

THE ASSESSMENT OF GENETIC DIVERSITY AND ANALYSIS OF PEDIGREE COMPLETENESS IN PUREBRED SLOVAK SPOTTED CATTLE

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ABSTRACT

The aim of this study was to assess the diversity based on pedigree information in purebred Slovak Spotted cattle. The pedigree information was available from The Breeding Services of the Slovak Republic, s. e. The pedigree file consisted of 109,686 individuals (105,229 dams and 4,457 sires). The 17,355 individuals (17,234 dams and 121 sires) born from 1987 to 2009 and registered in Herd book set up the analyzed reference population (RP). The 66.76% of animals in the RP were inbred. The maximum number of generations traced was 8.34 ± 1.35 , number of fully traced generations 2.34 ± 0.65 and equivalent complete generations 4.79 ± 0.71 . The average inbreeding coefficient for reference population was $F_i = 0.57\%$ and the average individual increase of inbreeding was $\Delta F_i = 0.14\%$. The average individual relatedness coefficient was $AR = 0.9\%$. The effective number of founders was 265, the effective number of ancestors was 69 and 27 individuals explained 50% of genetic diversity. Results will be used in genetic management of breeding work in Slovak Spotted population and monitoring of parameters characterizing genetic diversity and their development, as well.

Key words: cattle / breeds / Slovak Spotted cattle / average relatedness / genetic diversity / inbreeding / pedigree completeness

1 INTRODUCTION

Slovak Spotted is a dual-purpose breed with a good milk and meat production (Bujko, 2011), which belongs to the Simmental type of cattle. Maintaining genetic diversity in a population is important, because its loss limits mating choices and has adverse effects on economically (Golden *et al.*, 2000) and biologically relevant traits (Márquez *et al.*, 2010). The reduction of genetic diversity is associated with various adverse incidents such as inbreeding depression in fitness-related traits and an increased fluctuation in selection response (Falconer and Mackay, 1996). Pedigree analysis is an important tool to describe genetic variability and its evolution across generations (Gutiérrez *et al.*, 2003). The level and rate of inbreeding in the population have to be monitored at time (Vahlsten, 2004). A suitable method of assessment of genetic diversity within population based on the parameters derived from the probabilities of gene origin, such

as effective number of founders and effective number of ancestors. These parameters precisely describe genetic diversity of populations after a small number of generations, while inbreeding coefficient is important in monitoring the diversity over longer time periods (Boichard *et al.*, 1997).

The aim of this study was to assess levels and trends in inbreeding and to determine the depth of pedigree known, the average relatedness, the effective number of founders and ancestors in purebred Slovak Spotted cattle.

2 MATERIALS AND METHODS

Evaluation of the genetic diversity of the cattle population was made on the basis of pedigree information from the database of the Breeding Services of Slovak Republic. Basic characteristic of the assessed files are given in Table 1. As purebred were used individuals that have

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Table 1: Basic characteristic of the assessment files

Populations	N	Cows	Bulls	Inbred animals
pedigree file	109,686	105,229	4457	15.95%
active population	36,949	36,820	129	42.77%
reference population	17,355	17,234	121	66.76%

more than 87.5% of the genes originating from the Slovak spotted breed. The pedigree file (PP) consisted of 109,686 individuals and the 36,949 individuals registered in Herd book set up the analyzed active population (AP). The reference population (RP) set up 17,355 purebred individuals (17,234 dams and 121 sires) born from 1987 to 2009. The percentage of inbred animals in RP was above 66%, while in PP was only 15.9%. For data processing software SAS V9.2 was used. For monitoring of genetic variability in populations was used ENDOG v4.8 software (Gutiérrez and Goyache, 2005).

Several different types of measures were used to describe quality of pedigrees and the genetic variability of Slovak Spotted breed. The pedigree completeness level of the population was assessment by using 3 parameters:

1. the maximum number of generation traced (*GenMax*),
2. the number of fully traced generation (*GenCom*) and
3. the equivalent complete generations (*GenEqu*),

which is computed as the sum over all known ancestors of the terms computed as the sum of $(1/2)^n$ where n is the number of generations separating the individual to each known ancestor (Gutiérrez and Goyache, 2005).

As it was proposed by MacCluer *et al.* (1983), the pedigree completeness index (PCI) of Slovak Spotted was calculated using the formula: $PCI = 4C_{sire} C_{dam} / (C_{sire} + C_{dam})$. Where C_{sire} and C_{dam} are contributions from the paternal and maternal lines respectively and, $C = 1/g \sum_{i=1}^g a_i$ where a_i is the proportion of known ancestors in generation i , and g is the number of generations considered in this study ($g = 5$).

2.1 MEASURES BASED ON THE PROBABILITY OF IDENTITY BY DESCENT

The *inbreeding coefficient* (F_i) of an individual is the probability of having two genes which are identical by descent (Wright, 1923). The individual increases in inbreeding (ΔF_i) are computed as: $\Delta F_i = 1 - \sqrt[1-F_i]{1-F_i}$, where F_i is the individual coefficient of inbreeding and t is the equivalent complete generations (Gutiérrez and Goyache, 2005).

