



Effective population size and genomic inbreeding in Slovak Pinzgau cattle



**NINA MORAVČÍKOVÁ, RADOVAN KASARDA,
VERONIKA KUKUČKOVÁ, ONDREJ KADLEČÍK**

**DEPARTMENT OF ANIMAL GENETICS AND
BREEDING BIOLOGY, SLOVAK UNIVERSITY OF
AGRICULTURE IN NITRA, TR. A. HLINKU 2, 949
76 NITRA, SLOVAK REPUBLIC**



Introduction

- ❖ Why **Pinzgau** cattle?
- ❖ **Breeding strategy** → to minimize level of inbreeding
→ to increase effective population size
- ❖ **N_e and F** → prediction of genetic variation loss
→ rate of increase in inbreeding
→ population history

Objectives



In Slovak Pinzgau population to estimate based on high-density data :

- ❖ effective population size,
- ❖ genomic inbreeding.



Material & Methods

Analysed individuals

- ❖ 152 animals (37 sires, 35 nucleus cows, and 80 cows) → the nucleus of Pinzgau breed kept in Slovakia

Genotyping data

- ❖ Illumina BovineSNP50 BeadChip V2

Quality control

- ❖ only autosomal SNPs with known chromosomal position
- ❖ SNPs with call rate \uparrow than 90%
- ❖ MAF frequency \uparrow than 0.05
- ❖ HWE limit of 1×10^{-5}

Material & Methods



Inbreeding coefficient (F_{ROH})

- ❖ expressed as the length of the genome present in ROH divided by specified length of the autosomal genome covered by SNPs
- ❖ **3 length categories** ($F_{ROH>4\text{ Mb}}$, $F_{ROH>8\text{ Mb}}$, $F_{ROH>16\text{ Mb}}$)
- ❖ **Criteria for the ROH segments:**
 - ❖ the minimum number of SNPs in ROH: 15
 - ❖ the minimum length of ROH: 1 Mb
 - ❖ minimum density of one SNPs on every 100 kb
 - ❖ maximum gap between consecutive SNPs of 1 Mb
 - ❖ one heterozygous call allowed for length >16 Mb, one missing call allowed for length >4 Mb, >8 Mb and 4 for >16 Mb

Material & Methods



Ancestral and recent effective population size (N_e)

- ❖ according to Corbin et al. (2012)
- ❖ **historical effective population size** → expressed as a function of time and physical genetic distance between two loci
- ❖ **current N_e** → predicted based on the linear regression performed on estimates obtained for the past generations (N_{eLD10} to N_{eLD60})

Results & Discussion

ROH greater than 4 MB → 2.22%

ROH greater than 16 MB → 0.81%

recent inbreeding in analysed population

Summary statistics of F_{ROH} within each of analysed group

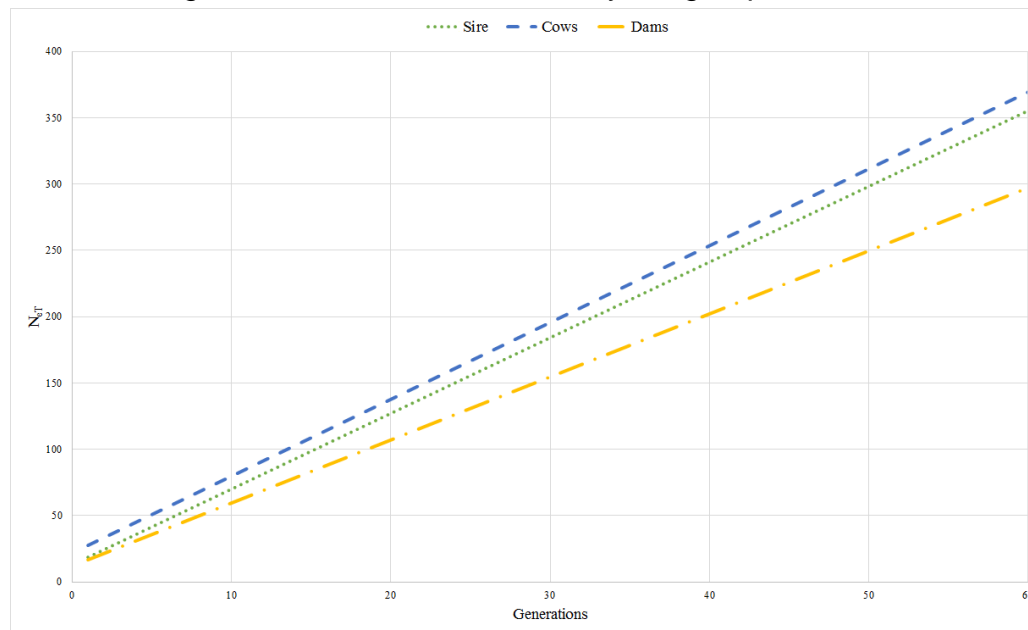
Inbreeding coefficient	Mean	Standard deviation	Lower 95% CI	Upper 95% CI	Range
Cows					
$F_{ROH > 4 \text{ Mb}}$	0.024	0.022	0.019	0.029	0.000 – 0.133
$F_{ROH > 8 \text{ Mb}}$	0.016	0.020	0.012	0.020	0.000 – 0.131
$F_{ROH > 16 \text{ Mb}}$	0.008	0.016	0.005	0.012	0.000 – 0.105
Dams					
$F_{ROH > 4 \text{ Mb}}$	0.017	0.016	0.012	0.023	0.000 – 0.072
$F_{ROH > 8 \text{ Mb}}$	0.011	0.013	0.006	0.015	0.000 – 0.062
$F_{ROH > 16 \text{ Mb}}$	0.007	0.012	0.003	0.012	0.000 – 0.048
Sires					
$F_{ROH > 4 \text{ Mb}}$	0.023	0.019	0.017	0.030	0.000 – 0.072
$F_{ROH > 8 \text{ Mb}}$	0.016	0.015	0.011	0.021	0.000 – 0.048
$F_{ROH > 16 \text{ Mb}}$	0.009	0.011	0.005	0.012	0.000 – 0.033

Results & Discussion

Historical N_e (N_{eT}) \rightarrow linear decrease within each of group

Predicted current N_e across animals \rightarrow 30.29 (90% CI 28.95-33.46)

The N_e trends across generation within each of analysed group based on the linear regression



Conclusions



- ❖ The estimates of historical effective population size indicated the linear decrease within each of analysed group.
- ❖ The predicted current N_e across all of animals clearly demonstrated the endangered status of Slovak Pinzgau population that was previously described based on both pedigree and genomic information.

Conclusions



- ❖ The results of this study mainly reflected the need for constant monitoring to increase population size without reduction of genetic diversity due to inbreeding.

- ❖ The study will contribute to the conservation management strategy of Pinzgau cattle in Slovakia.

Thank you for your attention!

Acknowledgement

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