

# Evaluation of Genetic Diversity Using Parameters Based on Probability of Gene Origin in the Slovak Spotted Bulls

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## Abstract

The aim of this study was to assess the diversity based on probability of gene origin in Slovak Spotted bulls. The pedigree information was available from The Breeding Services of the Slovak Republic, s. e. The pedigree file consisted of 752 individuals. The 62 sires born from 1995 to 2009 and registered in Herd book set up the analyzed reference (RP) population. Total number of founders in the RP was 308, effective number of founders was 115 and the effective number of ancestors was 37. The number of ancestors explaining 50 % of the diversity was 15 and founder's genome equivalent was 20.46. The sire GS Malf and Horwein were with 16 offspring's the most frequently used bulls in the artificial insemination. We found that the genetic conservation index for RP was 16.34 %. Results will be used in genetic management of breeding work in Slovak Spotted and monitoring of parameters characterizing genetic diversity and their development, as well.

**Keywords:** diversity, effective number of ancestors, effective number of founders, founders genome equivalent, Slovak Spotted cattle

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## 1. Introduction

Slovak spotted is dual-purpose cattle and belongs to the type of Simmental cattle. The genetic variability in a population is influenced by the number of founders, selection intensity, inbreeding and genetic drift [1]. The use of only a few sires could lead to a decreasing of the genetic diversity of breed. The rate of inbreeding is a common means to monitor changes of genetic diversity through its transformation to the effective size of population. However, this method is very sensitive to incomplete pedigree information. Another complementary approach is to analyze the probabilities of gene origin as effective number of founders, ancestors and genome equivalent. Evaluation of genetic diversity using parameters based on probability of gene origin, such as effective number of founders and ancestors from cattle to treated [2- 4]. The aim

of this study was to evaluate the genetic diversity based on probability of gene origin in Slovak Spotted bulls.

## 2. Materials and methods

Evaluation of the genetic diversity of the cattle population was based on analysis of pedigree information. The database was created from Central register of the Breeding Services of Slovak Republic. The pedigree file (PP) consisted of 752 individuals (435 dams and 317 sires). The 62 sires born from 1995 to 2009 and registered in Herd book set up the analyzed reference population (RP). For data processing software SAS V9.2 was used. The population parameters were calculated using the algorithms of the freely available ENDOG v4.8 software [5].

### *Effective number of founders ( $f_e$ )*

Each individual with unknown parents was considered as a founder. The expected genetic

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contribution of each founder to the reference population was defined as the probability of a gene taken at random within the reference population to come from a given founder. The effective number of founders is the number of equally contributing founders that would be expected to produce the same genetic diversity as in the population under study [6]. This is computed as:  $f_e = \frac{1}{\sum_{k=1}^f g_k^2}$  where  $q_k$  is the

probability of gene origin of the  $k$  ancestor [7]. The effective number of founders reflects the unequal contributions of founders to the current population due to selection rates and variation of family size [8]. When founders contribute unequally, the effective number of founders is smaller than the actual number [9].

*Effective number of ancestors ( $f_a$ )*

The effective number of ancestors represents the minimum number of equally contributing ancestors that are necessary to explain the complete genetic diversity in a population. The effective number of ancestors  $f_a$  is less than the effective numbers of founders  $f_e$  and the comparison of both numbers can be used to find the bottlenecks that have occurred from the founders to the present population [10] the greater the  $f_e/f_a$  ratio, the more stringent the bottleneck [11]. This parameter complements the information offered by the effective number of founders accounting for the losses of genetic variability produced by the unbalanced use of reproductive individuals producing bottleneck [5]. This is computed as:  $f_a = \frac{1}{\sum_{j=1}^a q_j^2}$  where  $q_j$  is the

marginal contribution of an ancestor  $j$ , which is the genetic contribution made by an ancestor that is not explained by other ancestors chosen before.

*Effective number of founder genomes ( $f_g$ )*

The effective number of founder genomes is defined as the number of equally contributing founders with no loss of founder alleles that would give the same amount of genetic diversity as is the reference population. The effective number of founder genomes  $f_g$  accounts for the loss of genetic diversity that occurred in population due to genetic drift and bottlenecks [8].

