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ABSTRACTS

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caused by a point mutation located on Chromosome 9. The objectives of this work were the development of an alternative molecular assay for detecting the point mutation and the evaluation of the incidence of the disease in the Italian Bolognese population. To this aim, the High Resolution Melt Analysis (HRMA) was used to identify carrier (GA), clear (GG) and affected (AA) animals. Several 110 blood samples was collected from 42 males and 68 females, and the DNA was extracted. The samples represented about 2% of the living animals corresponding to 11% of the breeding subpopulation. A PCR was conducted in the presence of a dsDNA-binding dye followed by a melting step. The analysis is based on the detection of fluorescence changes associated with the release of dye following dissociation of the dsDNA into single strands. The output is a melt curve profile, which is characteristic of a specific genotype. The results showed that the causative mutation was present for 30% in heterozygosity (carrier) and for 1% in homozygosity (affected). The samples were further submitted to Sanger sequencing analysis. The results confirmed the totality of genotypes detected by HRMA thus providing for the validation of the HRMA protocol as a valid method for the detection of the mutation responsible for prcd-PRA. Moreover, given the number of animals sampled in the present work with respect to the current population, it is possible to conclude that the results are representative of the real incidence of the disease in the Italian Bolognese population.

Key Words: oculopathy, Bolognese, dog, prcd-PRA, HRMA

OP57 Analysis of clinical samples from Doberman and Toy Poodle dogs with a targeted next-generation genotyping system.
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Next-generation sequencing (NGS) is a powerful tool to study DNA or RNA samples. New methods and protocols based on NGS have been developed to carry out the analysis of genetic variation for animal parentage testing, disease screening and trait detection. Targeted NGS is aimed at achieving 'targeted enrichment' of genome subregions to reduced significantly the sequencing of genomic loci of interest, as well as costs and efforts, compared with whole-genome sequencing (WGS). We generated genotyping information of 387 targets from 95 clinical canine samples (76 Doberman and 19 Toy Poodle dogs) and 3 control samples using AgriSeq Targeted GBS. Based on these data, we calculated the exclusion power of 228 parentage markers with Cervus 3.0 software. Furthermore, we detected disease/trait markers presenting polymorphism and calculated their allele frequencies within each breed. In the case of parentage markers, the assigned parents showed a higher LOD score (>1.22 x10^{16}), and the available pedigree data of offspring agreed with the assigned parent information. Interestingly, full siblings were also assigned like parents. On the other hand, we found 19 polymorphic disease/trait markers in the total sample, 3 of which (progressive rod-cone degeneration, von Willebrand disease 1 and dilated cardiomyopathy) were validated by pyrosequencing with 100% concordance. The mutant allele for cone-rod dystrophy3 (CRD3) was found in both groups, a variant which had not been reported in either breed to date. Sequencing of genomic loci of interest, costs and efforts can be reduced significantly with targeted NGS as compared with WGS. The AgriSeq Targeted GBS is a very good alternative for the massive genetic evaluation of animal populations.

Key Words: dogs and related species, animal breeding, high-throughput sequencing (HTS), polymorphism, genomic selection

OP58 First steps in animal genetic testing in Bulgaria.
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Molecular genetic testing can be a powerful tool for identification of autosomal recessive mutations to avoid crossbreeding between carriers, inborn predispositions aiming to prevent or at least delay the development of a disease or eventually for determination of the sex of an individual. Here we resume our work representing as far as we know the first genetic testing experience of animals, more precisely companion animals and birds, in Bulgaria. Having a solid background in human molecular genetics, we decided to expand our interests and enter the field of animal genetics. So far, we have implemented basic genetic tests for variations with well-known correlation to a pathological condition and thus with a high practical value. The Multi-Drug Resistance Gene (*MDR1*, *ABCB1*) has been our first target. We successfully performed genetic testing for identification of the c.227_230delATAG mutation in a control group and identified the ABCB1–1? genotype in a pair of Australian Shepherd dogs – the one being a heterozygous and the other a homozygous carrier. Concerning the feline genetics, so far, we have focused our work on the Polycystic Kidney Disease (PKD). Optimization of the molecular genetics assay and application of the protocol to a control group of cats of different breeds has been executed. The c.10063C > A variant in the *PKD1* gene was as expected detected in Persian cats. Using a widely known primer set (Lyons et al., 2004) for PCR amplification and Sanger sequencing, we even observed an allele drop-out of the wild type allele reminding us of the necessity to be careful when implementing new protocols. Lastly but not least, we determined the sex of different species of birds: domestic chicken (*Gallus gallus domesticus*) and parrots (*Serinus canaria*, *Nymphicus hollandicus*, *Melopsittacus undulatus*). For the future, we plan expanding our activities in canine and feline genetics by identifying typical for certain breeds mutations. A test for Lagotto Storage Disease (LSD, Lysosomal Storage Disease) is currently in a process of introduction.

Key Words: cats, dogs, genetic disorder, avian, Bulgaria

Avian Genetics and Genomics

OP59 Initiative for Global Chicken Genome Project (GCGP).
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As the most abundant and widespread domesticates, the characterization of genomic diversity of chickens at a global scale are indeed required for the sustainable conservation and utilization of these valuable resources. The Kunming Institute of Zoology - Chinese Academy of Sciences (KIZ, CAS) has been leading a 2-phase international collaborative project to improve our current knowledge on chicken genomics. Through collaboration with the International Livestock Research Institute (ILRI), Zoological Survey of India, and the University of Oxford among many others, we have completed the phase one project in generating around 1000 genomes of chickens and wild junglefowls sampled from East Eurasia where the putative center of chicken domestication is located. The population genomic analyses revealed a remarkably complex picture of the early domestication and subsequent dispersal of chickens in this region. In the phase 2 project, the topic will be the genomic changes of chickens during their global dispersal out of the East Eurasia. We plan to generate at least 2000 genomes of different indigenous populations/ecotypes, improved lines, and fancy chickens from Asia, Africa and Europe. The genomic data will be important resourc-