

## Assessment of genetic diversity using characteristics based on the probability of gene origin in the Slovak Spotted breed

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The aim of this study was to assess the diversity characteristics based on the probability of gene origin in the Slovak spotted breed. The animals were divided into 3 groups by breed types: group  $S_0$  where the ration of other than Slovak Spotted breed was <12.5 %; group  $S_1$  =12.5–25 %; group  $S_2$  = 25–50 %. The reference population of these groups consisted of 17355; 6251 and 12683 individuals, respectively. The pedigree information was available from the Breeding Services of the Slovak Republic, s. e. The total number of founders ( $f$ ) ranged from 10484 in the  $S_1$  to 20402 in the  $S_2$ , the effective number of founders ( $f_e$ ) ranged from 265 in the  $S_0$  to 507 in the  $S_2$ , the effective number of ancestors ( $f_a$ ) ranged from 69 in the  $S_0$  to 112 in the  $S_2$  and founder's genome equivalent ( $f_g$ ) ranged from 41.71 in the  $S_0$  to 70.75 in the  $S_2$ . The most important ancestor EGE 003 originating from Slovakia explained 5.30 % of the genetic diversity in purebred Slovak Spotted population. The results derived from the probabilities of gene origin indicate that the genetic diversity within each group was still relatively large.

**Keywords:** genetic diversity, founder, Slovak Spotted cattle

### 1. Introduction

Genetic improvement of cattle involves a struggle to achieve the proper balance between selection intensity of a small number of parents in the current generation and maintenance of sufficient genetic diversity for future generations. Slovak Spotted breed is dual-purpose cattle and belongs to the group of Simmental breeds. Genetic diversity was assessed using parameters based on the probability of gene origin, because these parameters describe population genetic diversity precisely. Contrariwise, the inbreeding coefficient and effective population size are important in monitoring the diversity over longer time periods (Boichard et al., 1997). An assessment of genetic diversity based on pedigree information was published in papers by Kadlečík et al. (2011, 2012), Mc Parland et al. (2007), Baumung and Sölkner (2002), Sölkner et al. (1998). Danchin-Burge et al. (2012) described evolution of the genetic variability of eight French dairy cattle breeds assessed by pedigree analysis. They found out that all analysed breeds could be considered quite poor from a genetic diversity point of view.

The aim of the study was to assess genetic diversity of three subpopulations existing within Slovak Spotted breed using characteristics based on the probability of gene origin.

### 2. Material and methods

Assessment of the genetic diversity (GD) in Slovak Spotted breed was based on pedigree information. The data was obtained from the Central Register of Farm Animals of the Breeding Services of Slovak Republic, s. e. The animals

born from 1987 to 2009 and registered in Herd book set up the analyzed reference population. The animals of reference population were divided into three groups as follows: group  $S_0$ : (purebred animals where the proportion of other than Slovak Spotted breeds was <12.5 %, this group represents the breed); group  $S_1$ : (12.5–25 %); group  $S_2$ : (25–50 %). The group  $S_0$  consisted of 17355 animals (129 males and 17234 females);  $S_1$  of 6251 (121 males and 6243 females) and  $S_2$  of 12683 (8 males and 12683 females) individuals.

The SAS 9.2 software was used for data processing.

Parameters of genetic variability were estimated using ENDOG v4.8 software (Gutiérrez and Goyache, 2005).

#### 2.1 Pedigree completeness

Quality of pedigree information was evaluated with pedigree completeness index as proposed by MacCluer et al., (1983):

$$PCI = \frac{2C_{sire}C_{dam}}{C_{sire} + C_{dam}}$$

where:

$C_{sire}$  and  $C_{dam}$  are the indexes for paternal and maternal contribution, respectively and

$$C = \frac{1}{d} \sum_{i=1}^d a_i$$

where:

$a_i$  is the proportion of ancestors known in generation  $i$  and  $d$  is the present number of generations traced back in pedigree

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## 2.2 Parameters based on the probability of gene origin

- Number of founders ( $f$ ) – in the whole analysis, it was assumed that all founders are animals with unknown parents and that they are non-inbred.
- Effective number of founders ( $f_e$ ) which is defined as the number of equally contributing founders that would be expected to produce the same genetic diversity. It was estimated as;  $f_e = \frac{1}{\sum_{k=1}^f q_k^2}$ , where  $q_k$  is the probability of gene origin of the  $k$  ancestors.

- Effective number of ancestors ( $f_a$ ) is the minimum number of ancestors explaining the complete genetic diversity of a population. It was computed as;  $f_a = \frac{1}{\sum_{j=1}^a q_j^k}$ ,

is the marginal contribution of an ancestor and  $j$  which is the genetic contribution made by an ancestor that is not explained by other ancestors chosen before. The  $f_a$  account for recent bottleneck and thus partially account for the loss of allelic diversity in descendant population (Boichard et al., 1997).

