

WBFSH Scientific Collaboration Department:

Summary of the webinar 'How to bring genomic development to horse breeding?' (held December 7th, 2021)

After a welcome and introduction by Karina Christiansen, the webinar started with some information from the scientific community, highlighting the points of contact with WBFSH. The member studbooks were made aware of the opportunities, the Horse Commission of the European Federation of Animal Science (EAAP-HC) can provide to them. Through its strong network, EAAP-HC can not only take up and promote topics of major practical importance for the annual scientific meetings, but also bring into touch studbooks and equine scientists from different fields to address specific questions. Spotlights from the annual conferences of the EAAP have become integral parts of the WBFSH general assembly. Supposed to link between science and practice, these reports can help keeping update with regard to developments in animal science which are important for horse breeding.

Over the last couple of years, the number of studies using so-called genomic data of horses has increased among those summarized by the EAAP-HC representative Kathrin Stock for the WBFSH member studbooks. The equine community can build upon improved understanding of how data on genetic markers spread over the whole genetic code can strengthen breeding programs and help breeders. However, transfer from science to practice has so far been rather slow, with currently just a very few routines for genomic evaluation and selection in horse breeding. The brief information on on-going research and current work towards more and improved genomically enhanced breeding applications in horses made it clear for the audience why the WBFSH Scientific Collaboration Department had chosen the title and structure of the webinar: Its second part focused on the options to use the routine parentage testing of horses as a straightforward entry of the studbooks to the genomics era.

The four speakers brought back to minds of the participants what had already in 2019 been summarized in the <u>fact sheet on parentage testing</u>, with the newly introduced webinar format allowing to explain in more detail what helps the studbooks in their decision making.

The established system of parentage testing in horses uses a type of molecular genetic markers called microsatellites (MS) or short tandem repeats (STRs). These markers proved suitable and robust for routine testing in the molecular genetic laboratories and are what makes the individual horse's 'DNA profile' used to compare it with the respective profiles of the declared parents. Nevertheless, the usability of STRs for nothing else but parentage



testing implies high motivation to establish systems which are based on markers usable for multiple purposes: single nucleotide polymorphisms (SNPs). Thanks to the technological progress and worldwide extensive use of SNP based technology, SNP genotyping is nowadays affordable standard service in many larger molecular genetic laboratories. Commercially available testing systems, referred to as SNP array or SNP chip, include 10,000s of markers and allow horse breeding to benefit from investing in what can be used for currently existing routines (like parentage testing and related services) and potential future routines (genomic evaluation and selection).

The webinar contributions from the Netherlands, France and Germany provided easy-tounderstand information on how the transition to SNP based applications could be made. And most valuably, experiences from the introduction of routine SNP genotyping in replacement of the former standard technology based on STRs were shared. Key messages of the presentations were:

In a situation where parentage testing of horses often requires that triplets, so progeny plus indicated parents, are genotyped, direct transition to SNP genotyping of all newly sampled and analyzed horses may be considered. The basic principle of marker comparison across generations requires that the genotype data of the same type of genetic markers is available for foal, sire and dam. This implies for studbooks with a lower the quota of STR genotyped older horses that investments in molecular genetic analyses may preferably target at SNPs. Costs per genotyping are nowadays similar for genome-wide SNPs than for STRs, so the much higher versatility of SNP genotype data may make a reasonable cost-benefit ratio for SNP genotyping of triplets. (laboratory analyses for 3 horses, costs for 3x SNP genotyping)

KWPN had chosen that way with direct switch from STR based to SNP based parentage testing, so comparison between offspring SNP genotypes and parental SNP genotypes for a pre-defined set of SNPs. With this procedure, the studbook was actually ahead of the ISAG which focuses for the horse still on the STR based parentage testing, although first preliminary SNP based ring-tests have been organized.

In a situation where parentage testing of horses usually requires only the progeny to be genotyped, because coverage of the breeding population with MS genotype data is high, there are two options for the transition to routine SNP genotyping.

(a) Newly sampled horses may be genotyped for both STRs and SNPs to get the former for regular STR based parentage testing and the latter for future SNP based routines. (laboratory analyses for 1 horse, costs for 1x STR genotyping plus 1x SNP genotyping)



(b) If a validated routine for deriving STR genotypes from SNP genotypes is available, newly sampled horses may be genotyped for SNPs only. A statistical method called imputation is then applied to obtain imputed STR genotypes which can subsequently be used for comparison with parental STR profiles. To account for the remaining uncertainty of the estimation procedure, indications of Mendelian conflicts should be verified by STR genotyping in the laboratory. If reliability of the imputing algorithm is high and the quota of re-analysis of samples is accordingly low, SNP based imputation of STRs a cost efficient method for the transition. (laboratory analyses for 1 horse, costs for 1x SNP genotyping; additional STR genotyping costs for <5% of the horses)

There are two countries that have independently developed imputing routines: France and Germany. Motivation and starting conditions were similar, with good population coverage and large amounts of historical STR data on the one hand and thousands of horses with both STR and SNP genotype data (required for setting up and training the imputation algorithm) on the other hand. Important differences were much larger heterogeneity of SNP panels used and of breeds included in the French imputation system which has not yet been practically implemented. Thanks to a strong training set from sport horse breeds with high consistency of SNP data, the German imputation system could already be implemented in routine.

The closing presentation from Sofia Mikko, representing the International Society for Animal Genetics (ISAG), clarified the role of ISAG in connection with international parentage testing and extended the overview of options. The well-established system of STR based parentage testing is still standard for horses, and many laboratories from all over the world are participating in the ring tests (quality control). SNP based parentage testing has become standard in other species, but not yet in the horse. Concerning the methodology to generate SNP data, the widely used SNP chip technology was explained and genotyping-by-sequencing (GBS) was presented as possible alternative. Pros and cons from technical and practical perspectives were discussed, underlining the importance of considering the whole system (molecular genetic laboratories, data bases / data service providers, computation centers, studbooks; national and international data use and exchange) and long-term strategic planning of individual studbooks and horse breeding. The discussion of the participants of the webinar was very helpful in this respect, because it allowed taking a closer look at specific technical as well as practical aspects of the overall topic. The open exchange of considerations and first practical experiences with the transition to routine use of genomic data in horse breeding was very much appreciated by the participating studbooks and their collaboration laboratories and considered very helpful for decision making. Accordingly, the WBFSH webinar was obviously successful in promoting the future-oriented development of sport horse breeding.