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# THE IMPACT OF ARTIFICIAL SELECTION ON RUNS OF HOMOZYGOSITY IN SLOVAK SPOTTED AND PINZGAU CATTLE

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## ABSTRACT

The aim of this study was to analyse the distribution of runs of homozygosity (ROH) across the genomes of Slovak Spotted and Slovak Pinzgau cattle and to describe the autozygosity islands resulting from the selective breeding for traits of interest during the development of these breeds. The genome-wide data for a total of 236 animals were obtained by using two platforms: Illumina BovineSNP50v2 BeadChip and ICBF International Dairy and Beef v3. After quality control, the database of genotyping data consisted of 39,261 common SNPs across both breeds that covered overall length 2,497,077 kb of the genome with average distance between adjacent SNPs 63.67 kb. The ROH segments were defined as genomic regions with 15 or more consecutive homozygous calls with maximum gap between SNPs of 1 Mb and minimum density of one SNPs on every 100 kb. The distribution of ROH was analysed for five length categories (> 1 Mb, > 2 Mb, > 4 Mb, > 8 Mb, and > 16 Mb). The results showed that the ROH segments were present across the genome of all animals, with the average number of 54.59 ± 18.58 segments and the average length of 130.33 ± 58.40 Mb. The short segments (> 1 Mb) were the most frequent through the genomes and accounted for 70.26 % (Slovak Spotted) or 65.99 % (Slovak Pinzgau) of all segments detected. Thus, our results indicated that on average 6.11 % (Slovak Spotted) and 4.72 % (Slovak Pinzgau) of the genomes are autozygous. Moreover, the proportion of ROH > 16 Mb revealed that on average 0.45 % of the Slovak Spotted and 0.88 % of the Slovak Pinzgau genomes could be affected by recent inbreeding. Despite the fact that the distribution of ROH differentiated between breeds, the major fraction of chromosome residing in ROH was observed on BTA6 (13.49 % resp. 14.26 % of autosomal length in ROH). In this region we identified various QTLs and genes responsible for milk production (CSN1S1, CSN1S2, CSN2, CSN3), and coat colour patterns (KIT). Generally, our results confirmed that the regions displaying autozygosity in Slovak Spotted and Slovak Pinzgau cattle are linked mostly to milk production and muscle development thus ensuring selection for dual-purpose performance.

**Key words:** autozygosity islands; high-throughput SNP platforms; dual-purpose cattle; selection signatures

## INTRODUCTION

Slovakia has a long tradition in breeding dual-purpose cattle, namely the Slovak Spotted and Pinzgau breeds. Both of those breeds, whose origin is composite of autochthonous Carpathian Red (extinct) and Carpathian Grey (extinct) from the 17th to the 18th century as well as Swiss Simmental and Austrian Pinzgau from the 19th century common in Austro-Hungarian Empire, belong to the main cattle breeds of national interest in Slovakia (Kasarda *et al.*, 2015). First imports of Pinzgau and Simmental purebred animals were organized long time ago before 1894 when system of cattle recording has started in the territory of Slovakia. Over the following decades, the population sizes of both breeds have been improved and in 1958 they were officially accepted as Slovak Spotted and Slovak Pinzgau cattle. The population sizes reached maximum in 1975 and 1978 for Slovak

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Spotted and Slovak Pinzgau, respectively. However, due to post-1990 changes of economic conditions the population size of both breeds has significantly decreased mainly due to the transformation processes in agriculture and utilization of Holstein cattle for crossbreeding (Kadlečík *et al.*, 2013). From long-term perspective, the population size is unfavourable mainly in case of Slovak Pinzgau cattle, which is registered by the UN FAO as threatened with extinction and it is classified as Animal Genetic Resource (AnGR) since 1994 (Kadlečík *et al.*, 2008).

Globally, local breeds are rarely subject to modern selection techniques. However, selection programs will be required if local breeds are to remain a viable livelihood option for farmers. Selection in such small populations needs to take into account accurate inbreeding control that is one of the most important criteria in the evaluation of the degree of endangerment of the given breed (Gandini *et al.*, 2014). It is generally accepted that the population is endangered if the increase in inbreeding in the population per generation is higher than 1 % (Kasarda and Kadlečík, 2007). It should be noted also that the rate of inbreeding in cattle still has increasing tendency and there is a strong correlation between level of inbreeding and reduced fitness. Moreover, the high level of inbreeding and reduced variability in populations will result in inbreeding depression and reduced selection response in breeding programs. Thus, maintaining genetic diversity is crucial in cattle breeding populations (Zhang *et al.*, 2015).

Traditionally, the inbreeding coefficient is estimated by the degree of parental relatedness based on the pedigree data, while the genomic inbreeding is based on the proportion of the genome that is autozygous. Moreover, pedigree-based inbreeding is based on Mendelian sampling probabilities, so that the inbreeding coefficients of full-sibs are always identical. Using pedigree information for calculation of the level of inbreeding usually underestimates the true inbreeding coefficient mainly due to incomplete pedigree information, especially for distant generations (Zhang *et al.*, 2015; Forutan *et al.*, 2018). Calculating the inbreeding coefficient based on genomic data is more accurate for estimation of genome autozygosity and for detection of both past and more recent inbreeding effects than are estimated from pedigree data. The better results of genomic inbreeding coefficient suggest that it can be used to infer information about the history and inbreeding levels of a population in the absence of genealogical information (Peripolli *et al.*, 2017).