- The  $f_d/f_e$  ratio reflects the role of bottleneck in the development of the population (Melka, et al., 2010). The  $f_g/f_e$  ratio measures the influence of genetic drift. Lower values of the ratio are connected with higher loss of genetic diversity due to genetic drift. Founder genome equivalents ( $f_g$ ) is the number of founders that would be expected to give the same level of genetic diversity in the population under study if the founders were equally represented and no loss of alleles occurred (Lacy, 1989). It was computed as:

$$f_g = \frac{1}{\sum_{j=1}^{N_f} \left( \frac{p_j^2}{r_j} \right)}$$

contribution of the founder  $j$  and  $r_j$  is retention of alleles. The  $f_g$  accounts for unequal contributions of founders, bottleneck, and random loss of alleles due to genetic drift (Lacy, 1995).

## 3. Results and discussion

Results of pedigree completeness index are shown in Figure 1. The percentage of known ancestors in the first generation

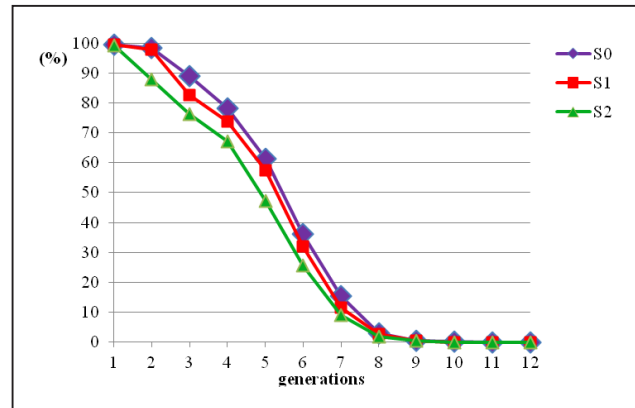


Figure 1 Pedigree completeness in subpopulations Slovak Spotted cattle

was more than 99 % in each group.  $S_0$  showed a greater proportion of known ancestors in the fifth generation (61.42 %) compared to  $S_1$  (57.41 %) and  $S_2$  (47.23 %). Pedigrees of the first 5 generations are more or less complete for all assessment of groups (above 75 %). The proportion of known ancestors decreased to less than 50 % after 5 generations in all groups. Analysed pedigrees were more complete than those published by Maignel et al. (1996) in Simmental and some dairy breeds as well as by Kadlečik and Pavlík (2012) in four beef cattle breeds. However, several authors warned about the sensitivity of pedigree quality to parameters of genetic variability. Boichard et al. (1997) estimated that 10% of incomplete data was enough to strongly underestimate inbreeding. According to the same article, the advantage of the theory of the probability of gene origin when assessing the genetic variability of a breed is its robustness in comparison with the methods based on the probability of identity computation.

Parameters based on the probability of gene origin detected recent significant changes in breeding strategies. The results of the parameters based on the probability of gene origin are shown in Table 1. We found out that the number of founders ( $f$ ) ranged from 10484 in the  $S_1$  to 20402 in the  $S_2$ . The size of the founder population was larger when the pedigree completeness level was lower, as

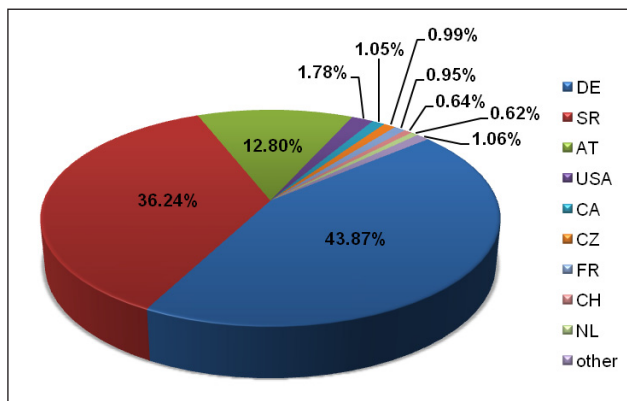
Table 1 Characteristics based on the probability of gene origin in subpopulations of Slovak Spotted cattle

Variable	$S_0$	$S_1$	$S_2$
N	17355	6252	12683
$f$	20260	10484	20402
$f_e$	265	332	507
$f_a$	69	78	112
$f_g$	41.71	49.54	70.75
Ancestors to explain 50% of the genetic diversity	27	38	77
$f_d/f_e$	0.26	0.23	0.22
$f_g/f_e$	0.15	0.14	0.13

