibroblast feeder cells and six adult tissues by RT-PCR. Transcripts of *POU5F1* and *NANOG* were detected at a high level in ES-like cells and blastocysts. However, these two genes were undetectable in cat fibroblast feeder cells and six adult tissues. We also examined ES-like cells by immunocytochemistry, and demonstrated that *POU5F1* and *NANOG* are present at a high level in cat

ES-like cells, and are undetectable in cat fibroblast feeder cells. These results confirm that cat ES-like cells can be successfully isolated from blastocysts produced *in vivo*. Moreover, the expression of *POU5F1* and *NANOG* can be used as a biomarker for the characterization of cat ES-like cells; and *SOX2*, *LIN28*, *KLF4* and *MYC* are necessary for additional analyses using cat ES-like cells.

Poster 2222

Title: A comparison of discriminating power between 14 STR and 60 SNP makers for individual cattle identification

Presenting Author: Hyun-Tae, Lim, Division of Applied Life Science, Gyeongsang National University, Jinju 660-701, Korea

Other authors

1. Jay You
2. Han-O, Park
3. Jung-Gyu, Lee
4. Jin-Tae, Jeon

Abstract

Short tandem repeat (STR) and single nucleotide polymorphism (SNP) typing methods have been widely used for cattle paternity and identity testing. Eleven STR markers used in the StockMarks® of Applied Biosystems, and three additional STR markers (*BMS1747*, *BM4305* and *BL1009*), were selected and prepared in a multiplex PCR. Sixty SNP markers applied in the iPEX™ assay for the cattle paternity test of SEQUENOM were used. Genotyping data were collected from 480 Korean cattle. The discriminating efficiency of the two types of markers was compared by calculating *He*, *PIC*, *F*-statistics and probability of homolog using the MSA, CERVUS, FSTAT, GENEPOP and API-CALC programs. The probabilities of homolog for the STR markers were estimated as 3.43×10^{-27} , 4.18×10^{-19} and 3.98×10^{-8} in random mating, half-sib mating and full-sib mating populations, respectively. For the SNP markers, the probabilities were estimated at 2.09×10^{-24} , 4.69×10^{-20} and 8.02×10^{-12} , respectively. For cattle individual identification, the STR markers would be about 1000 times more efficient than the SNP markers in a random mating population, and the SNP markers would be more effective than the STR markers in half-sib mating and full-sib mating populations. This indicates that a high number of makers is more informative in populations with blood relationships, although an STR markers has more alleles than an SNP markers in general.

Poster 2223

Title: High-Resolution RH Mapping of a Potential QTL Region on Sheep Chromosome 9 (OAR9)

Presenting Author: Tracy Hadfield, Utah State University, Old Main Hill 4700 Logan UTAH 84325 USA

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Abstract:

Gastrointestinal parasites have a profound detrimental effect on sheep production. Molecular markers associated with parasite resistance in sheep could be useful for genetic selection of superior animals. As a first step in identifying suitable markers, a genome-wide QTL scan has been implemented using DNA samples from 42 grandparents, 3 F1 sires, 97 F1 dams, and 195 F2 offspring, for which fecal egg count (FEC) for *Haemonchus contortus* were measured after natural and experimental challenges. Selective genotyping of the upper and lower 20% of the lambs for FEC after natural challenge identified a potential QTL region on ovine chromosome 9 (OAR9) between markers BMS108 and MCM63 within a 3.5 cM interval. In order to refine the QTL region, a high-resolution radiation hybrid (RH) map of OAR9 was constructed using the USUoRH5000 panel. Using a logarithm of odds (LOD) threshold of 6.0, a single linkage group was constructed and aligned with the genetic linkage map of this chromosome. The resulting RH map contains assignments for 28 microsatellite sequences, 21 BAC-end sequences and 8 genes. These experiments allowed resolution of the parasite resistance QTL region to an 8.0 Mb interval at position 90.0 Mb on OAR9.