The application of high-throughput single nucleotide polymorphism (SNP) platforms allows for determination of autozygous segments based on the identification of runs of homozygosity (ROH) genotypes (Kim *et al.*, 2013), whose frequency, size and distributions in the genome are affected by many factors such as artificial selection, recombination rate, linkage disequilibrium and population structure or mutation rate (Peripolli *et al.*, 2017). Generally, the ROH are defined as continuous homozygous segments that are common in individuals and populations. ROH segments would be expected within an individual when both identical haplotypes share a common ancestor, and this should be correlated to the inbreeding coefficient as defined by the probability that two genes at a locus are identical by descent (Kim *et al.*, 2013; Mastrangelo *et al.*, 2018). One of the most important factors that affect the ROH patterns in various genomic regions is artificial selection of superior animals. Such selection pressure increases mainly the proportion of homozygous genotypes around the target locus involved in the genetic control of phenotypic traits of interest. Thus, ROH segments can be also defined as genomic regions with reduced diversity and, consequently, high homozygosity around the selected locus that might harbour targets of positive selection and are under strong selective pressure (Pemberton *et al.*, 2012). Because of this, the analysis of ROH segment distribution in the genome provides an information about how the architecture of genome can disclose a population's genetic background. By revealing the molecular changes in populations over time, genome-wide information is crucial to understanding antecedent genome architecture and, therefore, to maintaining diversity and fitness in endangered livestock breeds (Peripolli *et al.*, 2017).

The aim of this study was to analyse the ROH segment distribution in the genomes of Slovak Spotted and Slovak Pinzgau cattle, to determine the impact of artificial selection on the architecture of their genomes and to describe the genomic regions mostly affected by strong selection pressure during the development of those breeds.

## MATERIAL AND METHODS

**Database of genotyping data**

In order to analyse the impact of artificial selection on genome architecture the genotyping data for a total of 236 animals, representing the nucleus of both Slovak Spotted and Slovak Pinzgau cattle, were obtained. The sample from Slovak Spotted cattle consisted of 37 AI sires and 48 dams that were genotyped by using two platforms, Illumina BovineSNP50v2 BeadChip (AI sires) and ICBF International Dairy and Beef v3 (dams). The samples of Slovak Pinzgau cattle covered living animals (19 active breeding bulls, 35 dams of sires, and 79 dams of dams) as well as AI doses deposited in reproduction centres (18 animals). In case of Slovak Pinzgau cattle, all of animals were genotyped by using Illumina BovineSNP50v2 BeadChip in a commercial lab.

**Quality control of data**

The data cleaning was performed by using PLINK 1.9 (Purcell *et al.*, 2007; Chang *et al.*, 2015). The quality control of genotyping data was carried out to remove markers assigned to unmapped regions or with unknown chromosomal position according to the latest bovine genome assembly (Btau 4.0) and SNPs positioned to sex chromosomes. In the following step, the consensus map had to be constructed, because of the two different genotyping platforms used for animals' genotyping. The final consensus map file consisted of 40,033 markers. In the subsequent SNP pruning only samples with lower than 10 % of missing genotypes, autosomal SNPs with call rate higher than 90 % and minor allele frequency higher than 1 % that adhered to mendelian inheritance patterns were retained.

**Distribution of ROH segments in the genome**

The ROH segments were defined according to Ferenčaković *et al.* (2013) as genomic regions with 15 or more consecutive homozygous calls with maximum gap between consecutive SNPs of 1 Mb and minimum density of one SNPs per every 100 kb. Because of the theoretical relationship between the distribution of identity by descent (IBD) fragments and the number of generation since common ancestor the minimum length of ROH segments was set to 1 Mb. The distributions of ROH segments in the genome were analysed separately for five length categories (> 1 Mb, > 2 Mb, > 4 Mb, > 8 Mb, and > 16 Mb). Heterozygous calls were not allowed across ROH categories, except length > 16 Mb with one permissible call. Missing calls per windows were not allowed for lengths > 1 Mb and > 2 Mb, while one missing call was accepted for length > 4 Mb, two for > 8 Mb and four for > 16 Mb. The total number of ROH detected, the average length of ROH (in Mb) and the sum of all ROH segments per animal were calculated for each ROH length category within and across analysed breeds. The proportion of autosomes covered by ROH was then expressed for length > 1 Mb as the pools of overlapping segments within animals per each breed.

The subsequent analysis of genome-wide selection signatures was based on the assumption that the identified autozygosity islands across the genome of Slovak Spotted and Slovak Pinzgau cattle are results of selective breeding for traits of interest defined in their breeding objectives. The autozygosity islands, characterized by SNPs with extreme frequency in ROH segments > 4 Mb across specific genomic regions, were determined based on the calculation of runs incidence per each SNP by using Plink 1.9 (Purcell *et al.*, 2007; Chang *et al.*, 2015). The genome-wide occurrence of SNPs in ROH was then expressed as the frequency (%) of overlapping ROH shared among samples and visualised by R package qqman (Turner, 2014). The genome-wide significance threshold for SNPs under selection (with extreme ROH frequency) was determined based on the corresponding boxplot distribution. All of SNPs with appropriate level of significance were assigned to the genomic QTL (quantitative trait loci) location according to the Bovine Genome Database (http://bovinegenome.org). To identify genes located in ROH regions under the most intense selection pressure, the Genome data viewer of the bovine genome UMD3.1.1 was used (https://www.ncbi.nlm.nih.gov/genome/gdv/).

