

The exploration of Single Nucleotide Polymorphisms density in *Bos taurus* genomes reveals their role in the immune response



M. Frąszczak¹, M. Mielczarek^{1,2}, P. Grzesiak¹, J.Szyda^{1,2}

¹Biostatistics group, Wrocław University of Environmental and Life Sciences

²National Research Institute of Animal Production



MATERIAL

- Whole genome sequence with IlluminaHiSeq2000
- 7-28 x genome average coverage
- 48 Brown Swiss (BSW) bulls

METHODS

Bioinformatics pipeline

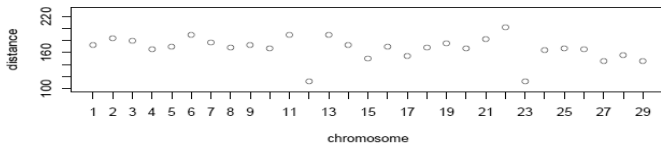
- Alignment to UMD3.1 (BWA-MEM)
- Post alignment filtering (Picard & Samtools)
- Variant calling (GATK)
- Functional annotation (VeP)

Statistical analysis

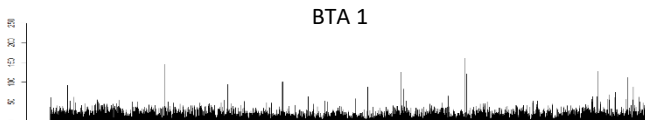
- Descriptive statistics
- Kolmogorov – Lilliefors test
- Kruskal – Wallis and Nemenyi tests
- χ^2 test
- Enrichment analysis

RESULTS

The average distance between neighbouring SNPs (in bp)



SNP density per 1000bp



BTA 1



BTA 12

Inter SNPs distance distribution

	BTA	P-value*
gr 1	1, 3, 13, 20, 24, 25,26	0.3191
gr 2	2, 6, 9, 11, 22	0.1562
gr 3	7, 8, 10, 14, 16, 19, 21, 28	0.5471
gr 4	5, 15, 17, 18, 27, 29	0.1433

* P-value – Kruskal-Wallis test

Without group – BTA 4, 12 and 23

Functional annotation

73% of SNPs – intergenic regions

0.01% of SNPs – exons

The most dense regions:

20 genes (6 on BTA12) with more than 200 SNPs in coding regions

54 genes (17 on BTA4) with average density of SNPs in exons <11bp.

Biological processes overrepresented among SNP-dense genes:

- antigen processing and presentation of endogenous peptide antigen via MHC class I via ER pathway, TAP-independent (GO:0002486)
- antigen processing and presentation of endogenous peptide antigen via MHC class Ib (GO:0002476)
- positive regulation of T cell mediated cytotoxicity (GO:0001916)
- immune response (GO:0006955)

CONCLUSIONS

- **SNPs are not uniformly distributed along the genome**
- **The distances between neighboring SNPs are not identically distributed in each chromosome, but 4 groups of BTAs with the same distribution were identified based on SNP density**
- **Biological processes related to immune system response were overrepresented among SNP-dense genes**

THANK YOU FOR YOUR ATTENTION

Acknowledgements: 