The *average relatedness coefficient* (AR) of each individual is defined as the probability that an allele randomly chosen from the whole population in the pedigree belongs to a given animal. AR can then be interpreted as the representation of the animal in the whole pedigree regardless of the knowledge of its own pedigree (Gutiérrez and Goyache, 2005). This parameter provides complementary information to that provided by the inbreeding coefficient (Gutiérrez *et al.*, 2003).

2.2 MEASURES BASED ON PROBABILITY OF GENE ORIGIN

2.2.1 EFFECTIVE NUMBER OF FOUNDERS

All animals with both parents unknown are regarded as founders in this study. *Effective number of founders* (f_e), is defined as the number of equally contributing founders that would be expected to produce the same genetic diversity. This is computed as: $f_e = \frac{1}{\sum_{k=1}^r q_k^2}$, where q_k is

the probability of gene origin of the k ancestors (Gutiérrez and Goyache, 2005). When founders contribute unequally, the effective number of founders is smaller than the actual number (Sørensen *et al.*, 2005).

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

where q_j is marginal contribution of an ancestor j , which is the genetic contribution made by an ancestor that is not explained by other ancestors chosen before (Gutiérrez and Goyache, 2005). The effective number of ancestors (f_a) is less than the f_e and the comparison of both numbers can be used to find the bottlenecks that have occurred from the founders to the present population (Boichard *et al.*, 1997).

3 RESULTS AND DISCUSSION

The pedigree completeness in the reference and active population of Slovak Spotted was above 80%, but it was only 45% in whole pedigree file. Results of the maximum number of generation traced the number of fully generations traced, the equivalent complete generations, and the percentage of known ancestors of the first, third and fifth generation of assessed populations are in Table 2. The percentage of known ancestors in the first generation was above 99%, while in the fifth generation was only 61.3% for reference and 55.7% for active population.

Table 2: Pedigree completeness characteristics in populations of Slovak Spotted

Variable		Reference population	Active population	Pedigree file
the maximum number of generation traced	\bar{x}	8.34	8.2	4.58
	s	1.35	1.36	3.57
	x_{\min}	0	0	0
	x_{\max}	12	12	12
the number of fully traced generation	\bar{x}	2.34	2.1	1.05
	s	0.65	0.64	1.01
	x_{\min}	0	0	0
	x_{\max}	5	5	5
the equivalent complete generations	\bar{x}	4.79	4.53	2.42
	s	0.71	0.79	1.94
	x_{\min}	0	0	0
	x_{\max}	6.57	6.57	6.57
percentage of known ancestors in:				
1 st generation back		99.37	99.33	69.47
3 rd generation		88.94	83.48	42.92
5 th generation		61.34	55.71	24.09

Mc Parland *et al.* (2007) found in the purebred Herefords born in 2004, that 84% had complete information on 4 generations, however only 3.5% of Simmentals had this degree of pedigree information. Melka *et al.* (2010) found greater proportion (31%) of known ancestors in the seventh generation for Canadian, compared to Guernsey and Milking Shorthorn, which might be due to the use of

some relatively old parents in the breeding program. We found GenMax, for reference, active and whole populations 8.34, 8.2 and 4.5, respectively. The GenMax for Slovak Spotted is lower than results obtained by Baumung and Sölkner (2002) from population Tux-Zillertal (6) and Original Pinzgau (13). The maximum numbers of generations traced don't gives reliable information about

Table 3: Characteristics based on the probability of identity by descent of assessed populations and inbred populations of Slovak Spotted

Variable		Reference population		Active population		Pedigree file	
		whole population n = 17355	inbred animals n = 11587	whole population n = 36949	inbred animals n = 15804	whole population n = 109686	inbred animals n = 17503
the average inbreeding coefficient (%)	\bar{x}	0.57	0.86	0.36	0.85	0.14	0.88
	s	1.7	2.03	1.48	2.17	0.95	2.25
	x_{\min}	0	0.001	0	0.001	0	0.001
	x_{\max}	26.17	26.17	26.17	26.17	37.5	37.5
the average relatedness coefficient (%)	\bar{x}	0.9	0.97	0.8	0.95	0.36	0.92
	s	0.38	0.35	0.38	0.35	0.44	0.37
	x_{\min}	0.000009	0.026	0.000009	0.02	0.000009	0.003
	x_{\max}	2.25	2.25	2.25	2.25	2.45	2.25
the average individual increases in inbreeding (%)	\bar{x}	0.14	0.21	0.094	0.21	0.038	0.24
	s	0.46	0.56	0.41	0.61	0.3	0.73
	x_{\min}	0	0	0	0	0	0
	x_{\max}	9.93	9.93	9.93	9.93	25	25