## RESULTS AND DISCUSSION

After quality control of genotyping data, the database consisted of 39,261 common SNPs for both Slovak Spotted and Slovak Pinzgau cattle that

covered overall length 2,497,077 kb of the genome. The average distance between adjacent SNPs was 63.67 kb; minimum distance was 0.02 kb and maximum distance between markers was 4428.95 kb.

The ROH segments were identified across the genome of all analysed individuals, with average number of 54.59 ± 18.58 segments and average length of 130.33 ± 58.40 Mb. The detailed descriptive statistics of the ROH number and length by categories for each breed is given in Table 1. The average sums of the ROH length calculated per animal and averaged per breed were higher in Slovak Spotted (152.48 Mb) compared to Slovak Pinzgau cattle (117.86 Mb) and represent in average 6.11 % or 4.72 % of their autosomal genome covered by SNP markers. As expected from these results, the average number of ROH per animals was similarly higher in Slovak Spotted (71.86) compared to Slovak Pinzgau cattle (44.88). The obtained proportion of the genome autozygosity per animals was comparable to results reported for dual-purpose breeds (Ferenčaković *et al.*, 2011; Marras *et al.*, 2015; Szmatoła *et al.*, 2016). On the other hand, both of the analysed breeds showed lower genome autozygosity compared to those reported for dairy cattle. For example Purfield *et al.* (2012), Kim *et al.* (2013) and Mastrangelo *et al.* (2016) identified dairy cattle as one of the most homozygous animals among various cattle breeds. Such high genome autozygosity in dairy cattle could be a consequence of intensive artificial selection as well as repeated use of superior and proven sires in breeding practices (Peripolli *et al.*, 2018). Figure 1 shows the chromosome-wise distribution of ROH segments with minimum length 1 MB across breeds. As can be evident from other results as well (Table 2, Figure 3), the major fraction of autosome residing in ROH was observed on BTA6 for both analysed breeds (13.49 % resp. 14.26 % of autosomal length in ROH), while the lowest coverage by ROH was found on autosome BTA19 for Slovak Spotted and on BTA24 for Slovak Pinzgau cattle.

Figure 2 shows the differences between the number of ROH segments (> 4 Mb, > 8 MB, and > 16 Mb) and the length of the genome covered by them. The results indicated that the number of ROH segments as well as the length of genome covered by them were considerably different

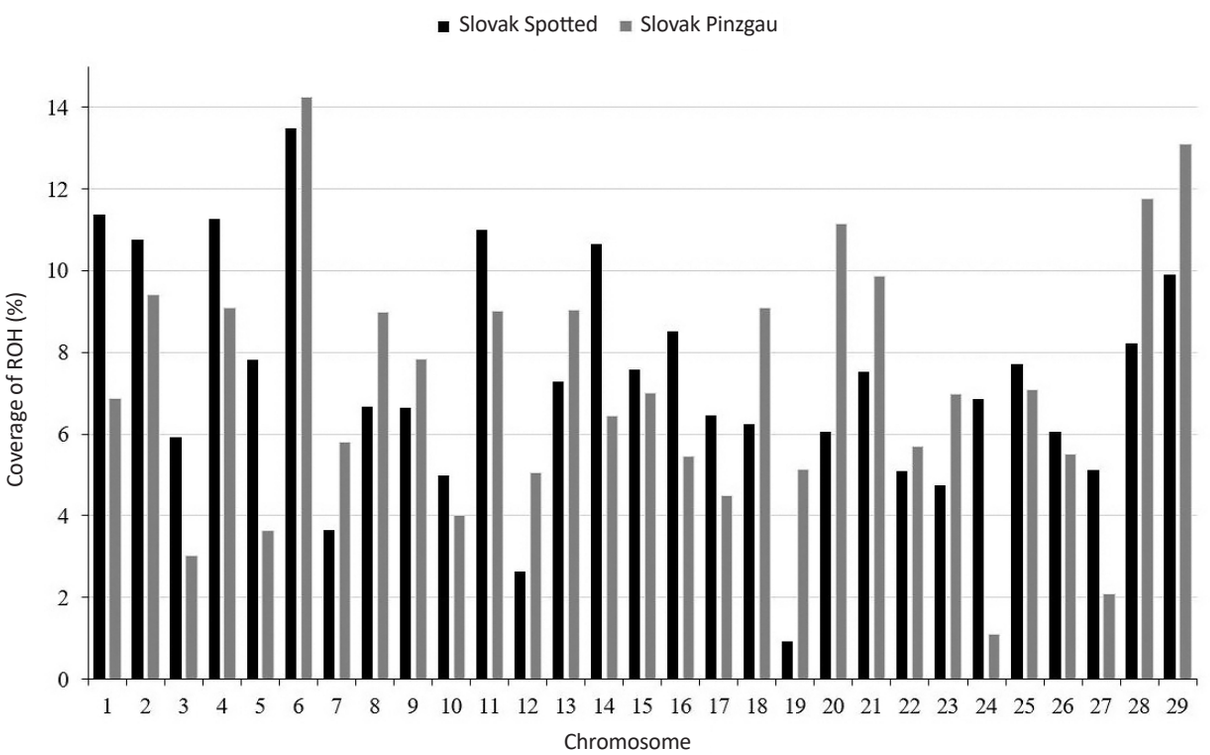


**Figure 1. Percentage of autosome coverage by ROH (minimum ROH length set to 1Mb)**

depending on the studied animal, which is likely a result of the distinct distances from the common ancestors (Mészáros *et al.*, 2015).