Poster 2224

Title: DIAGNOSTIC OF EYE HEREDITARY DISEASES IN SLOUGHIS

Presenting Author: MICHAELA PŘIBÁŇOVÁ¹, Ceskomoravska společnost chovatelů a.s., Laboratory of Immunogenetics, 252 09 Hradistko 123, Czech republic

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Abstract:

Progressive retinal atrophy (PRA) causing blindness occurs in many breeds of dog in different forms. Sloughis suffer from late onset PRA form (*rcd1a*). Small population in Czech republic is based on Sloughis imported from Algeria, later breeders used for mating dogs from France, Germany, Italy and Morocco.

In this study, mutation test, causing PRA in Sloughis (8-bp insertion in exon 21 of the *PDE6B* gene, Dekomien et al, 2000) was used for testing Czech population of Sloughis. We have tested total sum of 42 individuals, 41 Sloughis (30,4% of all living dogs registered in Czech studbook) from 13 different litters (86,7% of all registered litters) and 100% imported dogs and 1 Erdelterraier, clinically affected with unknown PRA form. For comparison of results, samples of Affected and Carrier (heterozygous) Sloughis, obtained from Germany was used. All tested dogs was homozygous Normal, so we can claim, Czech Sloughi's population is nearly clear from *rcd1a* mutation in contrast to German and American population.

We used the same test for clinically PRA affected Erdelterraier. In this dog breed PRA-causing mutation is unknown. There was observed no presence of *rcd1a* mutation in this sample, so we can except it as a cause of PRA in Erdelterraiers.

(This work was supported by IRP IAPG project no. AVOZ50450515)

Poster 2225

Title: Immunological Parameters of the Chicken Lines selected for the response in Graft-versus-host reaction *in ovo*

Presenting Author: S Mashima, Graduate school of Science and Technology, Niigata University, Niigata, Japan

Other authors (name only):

1. Ko-zaburo Yamamoto
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Abstract:

Chicken lines selected for the high and low competence in the response of the graft-versus-host reaction *in ovo* are valuable models for avian immunogenetics. Although *HA* and *LA* lines had been developed from a single colony and share the same *B* haplotype, *HA* is high responder in the reaction,

whereas the later exhibits lower response++ in the reaction.

Previously, *HA* and *LA* line showed significant differences in susceptibility to Marek's disease (MD). The higher mortality in the *HA*, as compared with the *LA*, had been confirmed by virus challenge and transplantation of MD-transformed cell line.

In this study, we characterized cellular immunological parameters such as constitution of T cell subsets, mitogenic response to lectins or allogenic cells in these lines.

Poster 2226

Title: Equine individual identification and parentage verification with 18plex and 22plex PCR

Presenting Author: J.A. Bouzada, LCV, Algete, Spain

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Abstract:

Multiplex PCR is a powerful technique typically used in genotyping applications where the simultaneous analysis of multiple markers is required. For parentage testing and individual identification *multiplex* PCR amplifying DNA microsatellites is routinely used. In this study we propose a main set of 18 markers (AHT4, AHT5, ASB2, ASB17, ASB23, CA425, HMS1, HMS2, HMS3, HMS6, HMS7, HTG4, HTG6, HTG7, HTG10, LEX3, LEX33 y VHL20) and an additional set of 22 markers, (AHT29, AHT39, HMS8, LEX22, LEX27, TKY19, TKY279, TKY287, TKY294, TKY297, TKY301, TKY312, TKY321, TKY325, TKY333, TKY337, TKY341, TKY343, TKY344, TKY374, TKY394 y UCDEQ405) from the 2005-2006 International Society of Animal Genetics (ISAG) Comparison Test. The markers of each panel can be co-amplified simultaneously and meet the needs for parentage testing an individual identification in horses. Some changes in the habitual primers were required to achieve a final configuration that allowed all markers to be analysed together. Robotic procedures were implemented to minimize the risk of genotyping errors and parentage verification mistakes. DNA from whole blood was extracted using *BioSprint 96 Blood Kit (Qiagen)* and each panel of markers was simultaneously amplified using the QIAGEN Multiplex PCR kit. PCR products were analyzed in a single run on an ABI PRISM® 3100 Genetic sequencer.

