Transcriptome profiling reveals interaction between two QTL for fatness in chicken

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In the "genetical genomics" field, different approaches are proposed to improve characterization of QTL regions for various traits combining QTL detection and transcriptome profiling. The most common approach is to identify genes whose eQTL colocalize with QTL of interest, providing new functional hypothesis about the QTL causative mutation. A second approach used only by Schadt et al (2003) and Le Mignon et al (2009) consists in performing linkage analysis for the trait using only some of the animal subgroups of a F2 population generated on the basis of their transcriptome profiles. Such an approach can refine some QTL and reveal other ones.

This approach was applied to hepatic transcriptome profiles for 45 offspring of a chicken known to be heterozygous for a QTL for abdominal fatness (AF) on GGA5 at 168cM. 688 gene expressions significantly correlated to the AF trait were obtained using a recent method taking into account the gene dependence independently of the trait of interest (Friguet *et al*, 2009; see ISAG2010 Blum *et al*). A hierarchical cluster analysis using these 688 genes distinguished five groups among the 45 birds. After removing a subgroup of 7 animals, linkage analysis revealed another QTL on the GGA5 at ~102cM. These 7 animals presented the same paternal haplotypes, suggesting that the two QTL are in interaction. We show by different approaches (ANOVA, linkage analysis) a significant interaction between the two QTL. These results show the power of the approach: transcriptome data allows separating a population into genetically homogenous subgroups, revealing the complexity of the genetic component of the complex traits.

P4015 High density linkage disequilibrium maps of chromosome 6 and 14 in Chinese Holstein cattle

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The extent and patterns of linkage disequilibrium (LD) determine the feasibility of association studies to map genes that underlie complex traits. In this study, fifteen sire families with 2,042 daughters were analyzed for LD map of chromosome 6 and 14 in a Chinese Holstein population. The Illumine Bovine GeneChip with 50K SNPs was used to genotype 2,208 cattle in total. A total of 1,937, 1,300 SNPs on chromosome 6,14 were chosen after filtering out SNPs not meeting quality control criteria (e.g. call rate <0.95, MAF<0.05, HWE<0.00001). HAPLOVIEW under the four gamete rule was implemented to analyze the haplotype block structure. Results show 179 and 320 haplotype blocks were identified in BTA6 and BTA14, respectively, and in which the largest one spans 460kb in BTA14. The analysis of pairwise D' value revealed that LD on BTA6 decayed more rapidly than on BTA14 and LD on BTA6 is much less extensive than those in other published researches. The research regarding LDU is also investigated in our study. The results presented here can be applied in future single or haplotype association analysis in Chinese Holstein cattle.

P4016 Whole genome single nucleotide polymorphism association with residual feed intake measured under different diet regimes

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The use of marker assisted selection will enhance the genetic progress for residual feed intake (RFI) in the cattle industry. Given that growing cattle are fed on diets with different compositions in the production cycle, limited information exists on whether genetic markers associated with variation in RFI are sensitive to diet type. Our objective was to determine whether similar SNP are associated with RFI in steers fed either a grower diet or finisher diet. A feeding trial was conducted over three years. There were two feeding periods within each year where either a grower diet or finisher diet was offered such that 402 and 419 steers were present in the grower-fed and finisher-fed groups, respectively. Feed intake was measured with the GrowSafe system, and RFI calculated by linear regression. Genotyping was done using the Illumina BovineSNP50 Beadchip while Haploview was used to select 11,257 tag-SNP from 40,653 SNP (mean r^2=0.6). Whole genome association analyses were implemented within each group using Proc Mixed in SAS with sire and dam as random factors. A total of 596 and 661 SNP were associated ($p<0.05$) with RFI in the grower- and finisher-fed groups, respectively. The majority of these SNP were diet specific while only 75 SNP were common between the two groups. For these SNP common to both groups, the Pearson and rank correlations of the allele substitution effects were 0.88 and 0.91 respectively. Our results indicate that despite the existence of diet-specific SNP, some SNP are associated with RFI regardless of diet type.

P4017 A 6-bp deletion in the *TYRP1* gene causes the brown coloration phenotype in Chinese indigenous pigs

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Brown coat color has been described in Chinese Tibetan, Kele and Dahe pigs and shows considerable phenotypic variation, ranging from silver to dark brown. The genetic basis of the brown coloration remains unknown. Here we performed a genome-wide association study on Tibetan and Kele pigs using the Illumina PorcineSNP 60K Beadchips, revealing that the brown colors in Chinese breeds are controlled by the same locus on pig chromosome 1. By using a haplotype sharing analysis, we refined the critical region to a 1.5-Mb interval that encompasses only one pigmentation gene: tyrosinase related protein 1 (*TYRP1*). Mutation screens of sequence variants in the entire coding region of *TYRP1* revealed a strong candidate causative mutation (c.1484_1489del), which results in loss of methionine and glycine at amino acids 945 and 946 in a predicted transmembrane domain and is likely to be of functional significance. The deletion showed complete association with the brown coloration across Chinese Tibetan, Kele and Dahe breeds by occurring exclusively in brown pigs (n = 121) and lacking in all non-brown coated pigs (n = 745) from 27 different breeds. The findings allow genetic testing for breeders to select for or against brown coat color and provide the compelling evidence that the brown colors in Chinese indigenous pigs are caused by the same ancestral mutation in *TYRP1*. Moreover, to our knowledge, this study gives the first description of genome-wide association study identifying causal mutation for a monogenic trait in the domestic pig.