Based on the applied criteria in ROH analysis across all length categories, overall 8,694 ROH segments for Slovak Spotted and 10,270 for Slovak Pinzgau cattle were identified. As described in many studies, the length and frequency of ROH can give insight into the history of the population in which an individual occurs and the history of that individual's ancestors (Howrigan *et al.*, 2011; Curik *et al.*, 2014; Szmatoła *et al.*, 2016). Thus, the length of ROH can be used to determine the age of inbreeding (Curik *et al.* 2014). It has been shown that very long autozygous ROH are expected to originate from recent common ancestors, while most of short ROH are likely derived from more remote ancestors. But sometimes, short ROH might persist in a population for a very long time, much above defined base population, as a consequence of the lack of recombination or just by chance (Curik *et al.*, 2017). Howrigan *et al.* (2011) revealed based on simulated sequence data that the expected length of autozygous segments follow an exponential distribution with average value equal to ½ g Morgan (g is the number of generations since common ancestor). In cattle, Ferenčaković *et al.* (2013) showed that the ROH > 1 Mb date back ~50 generations, > 2 Mb ~25 generations, > 4 Mb ~12.5 generations, > 8 Mb ~6 generations, and > 16 Mb ~3 generations. In our study, the total length of runs of homozygosity for both breeds was composed mostly from short segments (> 1 Mb) that accounted for 70.26 % (Slovak Spotted) and 65.99 % (Slovak Pinzgau) of all segments detected. The ROH segments of 2 – 4 Mb long, representing the 25 – 12 .5 generations from common ancestor, accounted for 27.4 % (Slovak Spotted) and 28.98 % (Slovak Pinzgau). The lowest proportion within the total length of ROH was found for the longest segments (> 16 Mb) that accounted for 0.45 % and 1.29 % of all segments detected. In terms of the genetic diversity those results indicated that on average 0.45 % of the Slovak Spotted genome and 0.88 % of the Slovak Pinzgau genome could be affected by recent inbreeding.

In subsequent analysis the impact of artificial selection on genome architecture was tested based

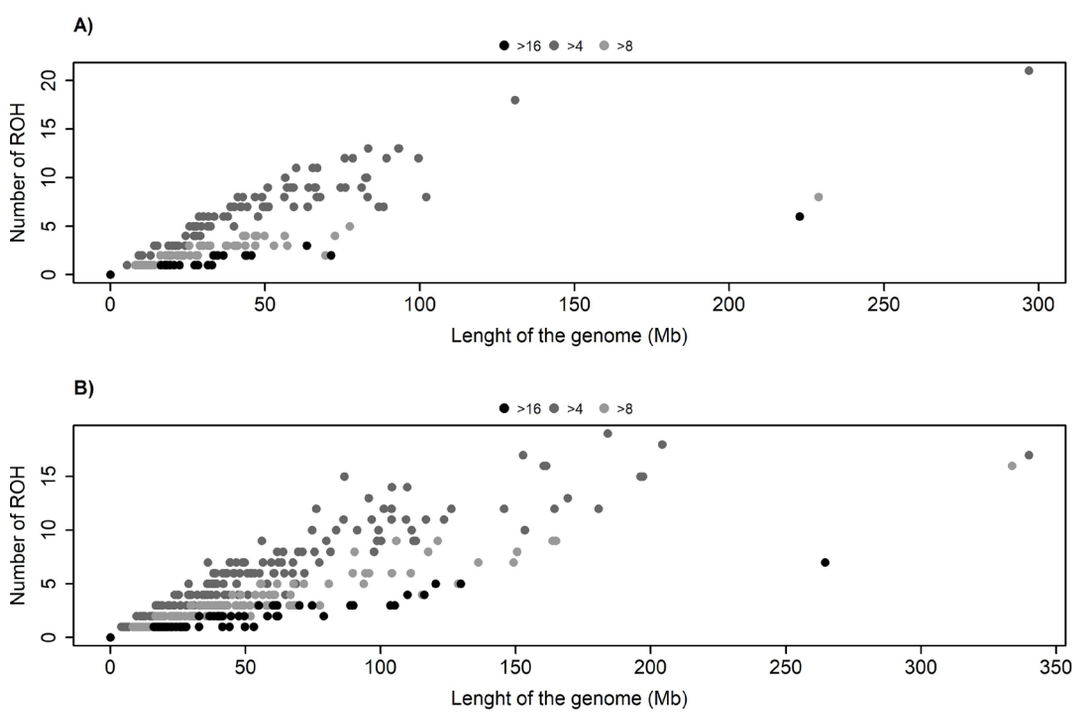


**Figure 2. The relationship between the number of ROH segments and the length of genome covered by them (minimum ROH length set to 4 Mb, 8 Mb, and 16 Mb) for Slovak Spotted (A) and Slovak Pinzgau (B) cattle**

