GENETICS AND GENOMICS

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Canine hip dysplasia - Understanding the genetic basis to apply Genomic Selection – A Master Thesis in Veterinary Medicine

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Canine hip dysplasia (CHD) is the most prevalent orthopaedic disease in large breeds of dogs such as the German Shepherd Dog, Golden and Labrador Retriever, Bernese Mountain Dog. It is characterized by an abnormal formation of the hip joints leading different degrees of laxity and subluxation with subsequent formation of progressive osteoarthritis. Nowadays the CHD diagnosis is carried out by physical examination, including the evaluation of hip pain, and the degree of hip laxity with the Ortolani test in young dogs, while in mature dogs the range of motion and pain of hip joint are evaluated. Moreover, the X-rays are essential to confirm the diagnosis and to establish the severity of the disease. Hip scoring (classes from A to E, according to increasing severity in the FCI classification) is a procedure used to determine the degree of hip dysplasia in dogs and it is the sum of the points awarded for each of the radiographic features of hip joints. The results of several studies reported in literature highlight that selective breeding programs, based on genetic and genomic information, are valid alternative to reduce the incidence of CHD in dogs. CHD is a complex trait, which expression is influenced by genetic, nutritional, environmental and hormonal factors. Several quantitative trait loci (QTL) on different chromosomes have been found to contribute to this trait expression in various dog breeds, including Labrador retrievers. The aim of this Master Thesis in Veterinary Medicine is to target re-sequence the genome of a sample Labrador retrievers dogs in a case/control (dogs classified as A/D-E) study of CHD under a selective DNA pooling approach (SDP). Blood samples will be collected during the routine CHD diagnosis practices and genomic DNA will be extracted with a commercial kit. The chromosome regions that will be investigated will be chosen on the bases of those reported in literature and associated with CHD. The resulting sequencing data will help to find new genomic/genetic variations which may be involved in the disease occurrence.

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Effect of genes related to meat quality traits on progeny of selected bulls of Italian Limousine breed

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The aim of this study was to analyze polymorphisms in candidate genes for meat quality traits in the Italian Limousine beef breed. Chemical (proximate analysis and fatty acid profile) and physical (sample cut, colour, pH, cooking loss, shear force and free water) traits from steaks of 97 steers progeny of 9 selected bulls were recorded. DNA was extracted from the same animals and analysed at 10 SNP mutations in 7 different genes: myostatin (MSTN), dyacilglycerol-O-acyltransferase (DGAT), calpain (CAPN), calpastatin (CAST), leptin (LEP), thyroglobulin (TG), fatty acid binding protein (FABP). Allele and genotype frequencies, heterozygosity, and Hardy-Weinberg equilibrium for each SNP were estimated. Haplotype reconstruction through Phase software was also performed for SNPs located on the same chromosome. The association among SNPs or haplotypes and the recorded traits has been performed through a linear mixed model incorporating the effect of the pedigree with the pedigreemm package in R. Two SNPs at myostatin gene (F94L and Q204X) and one located in the thyroglobulin gene (C422T) were fixed whereas the other SNPs presented a MAF 0.05. Fixation of the first two SNPs might be either the result of the selection program developed so far or the bias due to the small sample size. No significant departures from Hardy-Weinberg equilibrium were identified for the genotypes, except for the CAST282 located in the calpastatin gene where a moderate excess of heterozygous genotypes was observed. Accounting for haplotypes did not change the association results in any of the traits analyzed. Two out of three SNPs of the CAPN gene (CAPN316 and CAPN530) and one at CAST gene (CAST282) were associated with meat tenderness, free water content and fatty acid profile. Thus the combined effect of favourable alleles for CAPN and CAST markers resulted in improved meat quality traits providing further evidence that selection based on these markers might be feasible. There are also evidence that SNPs at FABP, DGAT and CAST genes were associated with fat traits confirming the important role of these genes in lipid metabolism of intramuscular fat. No significant association between LEP gene and meat quality traits has been found. Due to the moderate size of the sample is not possible to draw final conclusions but the result suggests to deepen the studies at the final aim of incorporating markers information in the selection criterion of Italian Limousine breed.