**Table 2** The most important ancestors in purebred Slovak Spotted cattle

Ancestor	Sex	Name	Year of birth	Country of origin	Marginal contributions		Offspring
					CC	IC	
EGOS-ET	♂	EGE 003	1998	SK	5.308628	5.308628	4023
SAMURAI	♂	STG 001	1992	DE	9.622047	4.313419	2334
HAXL	♂	DE000979317838	1966	DE	13.57274	3.950692	27
HORROR	♂	DE000809706945	1979	DE	17.40309	3.830352	31
RALBO	♂	DE000911825633	1988	DE	20.86895	3.465861	20

CC – cumulated contributions; IC – individual contributions; SR – Slovakia; DE – Germany



**Figure 2** The country of origin of ancestors explaining 100 % of genetic diversity in the Slovak Spotted cattle

in the  $S_2$  group. The decrease of genetic variability assessed via these parameters is reflected in lower values, such as the effective number of founders (ranged from 265 in the  $S_0$  to 507 in the  $S_2$ ) and effective number of ancestors (ranged from 69 in the  $S_0$  to 112 in the  $S_2$ ). The founder's genome equivalent accounts for the loss of genetic diversity due to unequal founders' contribution and random genetic drift. It is a more accurate description of variation. All determinations of genetic loss are relative to the genetic diversity in the founder population. The analysis of allelic loss is therefore a determination of the probability that alleles present in the founder population still reside within the descendant population (Lacy, 1989). We found out the founder genome equivalent ranged from 41.71 in the  $S_0$  to 70.75 in the  $S_2$ . The comparison between the  $f_e$  and  $f$  demonstrates declining genetic diversity as a consequence of unequal contributions of founders. Similar results were observed in the Charolais and Limousine cattle in Europe (Bouquet et al., 2011). Higher values of  $f_e$  than  $f_a$  indicate the occurrence of bottleneck and genetic drift of population in all the groups. Genetic drift was likely connected with the higher age of the most important ancestors. Our results were more significant than in the Simmental cattle in Ireland (McParland et al., 2007) and Simmental cattle in France ( $f = 7623$ ;  $f_e = 117$ ;  $f_a = 39$ ) (Danchin-Burge et al., 2012). However, Sölkner et al. (1998), found higher values of  $f_a = 113.7$  resp.  $f_g = 94.3$  in Austrian Simmental. Based on the  $f_a/f_e$  ratios, the loss of

genetic diversity attributable to bottleneck in all the groups is similar. The loss of genetic diversity due to a bottleneck is larger in Slovak Spotted cattle than in the French Simmental (Danchin-Burge et al., 2012), Slovak Pinzgau (Kadlečík et al., 2011) and Slovak Holstein (Pavlík et al., 2012) populations  $f_a/f_e = 0.33$ ; 0.36; 0.37 respectively. The  $f_g/f_e$  ratio measures the impact of genetic drift excluding the effect of founder contributions on genetic diversity, so that lower ratios are associated with higher impact of genetic drift. The loss of genetic diversity attributable to genetic drift in all the groups is similar. The effect of random genetic drift was higher in Brown Swiss (0.07) and Canadienne (0.08) populations (Melka et al., 2010) compared to the Slovak Spotted breed. Only a small number of ancestors was needed to explain half of the genetic diversity in the studied subpopulations. Thus it is likely that these groups will produce half sibs that will increase the average value of inbreeding in subsequent generations of the offspring (Pjontek et al., 2012).

The most important five ancestors in purebred Slovak Spotted cattle are shown in Table 2. All of them were male ancestors. Only one ancestor EGOS – ET (EGE 003) originates from Slovakia. The individual contribution of this bull explained 5.30 % of the genetic diversity in purebred Slovak Spotted population. The biggest proportion of individual contributions was detected in the ancestors originating from Germany (44 %). The ancestors originating from Slovakia explained 36 % of the genetic diversity in Slovak Spotted cattle.

#### 4. Conclusions

We have analyzed the genetic diversity using parameters based on the probability of gene origin of Slovak Spotted breed and its subdivided populations. Pedigrees of the first 5 generations are more or less complete in all assessed groups (above 75 %). The comparison between the  $f_e$  and  $f$  demonstrates a decline in the genetic diversity as a consequence of the unequal contributions of founders. This could happen due to the excessive use of some animals as parents of subsequent generations. Based on the  $f_g/f_e$  and  $f_a/f_e$  ratios, the loss of genetic diversity was attributable to bottleneck and genetic drift in all the groups. The results derived from probabilities of gene origin indicate that the

genetic diversity within each group was still relatively large. The most important ancestor EGOS – ET (EGE 003) was originally from Slovakia. The EGOS – ET (EGE 003) explained 5.30 % of the genetic diversity in purebred Slovak Spotted population.

## 5. Acknowledgements

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