**Table 1. The length (Mb) and number (in brackets) of ROH by categories within analysed breeds**

| Category | Mean ± SD | Lower 95 % CI | Upper 95 % CI | Range | Genome coverage % |
|---|---|---|---|---|---|
| Slovak Spotted | | | | | |
| > 1 Mb | 152.48 ± 53.18 (71.86 ± 16.42) | 141.01 (68.32) | 163.95 (75.40) | 21.38 - 396.13 (14.00 - 101.00) | 6.11 |
| > 2 Mb | 83.83 ± 46.66 (21.28 ± 8.64) | 73.76 (19.42) | 93.89 (23.15) | 3.11 - 317.54 (1.00 - 36.00) | 3.36 |
| > 4 Mb | 50.73 ± 37.90 (6.99 ± 3.65) | 42.56 (6.20) | 58.91 (7.78) | 0.00 - 296.79 (0.00 - 21.00) | 2.03 |
| > 8 Mb | 23.10 ± 29.57 (1.69 ± 1.49) | 16.73 (1.37) | 29.48 (2.02) | 0.00 - 228.91 (0.00 - 8.00) | 0.93 |
| > 16 Mb | 11.22 ± 28.04 (0.46 ± 0.92) | 5.17 (0.26) | 17.27 (0.66) | 0.00 - 222.70 (0.00 - 6.00) | 0.45 |
| Slovak Pinzgau | | | | | |
| > 1 Mb | 117.86 ± 57.67 (44.88 ± 11.23) | 108.58 (43.07) | 127.13 (46.69) | 18.83 - 398.71 (13.00 - 70.00) | 4.72 |
| > 2 Mb | 76.58 ± 53.98 (13.44 ± 6.52) | 67.89 (12.39) | 85.26 (14.49) | 2.16 - 350.75 (1.00 - 34.00) | 3.07 |
| > 4 Mb | 58.96 ± 51.28 (6.26 ± 4.21) | 50.71 (5.59) | 67.20 (6.94) | 0.00 - 339.89 (0.00 - 19.00) | 2.36 |
| > 8 Mb | 38.50 ± 43.76 (2.54 ± 2.39) | 31.47 (2.16) | 45.54 (2.93) | 0.00 - 333.68 (0.00 - 16.00) | 1.54 |
| > 16 Mb | 21.95 ± 35.06 (0.88 ± 1.22) | 16.31 (0.69) | 27.58 (1.08) | 0.00 - 264.49 (0.00 - 7.00) | 0.88 |

on the assumption that the regions with increased ROH frequencies are most likely consequences of selective breeding for traits of interest during the development of analysed breeds (Kim *et al*., 2013; Curik *et al*. 2014). The consecutive ROH segments identified in the genome close together reflected mainly the existence of a founder alleles broken down by recombination over generation (Biscarini *et al*., 2014). In our study, the autozygosity islands were defined as specific genomic regions where SNPs had extreme ROH frequency (minimum ROH length set to 4 Mb). According to the boxplot distribution the genome-wide significance threshold for those SNPs was set to 6.5 % for Slovak Spotted, 5.9 % for Slovak Pinzgau and 5.4 % for a whole population (Figure 3). As Figure 3A and 3B show, the overlapping ROH were evident across the genomes of both breeds, but the distribution of identified autozygosity islands was not uniform and differentiated between breeds. The detailed description of each region with extreme ROH frequencies, corresponding number of SNPs, number of genes and quantitative trait loci (QTL) is listed in Tables 3 and 4.

A total of 18 overlapping autozygosity islands located on seven autosomes (BTA4, BTA6, BTA11, BTA16, BTA18, BTA28, and BTA29) were detected across both breeds (Table 2). As shown in figure 3C, a strong selection signal was found mainly on BTA6. Several genes responsible for various traits were identified directly in the regions of these homozygous segments. Within the second autozygosity island on BTA2, the genes encoding insulin-like growth factor binding protein (IGFBP1 and IGFBP3) were found. IGFBP as a structural gene responsible for the multiple influences of insulin-like growth factors (IGFs) system is considered as a candidate gene for growth and production traits (Othman *et al*., 2014). The KIT gene was identified on the BTA6 in the fifth region. In cattle,

the KIT gene is responsible for coat colour pattern and is recognized as a candidate gene for the spotting locus (Fontanesi *et al*., 2016). In addition, the MC1R gene (melanocortin receptor 1) that determines the basic coat color in cattle (Dorshorst *et al*., 2015) was found in the homozygous region on BTA18. In the sixth region on BTA6 the genes for casein family were detected (CSN1S1, CSN1S2, CSN2, CSN3). For example the CSN2 A1 genetic variant is considered as a riskfactor in milk intolerance and in other important human diseases (Massella *et al.*, 2017). On BTA11 the POMC gene (proopiomelanocortin), which is a candidate for carcass traits in beef cattle, was identified. POMC is the precursor for several peptide hormones produced by post-translational processing, some of which are involved in energy homeostasis, including α-melanocyte stimulating hormone (MSH), corticotropic hormone (ACTH) and β-endorphin. In cattle, this gene plays an important role in ingestive behaviour, energy homeostasis and hot carcass and shipping weights (Garza-Brenner *et al.*, 2017).