Microsatellites included in the proposal panels were highly polymorphic. The mean allele number is 6.61 in the main and 7.00 in the additional panel. The cumulative exclusion probability reached 99.9999526% using the main set and 99.9999977% using the additional one. The main panel is used routinely for equine parentage verification at LCV, and the additional one, as a complementary set of markers in more complex paternity queries involving related sires or when exclusion for one only marker is observed in the main panel. DNA markers used in this study can be used in numerous fields of animal breeding, e.g. examination of genetic structure of populations, estimation of genetic distance between populations, estimation of genetic distance between populations and breeds, as well as planning of crossing programs.

Poster 2227

Title: Detection of QTL on BTA6 for carcass traits in a Korean beef cattle (Hanwoo) population

Presenting Author: Yun-Mi Lee, School of Biotechnology, Yeungnam University, Gyeongsan, 412449, Korea

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Abstract:

The purpose of this study was to detect QTL for carcass traits in a Korean beef cattle (Hanwoo) population. Four hundred and fifteen steers were produced from 25 grand-sires under the national progeny-testing program in National Livestock Research Institute, Korea. The traits analyzed in this study were final weight at 24 month, carcass weight, backfat thickness, average daily gain, longissimus dorsi muscle area, marbling score. DNAs were extracted and eight microsatellite markers were chosen on BTA6, genotyped and linkage-mapped using Cri-map. The used markers and map locations (cM) were IL90 (0), BMS2508 (15), BMS518 (34), BM4621 (63), BM2460 (81), BM8124 (117), IL35 (144), and BMC4203 (175). MQREML program was used to detect QTL, and threshold at the 5% chromosome-wise level was determined using chi-square distribution with one degree of freedom. Three QTL were detected at the 5% chromosome-wise level; for average daily gain at 33 cM, for final weight at 63 cM, and for backfat thickness at 0 cM, respectively. These results will provide useful information to find

potential candidate genes relevant to the traits that are located near the detected QTL.

Poster 2228

Title: A First-Generation Ovine Whole-genome Radiation Hybrid Map using the USUoRH5000 Panel

Chunhua Wu, Koh Nomura, Tom Goldammer, Jens Tetens, Cord Drögemüller, Tracy S. Hadfield, Brian P. Dalrymple, Jill F. Maddox and Noelle E. Cockett

Presenting Author: Noelle E. Cockett, Department of Animal, Dairy and Veterinary Sciences, Utah State University, 4900 Old Main Hill, Logan, Utah 84322-4900 USA

Abstract:

Whole-genome radiation hybrid (RH) mapping is a method for producing high resolution maps which can be used for integrating linkage and physical maps within a species and for determining gene order. Comparative maps can be created by incorporating genes/ESTs into the RH map that are common across species. An ovine whole-genome 5000 rad RH panel, referred to as USUoRH5000, was used to construct a first-generation ovine RH map that contains 918 type I and 388 type II markers clustered into 95 RH linkage groups distributed over all 26 ovine autosomes. The RH map has good agreement with existing ovine linkage and physical maps. The mapped loci were distributed, on average, every 18.42 cR (~ 2.63 Mb). By identifying the locations of mapped loci within the whole genome sequences of human and other closely related mammalian species, ovine-mammalian comparative maps were developed. Large blocks of synteny exist between sheep and other mammalian species, but previously unreported micro-rearrangements have been identified in this study. The USUoRH5000 panel will be an important tool for assembly of the ovine whole genome sequence and refinement of the virtual sheep genome, as well as aiding in the identification and positional cloning of genes influencing traits in sheep.

