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Runs of homozygosity and genomic inbreeding in the German riding horse population

Project motivation / Introduction

- routine genome-wide SNP genotyping of all registered foals in most German Warmblood studbooks
 - related to introduction of SNP-based imputation of STRs used for parentage testing
- broad availability of genome-wide SNP genotypes implying suitability for analyses of pedigree structure and inbreeding high coverage of cohorts of riding horses from 2021 onwards

Conclusion

moderate mean genomic inbreeding (F_{ROH} of 6.8%) in the German riding horse population

low frequencies of long ROH

- smaller role of recent inbreeding than historical inbreeding
- monitoring of genetic diversity as integral part of responsible breeding programs
 - reliable assessment of diversity measures by genomic tools

objective: using genome-wide SNP genotypic data to derive key genomic parameters and assess

- genomic inbreeding _____
- development of genetic diversity ____



results of pedigree structure analyses as basis of new tools for support of mating planning and improved breed management





Results

Table 1: Mean F_{ROH} with standard deviation (std.), minimum and maximum for N = 63,196 German Warmblood horses born 2021-2023, determined with different lengths of DNA stretches considered as ROH.

window size [kb]	mean	std.	minimum	maximum
500	0.0783	0.0229	0.0005	0.5424
1,000	0.0777	0.0228	0.0005	0.5256
1,500	0.0745	0.0225	0.0000	0.4478
2,000	0.0679	0.0219	0.0000	0.3555
3,000	0.0511	0.0203	0.0000	0.3140
4,000	0.0388	0.0185	0.0000	0.2899
5,000	0.0298	0.0169	0.0000	0.2722
6,000	0.0229	0.0153	0.0000	0.2628
10,000	0.0090	0.0101	0.0000	0.2306



ROH = Runs of homozygosity, F_{ROH} = genomic inbreeding coefficient based on ROH (McQuillan^[b])

Material and methods

70K+ SNP genotypes of N = 63,196 German Warmblood horses were used to determine **runs of homozygosity (ROH)** with **PLINK** software^[a] and the following functions / settings:

- -- homozyg
- -- homozyg-window-kb (different windows see Table 1)
- -- homozyg-snp 25 (minimum number of SNPs in one ROH)
- -- homozyg-window-het 0 (no heterozygotes allowed in ROH)

Figure 1: Principal component plot from all horses in the central genome database (N=78,779); PC1 (PC2, PC3) = Vector to the 1st (2nd, 3rd) principal component

Breeds and breed groups of equids as indicated in figure 1:

- = Oldenburg OL
- = Oldenburg Internat. (jumping) OS
- = Holstein HOL
- = Westphalian WESTF
- = Trakehner TRAK
- = Hanoverian HAN
- DSP = German Sport Horse
- Pony = different pony breed
- XXOX = Thoroughbred and Arabians
- COLD = different heavy horse breeds (draught horses, Coldblood)
- other = Warmblood horses with foreign ID and horses of unknown breed

N = 57,286 quality controlled autosomal SNPs entered the analyses

calculation of **genomic inbreeding** for each individual and each scenario (different window sizes) according to McQuillan^[b]:

 $F_{ROH} = \frac{total \, length \, of \, all \, ROH}{autosomal \, genome \, length \, covered \, by \, SNPs}$

afterwards: mean F_{ROH} over all horses for each scenario (Table 1)

Literature / Acknowledgements

^[a] Purcell, S., et al. (2007): PLINK: a toolset for whole-genome association and population-based linkage analysis. Am. J. Hum. Genet. 81, 559-575. ^[b] McQuillan, R., et al. (2008): Runs of homozygosity in European populations. Am. J. Hum. Genet. 83, 359-372.

Thanks to the studbooks and IAFH for providing the data



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