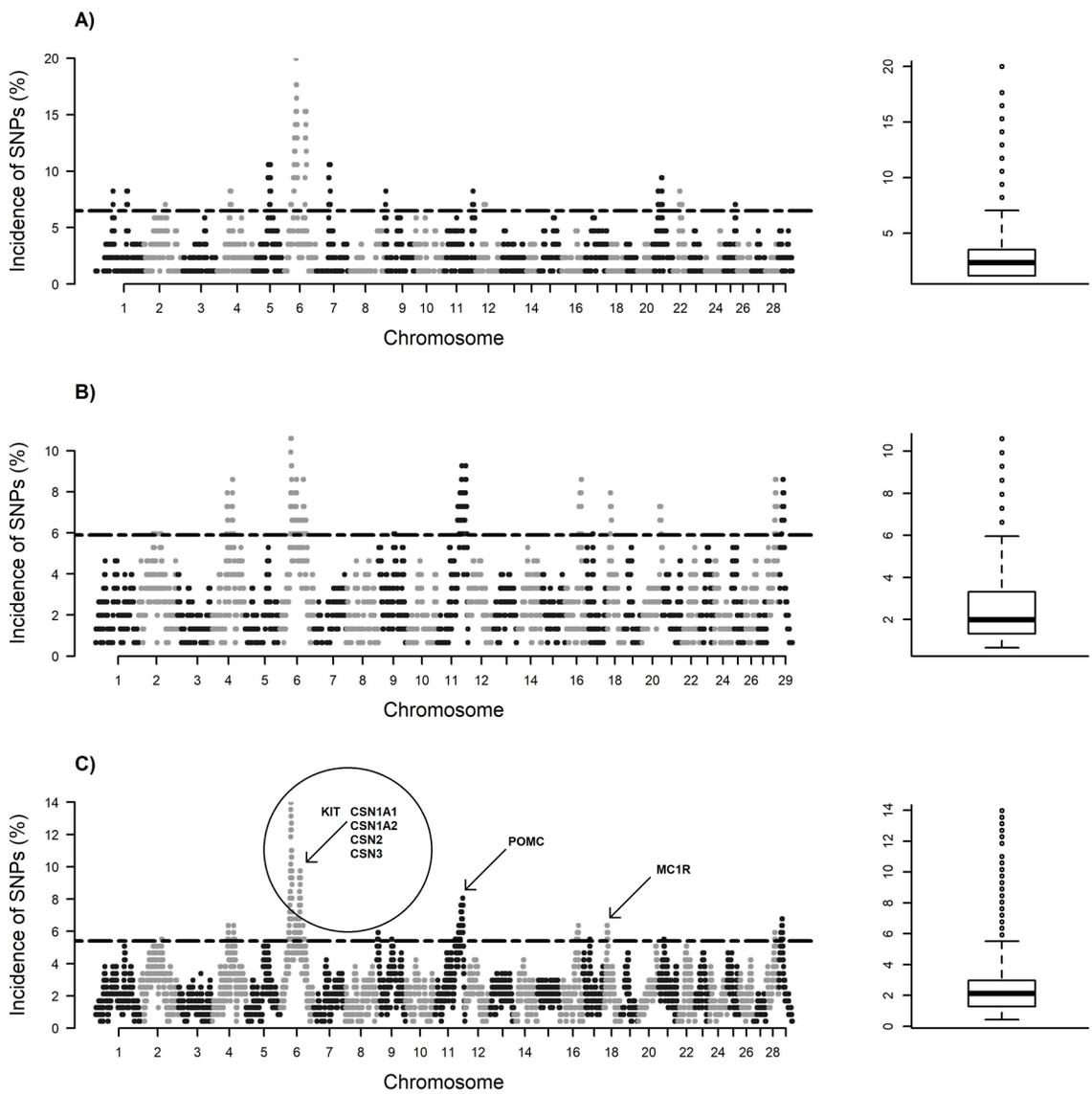


**Figure 3. Genome-wide occurrence of SNPs in ROH for Slovak Spotted (A) and Slovak Pinzgau (B) cattle and incidence of runs for each SNP across both populations (minimum ROH length set to 4 Mb)**