Poster 2229

Title: A 6540 bp deletion in the LAMA3 gene is associated with hereditary junctional epidermolysis bullosa (JEB) in the American Saddlebred Horse

Presenting Author: Kathryn T. Graves, Department of Veterinary Science, University of Kentucky, Lexington KY USA 40546

Other authors (name only):

- 1 Pamela J. Henney

Abstract:

Laminin 5 is a heterotrimeric basement membrane protein integral to the structure and function of the dermal-epidermal junction. It consists of three glycoprotein subunits; the $\alpha 3$, $\beta 3$ and $\gamma 2$ chains, encoded by the *LAMA3*, *LAMB3* AND *LAMC2* genes respectively. A mutation in any of these genes results in the condition known as hereditary junctional epidermolysis bullosa (JEB). A 6540bp deletion spanning exons 24 to 27 was found in the *LAMA3* gene in American Saddlebred foals born with the skin blistering disease epitheliogenesis imperfecta. The deletion confirms that this autosomal recessive condition in the American Saddlebred Horse can indeed be classified as junctional epidermolysis bullosa and corresponds to Herlitz-JEB in humans. A diagnostic test was developed and 9 of 175 randomly selected American Saddlebred foals from the 2007 foal crop were found to be carriers of the mutation (gene frequency 0.026).

Poster 2230**Title: Whole genome association study for protein yield in German Braunvieh****Authors:**

Presenting Author: Hermann Schwarzenbacher, Lehrstuhl für Tierzucht, Technische Universität München, Hochfeldweg 1, 85354 Freising-Weihenstephan, Germany

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Abstract:

Selective genotyping was applied to 140 bulls with highest estimated breeding values (EBVs) for protein yield and to 148 bulls with lowest EBVs among 973 progeny tested Braunvieh bulls. Animals were genotyped using the Illumina[®] BovineSNP BeadChip which comprises 54001 SNPs. PLINK software v1.02 and the R environment for programming and statistical computing were used for the analysis of genotyping data. SNP and animals with call rates <90% were omitted. Other exclusion criteria were HWE P-values ≤ 0.001 and a minor allele frequency <5%, resulting in 36821 SNPs in 287 animals to be included in the association study by allele regression. Comparison of observed with expected test statistics under the null hypothesis indicated substantial stratification. However, accounting for pedigree information and identical by state clustering successfully controlled for

population substructure. Several significant SNPs will be followed up by fine-mapping and candidate gene analyses.

Poster 2231**Title: Genetic relationships between native goat populations from Spain, Portugal and America**

Presenting Author: Amparo Martínez Martínez, Departamento de Genética, Universidad de Córdoba, Spain.

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Abstract:

Twenty seven microsatellite markers were analysed in 1450 goats belonging to 38 native goat populations from Spain, Portugal and related Creole populations from America in order to establish their genetic relationships. The genetic differentiation between populations was moderate with a mean F_{ST} value of 0.13. The neighbour-joining tree constructed with the D_A genetic distance values demonstrate that Creole populations from Argentina, Bolivia and the Spanish goat from USA grouped in the same cluster that the Iberian populations while the Cuban Creole goat grouped with the populations from Canary and Cabo Verde Islands. Clustering of populations and estimates of the proportions of the individual genomes that were derived from the respective inferred clusters were obtained using the model-based clustering program Structure (Pritchard et al. 2000). Results for $k=2$ showed that populations from the Iberian Peninsula (Spain and Portugal) and most Creole goats grouped in the same cluster, while the Brazilian, Canarian, Cuban and Arapawan populations formed another cluster. Setting $k=3$ generated a cluster with the Canarian, Cuban and Arapawan populations. The results obtained suggested a clear influence of the Iberian populations in the Creole goats, while the Cuban Creole goat is more influenced by the Canarian breeds than by Iberian goats.

