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Genomic patterns of homozygosity in Istrian sheep

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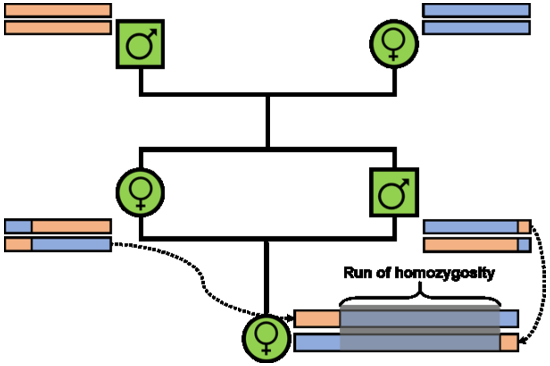
INTRODUCTION

- **Runs of homozygosity (ROH)**
  - Long homozygous regions → inherited from common ancestor
  - Long ROHs – recent inbreeding
  - Short ROHs – ancient inbreeding
  - Information → **history** & **genetic profile**
- **Istrian sheep**
  - Herdbook sheep (N=1,302)
  - Milk & meat production



AIM

- To determine: level of inbreeding – Istrian sheep:
  - F_{ROH} – by homozygous segments
  - F_{HOM} – by homozygous genotype count

Material & Methods (1)

- 719 Istrian sheep → OvineSNP50 BeadChip

- PLINK software
  - Autosomes
  - Quality control:

    MAF<0.05
    call rate: SNP 0.10 & animal 0.05
    GC<0.7, GT<0.4

656 sheep
38,351 SNPs

Material & Methods (2)

- **Defining sliding window** (*Lencz et al. 2007, Purfield et al. 201*
  - No SNP window (44); No het (0) & No miss SNP (1)

$$L = \frac{\log_e \frac{\alpha}{n_s n_i}}{\log_e (1 - het)}$$

- **Identifying ROH** *(Ferenčaković et al., 2013; Meyermans et al., 2020)*
  - Homozyg Kb (1 Mb); SNP ROH density (1 SNP/250 Kb)
  - SNP gap (1 Mb); Homozyg SNP (44)
  - Homozyg het (0); Homozyg miss SNP (0) - less than 4 Mb
- **ROH length category** (1-4 Mb, 4-8 Mb, 8-16 Mb, >16 Mb)
- $F_{ROH\ (>4Mb)} = \dfrac{\sum_i lenght\ (ROH_i)}{L\ (total\ length\ of\ genome\ covered\ by\ SNP)}$
- $F_{HOM} = \dfrac{OBS\ homozygous\ genotype\ count - EXP\ count}{total\ observations - EXP\ count}$

Results

- Total of 15,817 autosomal ROHs ranging from 1.9 to 70.1 Mb
- ~24 ROHs per animal with an average length of ~8.12 Mb
- Genomic $F_{ROH>4Mb}$ = **7.5%** and F_{HOM} = **6.0%** → (r=98%)

ROH Category	Frequencies (%)	Length proportion of category/total lenght of the genome covered by ROHs (%)
1-4 Mb	5.6	9.4
4-8 Mb	**51.4**	**29.2**
8-16 Mb	**31.4**	**34.5**
> 16 Mb	11.6	26.9

Conclusion

- F_{ROH} → attention in planning future matings to **decrease inbreeding** below the conventionally accepted level (6.25%)
- Results are a reliable source of information for developing **basic guidelines** for implementation of **optimum contribution selection**

THANK YOU FOR ATTENTION

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