**Table 2. Overlapping autozygosity islands across the genome of analysed breeds**

| BTA | Start position (Mb) | End position (Mb) | Length (Mb) | No. of SNPs | No. of genes | QTL traits |
|---|---|---|---|---|---|---|
| 4 | 54.55 | 58.31 | 3.76 | 56.00 | 22 | Somatic cell score |
| | 76.07 | 76.90 | 0.82 | 17.00 | 5 | |
| | 78.39 | 80.71 | 2.31 | 35.00 | 12 | |
| 6 | 51.59 | 51.99 | 0.40 | 12.00 | 1 | Birth weight, Yearling weight, Stature, Strength, Marbling score, Milk yield, Protein and Fat yield, Protein and Fat percentage, Backfat EBV |
| | 54.14 | 59.31 | 5.17 | 87.00 | 23 | |
| | 63.28 | 63.89 | 0.62 | 6.00 | 2 | Stature, Strength, Body, Rump width, Suspensory, ligament, Teat placement, Foot angle, Quality of udder, Quality of feet and legs, Udder depth, Protein and Fat percentage |
| | 67.32 | 73.31 | 5.99 | 88.00 | 67 | |
| | 82.05 | 87.40 | 5.35 | 39.00 | 61 | |
| 11 | 72.56 | 73.98 | 1.42 | 22.00 | 38 | Yield grade |
| | 75.50 | 76.35 | 0.86 | 14.00 | 6 | |
| | 79.66 | 81.28 | 1.63 | 24.00 | 9 | |
| | 85.74 | 89.02 | 3.28 | 52.00 | 37 | Fat yield |
| | 90.39 | 91.88 | 1.49 | 20.00 | 5 | |
| | 92.79 | 101.22 | 8.43 | 124.00 | 217 | |
| 16 | 57.89 | 66.11 | 8.22 | 134.00 | 79 | Fat depth, Yield grade, Hot carcass weight |
| 18 | 11.84 | 15.10 | 3.26 | 44.00 | 72 | Hot carcass weight, Dystocia (maternal effect) |
| 28 | 30.84 | 33.10 | 2.26 | 22.00 | 18 | Protein and Fat percentage |
| 29 | 7.48 | 14.12 | 6.64 | 108.00 | 47 | Milk Speed, Temperament, Marbling score, Milk yield, Birth weight |

In the genome of Slovak Spotted cattle, overall 19 genomic regions distributed across 12 autosomes (BTA1, BTA2, BTA4, BTA5, BTA6, BTA7, BTA9, BTA11, BTA22, and BTA25) were found to be under intense selection pressure. The longest ROH was found on BTA6 (30,812,012:42,866,973 bp), while the shortest on BTA2 (71,209,253:71,657, 308 bp). Inside the ROH island on BTA6 various QTLs were reported, including those that affect milk yield (Viitala *et al.*, 2003), protein and fat yield (Kühn *et al.*, 1999), birth weight (Casas *et al.*, 2000), stature and strength (Hiendleder *et al.*, 2003), longissimus muscle area (Casas *et al.*, 2003), and daily gain (Lee *et al.*, 2011). For the Slovak Pinzgau cattle a total of 21 genomic regions with extreme ROH frequencies on BTA2, BTA4, BTA6, BTA9, BTA11, BTA16, BTA18, BTA20, BTA28, and BTA 29 were detected. The strongest ROH pattern was identified on BTA4 in the region from 65,400,608 bp to 80,706,119 bp that included QTLs affecting teat length (Ashwell *et al.*, 2001), longissimus muscle area and marbling score (Mizoshita *et al.*, 2004). The subset of biologically and economically most important quantitative trait loci located in each region is summarized in Table 3 and 4. Besides them, we identified in target regions of selection several genes involved in multiple signalling and signal transduction pathways in a wide variety of biological processes, including the genetic control of milk production and reproduction (LCT, CSN1S1, CSN1S2, CSN2, CSN3, BMPR1B), body conformation and meat quality (GHRHR, POMC, MYO1G), coat colour (MCR1, KIT) and immunity response (IGFBP, IGJ, MR1, TLR10, TLR6).

Generally, the breeding objectives of dual-purpose breeds are mostly similar, but environmental and management condition may vary, giving rise to slightly different selection pressures applied to given traits which can lead to different selection pressures to loci across the genome (Howard *et al.*, 2015). The comparison of ROH patterns from observed breeds could also provide an insight into the effect of selection on the genome over varying periods of time and determine if the direction of selection was similar (Kim *et al.*, 2013). The results of our study indicated that the regions

**Table 3. Genomic region with extreme ROH frequencies in Slovak Spotted cattle**

| BTA | Start position (Mb) | End position (Mb) | Length (Mb) | No. of SNPs | No. of genes | QTL traits |
|---|---|---|---|---|---|---|
| 1 | 57.17 | 57.91 | 0.74 | 10 | 12 | Fat yield, Protein yield, Protein percentage |
| | 102.57 | 105.75 | 3.19 | 40 | 9 | Protein percentage, Resistance to BSE |
| 2 | 71.21 | 71.66 | 0.45 | 11 | 7 | Fat thickness, Birth weight, Marbling score, Slaughter weight, Functional herd life |
| 4 | 44.51 | 48.82 | 4.31 | 82 | 45 | Somatic cell score |
| 5 | 60.85 | 69.96 | 9.10 | 111 | 90 | Dressing percentage, Yearling weight, Birth weight, Longissimus muscle area, Protein yield, Twinning rate, Concentration of follicule stimulating hormone |
| 6 | 21.59 | 23.78 | 2.19 | 37 | 27 | Longissimus muscle area, Milk yield, Protein and Fat percentage, Protein and Fat yield, Pre-weaning average daily gain |
| | 30.81 | 42.87 | 12.05 | 193 | 53 | Birth weight, Milk yield, Protein and Fat yield, Protein and Fat percentage, Stature, Strength, Longissimus muscle area, Daily gain |
| | 68.29 | 72.97 | 4.68 | 64 | 57 | Stature, Strength, Body, Rump width, Suspensory ligament, Teat placement, Foot angle, Quality of udder, Quality of feet and legs, Udder depth, Protein percentage |
| 7 | 41.03 | 46.35 | 5.32 | 84 | 226 | |
| 9 | 4.13 | 5.68 | 1.55 | 30 | 1 | |
| 11 | 92.69 | 95.64 | 2.95 | 37 | 85 | |
| 12 | 33.78 | 39.02 | 5.24 | 57 | 56 | Milk yield, Protein and Fat yield, Protein percentage |
| 21 | 20.89 | 23.76 | 2.86 | 63 | 3 | Somatic cell score, Birth weight |
| | 30.01 | 34.61 | 4.60 | 64 | 85 | Birth weight |
| 22 | 26.26 | 29.10 | 2.85 | 36 | 10 | |
| | 30.99 | 31.48 | 0.49 | 10 | 2 | |
| | 33.19 | 33.77 | 0.57 | 9 | 2 | Somatic cell score |
| | 34.88 | 35.63 | 0.75 | 10 | 5 | |
| 25 | 36.79 | 37.99 | 1.20 | 18 | 55 | |

