C-133

Copy number variation in cattle breeds

Alessandro Bagnato[1], Johanna Vilkki[2], Terhi Iso-Touru[2], Erika Frigo[1], Maria Cristina Cozzi[1], Maria Giuseppina Strillacci[1], Fausta Schiavini[1], Raphaelle Teresa Matilde Maria Prinsen[1], Mario Vevey[3], Attilio Rossoni[4], Luca Fontanesi[5], Marlies A. Dolezal[1,6]

[1]*Dipartimento di Scienze Veterinarie per la Salute, la Produzione Animale e la Sicurezza Alimentare, Università degli Studi di Milano, Italy*
[2]*Natural Resources Institute Finland, Helsinki, Finland*
[3]*Associazione Nazionale Allevatori Bovini di Razza Valdostana, Gressan (AO), Italy*
[4]*Associazione Nazionale Allevatori Razza Bruna Italiana, Bussolengo (PR), Italy*
[5]*Dipartimento di Scienze e Tecnologie Agro-alimentari, Alma Mater Studiorum-Università di Bologna, Italy*
[6]*Institut für Populationsgenetik Veterinärmedizinische University Wien, Austria*
Corresponding author: alessandro.bagnato@unimi.it

Detecting all classes of genetic variation in livestock species, such as cattle, is a pre-requisite to studying their association to traits of interest. Copy Number Variations (CNVs) are classes of polymorphic DNA regions including deletions, duplications and insertions of DNA fragments of at least 0.5 kb to several Mb, that are copy number variable when compared to a reference genome. CNVs can be identified using various approaches, among those the SNP array data are low cost, dense coverage, and high throughput. The aim of this study was to obtain a consensus genome map of Copy Number Variable Regions (CNVRs) in the Brown Swiss (dataset of 192 bulls), Red Pied Valdostana (dataset of 143 bulls) and Finnish Ayrshire (dataset of 243 bulls) cattle breeds all genotyped on the Illumina Bovine HD BeadChip, and two SNP based CNV calling algorithms. Brown Swiss cattle originated in the Swiss Alps, kept as a triple purpose breed. Once imported in the US, it was mainly selected for increased milk production. The Valdostana Red Pied cattle is the most common autochthonous dual purpose breed in the region Valle d'Aosta in Italy (13,000 animals in 2013, almost all of them registered in the Herd Book). The Finnish Ayrshire is the most common cattle in Finland. CNVs were called with the PennCNV and SVS7 software and were summarized to CNVRs at the population level as overlapping CNV calls within breed. PennCNV identified 2,377, 1,723 and 1,689 for the Italian Brown Swiss, the Red Pied Valdostana and the Finnish Ayrshire, respectively. SVS7 detected 370, 235 and 2,063 for the three cattle breeds. These regions were annotated with Ensembl v78 Bos taurus gene set (UMD3.1) and genomic regions harboring QTL for production and functional traits. The comparison among CNVRs here identified provided common regions in the breeds. The results of this study are a comprehensive genomic analysis of cattle CNVs derived from SNP data, which will be a valuable genomic variation resource and will enrich the bovine CNV map in the cattle genome, providing new information for association studies with traits included in the selection programs.

Acknowledgments
This study was funded by EC-FP7/2007-2013, agreement n°222664, "Quantomics".

C-134

Haplotype detection for monogenic factors in the Italian Holstein cattle breed

Raffaella Finocchiaro, Jan-Thijs van Kaam, Maurizio Marusi

Associazione Nazionale Allevatori Frisona Italiana, Cremona, Italy
Corresponding author: raffaellafinocchiaro@anafi.it

Italian Holstein genomic data-base has more than 125.000 genotyped animals with several SNPs panels, and every month this number is increasing. These genotypes are not only used for genomic breeding values, but are used also as parental confirmation and detection for herd book animals. Moreover the availability of these enormous amount of SNPs allows the reconstruction of several haplotypes. Recently we have derived information on 11 haplotypes of which 9 contain a lethal disorder and the other 2 haplotypes are for two variations of the polled gene. The concept is to scan imputed chromosomes for a haplotype in common between carrier animals. If a disorder is lethal than the haplotype containing the mutated gene should never occur in homozygous form except when recombined. First we introduced the haplotype detection for recessive factors affecting fertility, these haplotypes are indicated with HH1, HH2, HH3, HH4, HH5. For each of these five haplotypes carrier females risk an embryonic loss during gestation. Lately carrier status for Holstein haplotypes with mutations for brachyspina (HH0), bovine leukocyte adhesion deficiency (BLAD), complex vertebral malformation (CVM), mulefoot (syndactyly), and polledness (2 carrier haplotypes) have been introduced. In total 125.498 unique genotypes were used. Also identical twins, imputed animals which are not genotyped but have several genotyped relatives and some ancestors get a haplotype assigned. This leads to 152.322 animals with information. Around 77% of the animals are free of a lethal genetic disorders; the remaining part have a small probability to have an abortion or a genetic disorder. For recessive disorders, problems occur only in 25% of the matings between carriers. Table 1 reports in detail the different carrier haplotype frequencies detected in the Italian Holstein population. A monitored mating plan like the one offered by Anafi Webpac would help farmers to avoid undesirable genetic disorders. Furthermore AI centers can use haplotypes as an additional early-stage selection tool.