Poster 2232**Title: Annotation, polymorphism and expression analysis of the bovine *INS-IGF2* locus**

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Wolka Kosowska

Abstract:

Transcription of insulin (*INS*) and insulin like growth factor (*IGF2*) genes results in a bicistronic mRNA. Expression of the two genes may therefore be subject to common regulation. Based on the bovine draft sequence Btau_3.1 and EST databases of different species, we determined the intron-exon structure and identified regulatory regions including five promoters at the *INS-IGF2* locus. Eight novel polymorphisms were identified in the regulatory regions, among them three in the *INS* promoter that potentially alter the binding capacity of AP-2 and NF-1 transcription factors. Additionally, a detailed quantitative analysis of tissue- and age-dependent expression of the *IGF2* derived sequences was performed. The mRNA level differed significantly between tissues. Highest expression was observed in the liver and kidney, whereas it was only spurious in muscle and spleen. Expression was fourfold higher in two-month male fetuses than in twelve-month old bulls. Examination of the DNA methylation patterns revealed full methylation (93%) of the *INS-IGF2* promoter independent of the age. Two SNPs localized in this region create new methylated CpG sites. These polymorphisms could affect the *cis*-activation of the bovine *INS-IGF2* transcript and may be associated with variation of developmental traits.

Poster 2233

Title: Genetic characterization of Italian heavy draft horse (IHDH) breed using microsatellite markers

Presenting Author: Fabio Maretto, Department of Animal Science, University of Padova, AGRIPOLIS, 35020 Legnaro (PD), Italy.

Other authors (name only):

1. Cinzia Ribeca
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Abstract:

The IHDH breed was established in 1926 by the Italian government and originated from crosses of Norfolk-Breton stallions with local derived Hackney, Percheron and Bretons mares. The breed was developed for agricultural and draft uses as well for artillery transport by the Italian army; nowadays it is mainly used for meat production and heavy draft works. This study compares genetic diversity of IHDH and other two unrelated breeds (Italian Haflinger, IH and Quarter Horse, QH breeds) by means of 23

microsatellite markers. A total of 95 animals (IHDH n = 55; IH n = 19 and QH n = 21) were genotyped. A total of 165 alleles were detected in the whole sample and the number of alleles (N_A) varied between 3 (locus Htg7 in the IH breed) and 13 (locus Tky343 in the IHDH breed) with a mean of 6.06 ± 1.72 . The average observed heterozygosity (H_0) was 0.66, 0.67 and 0.76 for IH, IHDH and QH breed, respectively. The IHDH breed showed a good genetic variability comparable to the other analysed breeds. This study provides the first baseline of data for the IHDH breed and contributes to the conservation and implementation of selection programme of the breed.

Poster 2234

Title: High throughput SNP discovery and validation in the Pig: towards the development of a high density swine SNP chip

Presenting Author: Crooijmans Richard P.M.A. Animal breeding and Genomics Centre, Wageningen University, Marijkeweg 40, Wageningen, The Netherlands

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Abstract:

Recent developments in sequencing technology have allowed the rapid and inexpensive generation of millions of short read sequences, enabling the cost effective identification of the hundreds of thousands of SNPs. Currently, a high density swine SNP chip is being developed as part of an integrated effort of several European and U.S. institutions involved in swine genomics research. The future pig SNP chip will include previously validated SNPs and SNPs identified *de novo* using second generation sequencing on the Illumina Genome analyzer (Solexa) and Roche 454 FLX sequencer. Towards this end, 20 DNA libraries were prepared using pooled DNA samples from individuals from five breeds (Duroc, Landrace, Large White, Pietrain, Wild Boar) digested with three restriction enzymes (*AluI*, *HaeIII*, *MspI*). The Solexa

short sequence and longer 454 reads will be filtered using several quality criteria to produce the dataset used for SNP discovery. Additional criteria for the selection of the 60K SNPs to be incorporated into an Illumina iSelect assay include the Illumina design score, Infinium I vs II, estimated minor allele frequency and genomic location of the SNPs. The 60K SNP chip will be an extremely valuable tool for a variety of applications including QTL and LD mapping, association studies and genomic selection.