displaying autozygosity in Slovak Spotted and Slovak Pinzgau cattle are linked mostly to milk production and muscle development, thus selection for dual-purpose performance.

## CONCLUSION

In this study we provided the insight into the distribution of autozygosity islands across the genomes of two dual-purpose breeds with historical and national importance in Slovakia; Slovak Spotted and Slovak Pinzgau cattle. We found that the number of ROH segments as well as the length of genomes covered by them are considerably different depending on the studied animal and breed. Despite the fact that the overall ROH length indicated higher total level of autozygosity in the genome of Slovak Spotted cattle, the proportion of ROH segments > 16 Mb indicated higher risk of genetic diversity loss for Slovak Pinzgau cattle due to the recent inbreeding. For both breeds the regions of genome that were most commonly associated with ROH and therefore could reflect the impact of artificial selection were identified and described. Generally, our results

**Table 4. Genomic region with extreme ROH frequencies in Slovak Pinzgau cattle**

| BTA | Start position (Mb) | End position (Mb) | Length of region (Mb) | No. of SNPs | No. of genes | QTL traits |
|---|---|---|---|---|---|---|
| 2 | 49.81 | 51.90 | 2.09 | 32 | 6 | Marbling score, Milk yield |
| | 53.43 | 55.48 | 2.04 | 47 | 7 | Marbling score, Milk yield |
| | 60.09 | 63.78 | 3.70 | 52 | 20 | Marbling score, Milk yield, Functional herd life |
| 4 | 54.55 | 59.33 | 4.78 | 79 | 24 | Somatic cell score |
| | 60.92 | 61.73 | 0.81 | 20 | 9 | |
| | 65.40 | 80.71 | 15.31 | 235 | 163 | Teat length, LMA, Marbling score |
| 6 | 37.10 | 42.99 | 5.88 | 110 | 25 | Birth weight, Milk yield, Protein and Fat yield, Protein and Fat percentage, Stature, Strength, Daily gain |
| | 49.21 | 59.31 | 10.11 | 178 | 30 | Birth weight, Yearling weight, Stature, Strength, Marbling score, Milk yield, Protein and Fat yield, Protein and Fat percentage |
| | 65.34 | 69.88 | 4.54 | 77 | 53 | Stature, Strength, Body, Rump width, Suspensory ligament, teat placement, Foot angle, Quality of udder, Quality of feet and legs, Udder depth, Milk yield, Protein and Fat yield, Protein and Fat percentage, Pre-weaning average daily percentage |
| | 71.91 | 73.31 | 1.40 | 30 | 19 | |
| | 79.84 | 88.59 | 8.75 | 115 | 83 | |
| | 90.56 | 93.94 | 3.37 | 66 | 57 | |
| 9 | 53.77 | 57.95 | 4.18 | 85 | 15 | Milk yield, Protein and Fat yield |
| 11 | 71.34 | 77.56 | 6.22 | 84 | 108 | Yield grade |
| | 79.17 | 91.88 | 12.70 | 176 | 78 | Fat yield, Pelvic and Heart fat |
| | 93.29 | 103.62 | 10.33 | 160 | 249 | |
| 16 | 56.49 | 66.22 | 9.73 | 160 | 105 | Fat depth, yield grade, Hot carcass weight |
| 18 | 10.83 | 16.03 | 5.20 | 69 | 109 | Dystocia (maternal effect), Hot carcass weight |
| 20 | 62.05 | 68.20 | 6.15 | 123 | 36 | Meat tenderness, Milk yield, Protein percentage |
| 28 | 27.46 | 37.20 | 9.75 | 149 | 110 | Protein and Fat percentage |
| 29 | 6.69 | 15.49 | 8.80 | 140 | 54 | Milk speed, temperament, Milk yield, Birth weight |

confirmed that the regions displaying autozygosity in Slovak Spotted and Slovak Pinzgau cattle are linked mostly to milk production and muscle development, thus selection for dual-purpose performance. However, due to the common history of analysed breeds, some selection signatures were found in the same genomic regions located on seven autosomes directly or very close to the genes involved in the genetic control of milk production, carcass traits and coat colour. In addition, our study confirmed that the genome-wide scan for ROH segments in cattle can be used as an alternative strategy to identify the genomic regions as well as genes related to traits with biological and economical importance.

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