

Rule-based autozygosity detection (PLINK)

Use of differing criteria

Outcomes: non comparable across studies

Objective

Compare autozygosity outcomes from PLINK⁽¹⁾ and ZOOHOH⁽²⁾ software

Materials & Methods

Data

- 40,753 SNPs on 46 sheep
- 34,066 SNPs on 51 pigs
- 26,932 SNPs on 6 horses (array)
- WGS-data on 6 horses

PLINK

- --homozyg-window-snp 50
 - --homozyg-window-het 3, etc.
 - $F_{ROH} = \frac{\sum \text{length of all ROHs in an individual}}{\text{Length of autosome}}$
- ROH: Runs of homozygosity

ZOOHOH

Hidden Markov model-based approach identifies homozygous-by-descent segments.

Inbreeding coefficients (F) and number of ROH segments (nROH) detected in the sheep, pigs and horses.

Species	Mean F(%)		nROH	
	PLINK	ZOOHOH	PLINK	ZOOHOH
Sheep	8.1 ^a	11.5 ^b	1,282	3,296
Pig	26.5 ^a	31.0 ^a	2,805	5,267
Horse (array)	11.1 ^a	16.7 ^a	177	940
Horse (WGS)	21.0 ^a	21.0 ^a	12,530	38,838

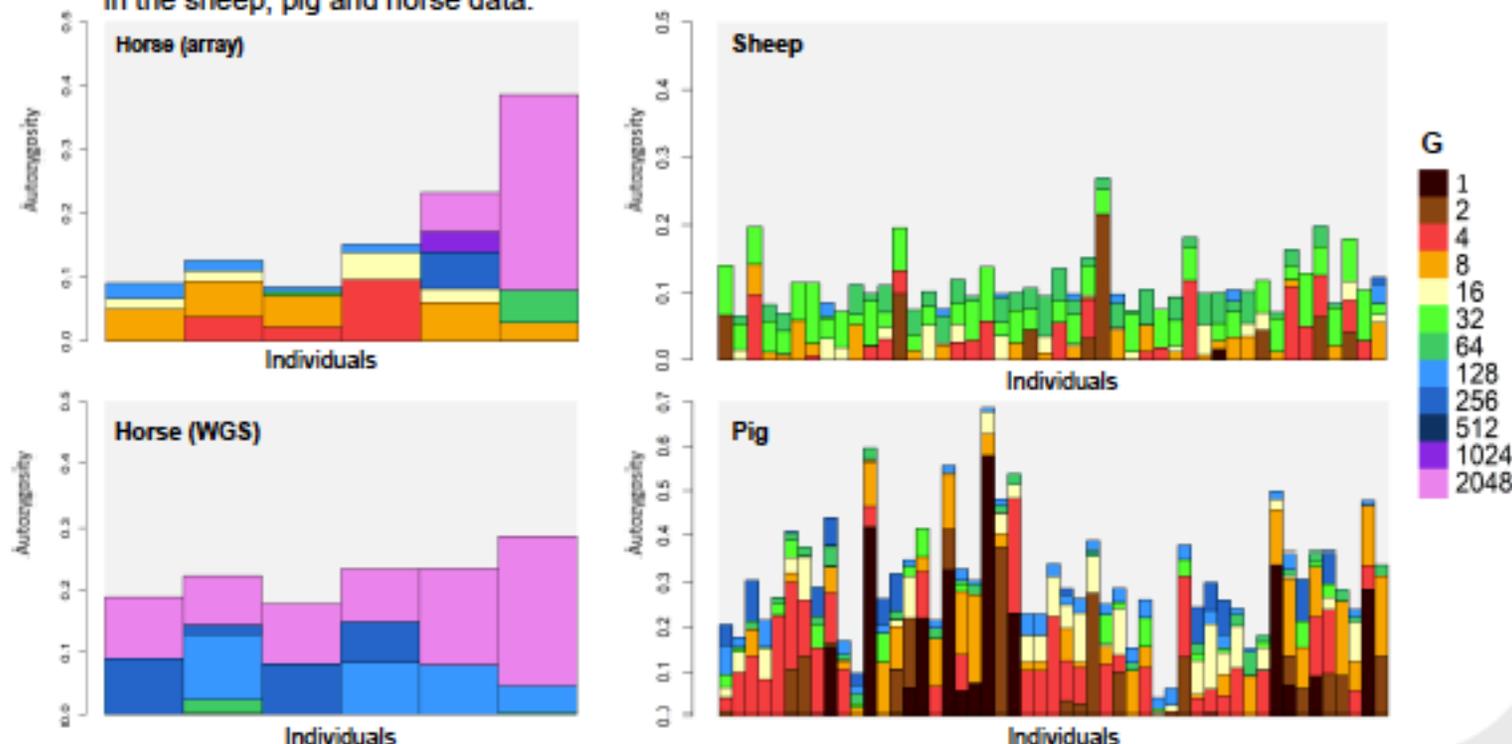
Within species/data type, means with common superscripts are not significantly different ($P > 0.05$).

References:

- (1) Purcell, S., B. Neale, K. Todd-Brown, L. Thomas, M. A. R. Ferreira, D. Bender, J. Maller, P. Sklar, P. I. W. De Bakker, and M. J. Daly. 2007. PLINK: a tool set for whole-genome association and population-based linkage analyses. *Am. J. Hum. Genet.* 81:559–575.
- (2) Bertrand, A. R., N. K. Kadir, L. Flori, M. Gautier, and T. Druet. 2019. RZooRoH: An R package to characterize individual genomic autozygosity and identify homozygous-by-descent segments. *Methods Ecol. Evol.* 10:860–866.

Results & Discussion

Tracing of individual's realised ZOOHOH-based autozygosity to different ancestral generations (G) in the sheep, pig and horse data.



Conclusion

ZOOHOH-based analysis provides valuable information for the optimisation of rule-based ROH/autozygosity detection.