Poster 2235

Title: Insulin induced gene 2 (*INSIG2*) is associated with fatty acid composition in the pig

Presenting Author: Anna Pertek, Lehrstuhl fuer Tierzucht, Technische Universitaet Muenchen, Hochfeldweg 1, 85354 Freising, Germany

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Abstract:

Insulin induced gene 2 (*INSIG2*) is involved in sterol regulatory element binding protein (SREBP) activation of fatty acids and thus a candidate gene for fatty acid composition traits. We investigated porcine *INSIG2* in a Mangalitsa x Piétrain cross. The genomic sequence of *INSIG2* was obtained by sequencing a porcine BAC clone. The coding sequence, flanking splice junctions and a putative promoter region were screened for polymorphisms in the parental animals of the cross. A total of 13 variants were identified, including a synonymous substitution (c.237A>G) and a microsatellite polymorphism (*121_122insGT) in the 3' untranslated region (3'-UTR). Four haplotypes were derived based on 12 variable positions, but haplotype 4 was not transmitted to the F2 generation. An association study was conducted including 324 F2 animals. Haplotype 1 significantly decreases saturated fatty acids percentage in the intramuscular fat. The content of polyunsaturated fatty acids (PUFA) is significantly associated with haplotypes 1 and 3. While haplotype 1 increases PUFA content, haplotype 3 reduces it. Haplotype 3 significantly increases monounsaturated fatty acids percentage. An obviously functional polymorphism could not be identified. However, microsatellite variation in the 3'-UTR may affect the expression of *INSIG2*.

Poster 2236

Title: Expression profiling and eQTL analysis

reveal biological pathways and candidate genes affecting water holding capacity of muscle

Presenting Author: Siriluck Ponsuksili, Research Institute for the Biology of Farm Animals (FBN), Research Group 'Functional Genome Analysis', 18196 Dummerstorf, Germany

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Abstract:

There is considerable variation in the water holding capacity (WHC) of meat, which is an important economic factor in pork production. WHC depends of numerous genetic and environmental factors that affect muscle fibre properties and metabolic processes ante and post slaughter. We aimed to identify candidate genes for WHC by QTL mapping, expression profiling and expressionQTL (eQTL) analysis. Expression microarray analysis of *M. longissimus dorsi* RNAs of 74 F2 animals of a resource population showed 1,279 transcripts with trait correlated expression. Negatively correlated transcripts were enriched in functional categories and pathways of extracellular matrix, receptor interaction, and Ca-signalling; positively correlated transcripts dominantly represented oxidative phosphorylation, mitochondrial pathways, and transporter activity. A eQTL analysis of trait correlated transcripts revealed 897 eQTL, with 104 eQTL matching previously identified QTL for WHC; 96 transcripts were trans regulated and 8 had cis acting regulation. Based on these findings, a priority list of genes was established out of the orchestra of genes of biological networks relevant to the liability to elevated drip loss. The complex relationships between biological processes in live skeletal muscle and meat quality are mainly influenced by the energy reserves and their utilisation in the muscle and by the muscle structure itself.

Poster 2237

Title: Identification of a SNP in *TRPM1* and its association with *Leopard Complex Spotting (LP)* and *Congenital Stationary Night Blindness (CSNB)* in horses.

Presenting Author (name & address): Rebecca Bellone
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Abstract:

The Leopard Complex Spotting patterns in horses is caused by an incompletely dominant gene (*LP*) mapped to a 6cM region on ECA1 containing the candidate gene *TRPM1* (Transient Receptor Potential Cation Channel, Subfamily M, Member 1). Homozygosity for *LP* is associated with congenital stationary night blindness (CSNB) in Appaloosa horses. One SNP, found within an intron, had a distribution identical to *LP* on a panel of 10 horses. To determine the strength of the association, 397 unrelated horses from different breeds were tested. There was a complete association of this SNP with *LP* ($P < 0.0005$) and CSNB ($N=30$, $P < 0.0005$) among Appaloosa and Knabstrupper horses. Among Noriker horses, a strong association was observed ($P < 0.0005$) however this association was not complete; 34 horses identified as non-patterned (*lp/lp*) possessed at least one copy of this SNP. These results suggest two possible conclusions: the SNP is not responsible for *LP* spotting but associated with the phenotype in Appaloosa and Knabstrupper horses while less so among Noriker horses; or the phenotypic assignments of *LP* were not accurate. Additional work is underway to identify other SNPs in the region and to assess their potential effects on the *LP* phenotype.