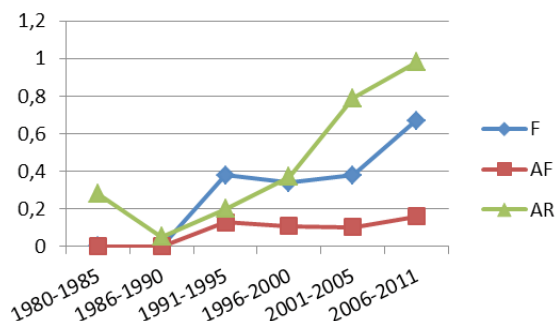


Figure 1: Inbreeding trend, mean average relatedness and average increase of inbreeding of purebred Slovak Spotted cattle

gaps in the pedigree. A good way of describing the quality of a pedigree is the average complete generation equivalent (Baumung and Sölkner, 2003). The GenEqu was comparable between reference and active populations. The average number of generation equivalents quantifies how many generations have been traced (Hammami *et al.*, 2007).

Table 3 shows the results of assessment based on the probability of identity by descent of assessed populations and inbred populations. The increase in population homozygosity can be measured through the inbreeding coefficient, while the loss of diversity is related to the average coancestry coefficient. The values of coefficient of inbreeding, increase in inbreeding and average relatedness for animals in reference population were greater than those found in the active population and pedigree file, i.e. 0.57%, 0.14% and 0.9% respectively. The values of coefficient of inbreeding, increase in inbreeding and average relatedness for inbred animals were similar for each of assessed populations. Our results were lower than results presented by Mc Parland *et al.* (2007). They estimated in the Holstein-Friesian population the average value of inbreeding coefficient 1.49% in 2004, increasing by 0.10% per annum. The level of inbreeding in the Holstein-Friesian in 2004 was lower than of the purebred Holstein (2.5%) and Friesian (1.61%) populations. As well as within the Simmental population they observed that inbreeding level has also been rising, with an annual increase of 0.06% between the years 1994 and 2004, reaching an inbreeding level of 1.35% in 2004. The AR

coefficient addresses the chance of recovery of the breed, since it also takes coancestry coefficients into account, not only for the animals of the same generation but also for those of previous generations whose genetic potential could be interesting to preserve (Gutiérrez *et al.*, 2003).

The inbreeding trend, average relatedness and average increase of inbreeding are shown Fig. 1. The inbreeding trend for reference population has ascending tendency, similarly to their respective trend in average coancestry. We could assume that in the next generation the number of inbred individual will increase.

Results of the analysis of gene origin probabilities are shown in Table 4. We found the number of founders (f) for reference population almost less by half (20,260) as in the active population (42,082). The number of founders in the pedigree influences inbreeding coefficient, and a greater number of founders in the pedigree leads to reduced inbreeding, as these are assumed to be non-inbred and unrelated to each other for computing purposes (Márquez *et al.*, 2010). The effective number of founders (f_e) was higher for the active population (350) than the reference population (265). The comparison between the f and f_e demonstrates a decline in genetic diversity because of unequal contributions of founders. This could happen due to excessive use of some animals as parents of subsequent generations (Melka *et al.*, 2010). Estimates of f_e are dependent on the size of the population and the number of actual founders, so smaller populations are expected to have a smaller f_e . A small f_e would suggest that genetic drift has been occurring since the founder generation (Sørensen *et al.*, 2005). The effective number of ancestors (f_a) was smaller than that of the effective number of founders (f_e) for all groups. The differences between f_e and f_a reflect the existence of bottleneck in the pedigree (Gutiérrez *et al.*, 2003). Our results were higher than results presented by Mc Parland *et al.* (2007). They estimated the number of founders 1,248, the effective number of founders 55 and effective number of ancestors 35 for Simmental breed. The numbers of ancestors explaining 50% of diversity was 44 for active population and only 27 for reference population. The f_a account for recent bottleneck, and thus accounts partly for the loss of allelic diversity since the founder population. However, it does not completely reflect the genetic diversity and the

Table 4: Characteristic based on the probability of gene origin in populations of Slovak Spotted

Variable	Reference population	Active population	Pedigree file
f	20,260	42,082	42,432
f_e	265	350	610
f_a	69	86	172
ancestors to explain 50% of genetic diversity	27	44	133

value also depends on the depth of pedigree (Sørensen *et al.*, 2005).

4 CONCLUSIONS

The level of inbreeding gain per generation and relatedness were under 1%. The average value of relatedness coefficient in reference as well as in the whole pedigree file was higher than inbreeding coefficient. Therefore we could assume that number of inbred individuals will increase in the next generation. The value of effective number of founders and ancestors for animal in reference population were lower as those found in the pedigree file. The comparison between the f and f_e advert to a decrease of genetic diversity as consequence of unequal contributions of founders. This could happen due to excessive use of some animals as parents of subsequent generations. In order to prevent loss of diversity within breed it is needed to monitor and use suitable mating strategy.

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