

# Genome-wide scan for selection signatures within two local cattle breeds in Slovakia

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#### Introduction

#### Selection signatures

- ✓ genetic changes that shaped the genome due to natural as well as artificial selection
- ✓ insight into the genetic adaptation to specific environments and history of selection for economically important traits
- ✓ domestication-related loci, immunity-related loci, loci associated with desired phenotypic traits...
- Why did we choose Slovak Spotted and Slovak Pinzgau cattle?

#### **Objectives**

Detection of selection signatures in the genomes of Slovak Spotted and Slovak Pinzgau cattle

Identification of genomic regions subjected to selection histories of those local populations

Characterization of regions located directly or close to QTLs and/or genes associated with economically important traits

#### **Material & Methods**

Analysed population (overall 236 animals)



Genotyping of animals: 2 SNP platforms Illumina BovineSNP50v2 BeadChip (54 609 SNPs) ICBF International Dairy and Beef v3 (53 262 SNPs)



### **Material & Methods**

# Quality control of genotyping data

## PLINK 1.9

- Consenzus map  $\rightarrow$  40,033 SNPs (only autosomal SNPs)
- Quality control criteria:

missing genotypes per sample max. 10 % min. SNPs call rate 90 % min. MAF 0.01

• Final database  $\rightarrow$  **39,261 SNPs** 

### **Material & Methods**

## Analysis of selection signatures

Genome-wide scan of Wright's F<sub>ST</sub> index

Variation in linkage disequilibrium (LD)

Integrated Haplotype Score (iHS) statistic

#### **Characterization of regions under selection**

- Genome map viewer, NCBI (Btau 5.0.1)
- Bovine QTL database

#### **Material & Methods**

# **Genome-wide scan of Wright's F**<sub>st</sub> index

- PLINK 1.9
  - according to Weir and Cockerham (1984) for each syntenic loci
  - F<sub>ST</sub> values averaged over 10 consecutive SNPs
  - > Genomic region under selection  $\rightarrow$  top 0.1 %

### **Material & Methods**

# Variation in linkage disequilibrium (LD)

- VarLD (Ong and Teo, 2010)
  - differences in genome-wide LD patterns between populations over sliding windows of 50 SNPs
  - ➤ Genomic region under selection → 0.01, 0.1, 1, and 5 percentile of signals

### Material & Methods

# Integrated Haplotype Score (iHS) statistic

- Haplotype reconstruction according to Scheet and Stephens (2006) → fastPHASE
- iHS score calculation
  - ➤ according to Voight et al. (2006) for each SNPs → R package rehh
  - > status of the ancestral and derived alleles → Rocha et al.
    (2014)
  - iHS score averaged into non-overlapping 500 kb segments across the genome
  - ▶ Genomic region under selection  $\rightarrow$  top 0.1 %

Rocha D, Billerey C, Samson F, Boichard D, Boussaha M. 2014. Identification of the putative ancestral allele of bovine single-nucleotide polymorphisms. J Anim Breed Genet, 131, 483-486.

Scheet, P., Stephens, M. 2006. A fast and flexible statistical model for large-scale population genotype data: Applications to inferring missing genotypes and haplotypic phase. American Journal of Human Genetics, 78, 629-644.

Voight, B. F., Kudaravalli, S., Wen, X., Pritchard, J. K. 2006. A map of recent positive selection in the human genome. PLoS Biology, 4, e72.

#### Results

# Genome-wide scan of Wright's $F_{sT}$ index

Strongest signals – 8 regions (BTA5, BTA6, BTA15) Longest region BTA6 Shortest region BTA5



Genome-wide distribution of  $\mathrm{F}_{\mathrm{ST}}$  values with corresponding boxplot

#### Results

# Variation in linkage disequilibrium (LD)

#### Strongest signals – 10 regions (BTA4, BTA5, BTA6, BTA7, BTA11, BTA12, BTA20, BTA22, BTA23) Longest region BTA5 Shortest region BTA11



Differences in genome-wide LD patterns between populations (upper abline is showing the genome-wide significance threshold)

#### Results

# **Integrated Haplotype Score (iHS) statistic** Strongest signals

- Slovak Spotted: 5 region (BTA5, BTA6, BTA11)
- Slovak Pinzgau: 7 region (BTA5, BTA7, BTA9, BTA15, BTA18)

Genome-wide plot of the iHS score averaged for 500 kb



#### Results

#### Genomic regions showing the strongest signal of selections across applied approaches

BTA	Start position (Mb)	End position (Mb)	Length (Mb)	No. of genes	QTL traits
4	44.09	46.82	2.74	27	somatic cell score
5	59.60	60.64	1.04	36	
5	63.65	68.01	4.36	55	dressing percentage, birth weight, longissimus muscle area, backfat EBV, follicle stimulating hormone concentration, twinning rate
5	70.31	76.92	6.61	95	
6	38.58	43.18	4.60	15	birth weight, stature, strength, daily gain, milk yield, protein and fat percentage, protein and fat yield,
6	62.09	73.09	11.01	105	rump width, suspensory ligament, teat placement, foot angle, quality of foot and leg, quality of udder, marbling score, milk yield, protein percentage and yield, fat yield
6	109.87	111.88	2.01	11	protein yield, fat percentage
7	42.06	44.92	2.86	110	
11	23.97	24.38	0.41	1	yearling and weaning weight
12	25.79	27.91	2.11	10	milk yield, protein and fat yield, ribeye muscle area, yield grade
15	30.46	35.20	4.74	51	
15	42.92	48.07	5.15	157	estimated kidney, pelvic and heart fat
15	51.53	55.64	4.11	109	meat tenderness
20	4.61	6.18	1.56	22	birth weight, protein and fat yield
22	32.76	34.64	1.88	7	
23	30.44	30.97	0.53	98	protein percentage, marbling score

#### Conclusion

- strongest signal of selection  $\rightarrow$  16 genomic regions
- common signals → genes related to milk production traits, beef quality traits, somatic cell score, coat colour pattern
- genomic regions under intensive selection pressure mostly linked to milk production and muscle development → dual-purpose performance
- applied approach → alternative strategy to identify the genomic regions and genetic markers related to traits with biological and economical importance

# Thank you for your attention!

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