*Genetic Conservation Index GCI*

The ENDOG can compute of genetic conservation index for each of the individuals of the analyzed population. The index is computed from the genetic contributions of all identified founders as:

$$GCI = \frac{1}{\sum p_i^2}$$

where  $p_i$  is the proportions of genes of founder  $i$  in the pedigree of an animal. The index is based on the assumption that the objective of a conservation program is to retain the full range of alleles possessed by the base population [5].

**3. Results and discussion**

The loss or gains of genetic diversity in the whole and reference populations are shown in Table 1. This table gives the contributions of each subpopulation to within, between and total gene diversity. We found the loss of genetic diversity in the whole population (-1.9897) by contrast in the reference population, where we found the low (0.06295) gain of genetic diversity.

**Table 1.** The loss or gain of genetic diversity in the populations

	GD <sup>1</sup>	I D <sup>2</sup>	M D <sup>3</sup>	Loss/Gain
whole population	0.9755	-1.8834	-0.1062	-1.98970
reference population	0.9959	0.16923	-0.1062	0.06295

<sup>1</sup>GD=gene diversity remaining after removal of the corresponding subpopulation, <sup>2</sup>I\_D=the contributions of each subpopulation to within-subpopulations, <sup>3</sup>M\_D=contributions of each subpopulation to between subpopulations

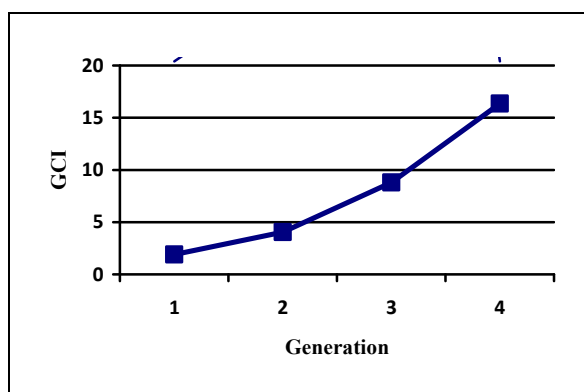
Results of the parameters based on the probability of gene origin are shown in Table 2. The number of founders was equal (308) for reference population and whole pedigree file. The values of effective number of founders and ancestors for animal in reference population were lower as those found in the pedigree file, i.e. 115 and 37 respectively. These results are comparable to numbers for the Slovak Pinzgau bulls, where they found  $f_e$  99 and  $f_a$  23[12]. The comparison between the number of founder  $f$  and the effective number of founders  $f_e$  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. The difference between the effective number of founders and the effective number of ancestors show that bottlenecks have occurred since the foundation of the population. The number of ancestors explaining 50% of diversity was 15 for reference population. Similar results were observed in Canadian Holstein population, where 11 ancestors explained 50 % of gene pool [13] and in Slovak Pinzgau bulls, where 10 ancestors explained 50 % of diversity [12]. The genetic contribution index was computed from the genetic contributions of all identified founders. We found GCI 16.34 % for reference and 4.18 % for whole pedigree file. The ideal individual would receive equal contributions from all the founder ancestors in the population and consequently, the higher the values of animal for conservation [5]. The increases in values of GCI in the individual's generations are shown in Figure 1. The values of GCI were almost over by half in the each next generation.

**Table 2.** Characteristics based on the probability of gene origin in population Slovak Spotted

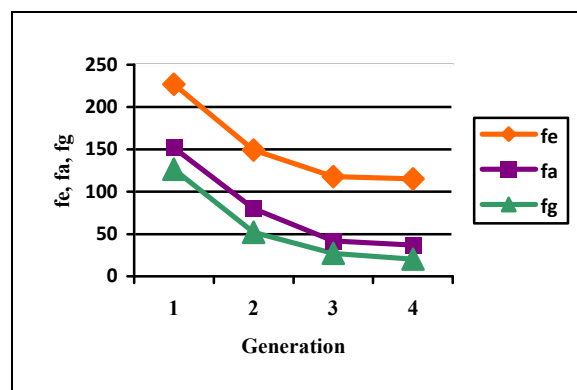
Variable	Pedigree file	Reference population
$N^1$	752	62
$f^2$	308	308
$f_e^3$	150	115
$f_a^4$	85	37
$f_g^5$	-	20.46
GCI <sup>6</sup>	4.18%	16.34%
Ancestors to explain 50% GD <sup>7</sup>	39	15

<sup>1</sup> $N$ =number of animal, <sup>2</sup> $f$ =number of founders, <sup>3</sup> $f_e$ =effective number of founders, <sup>4</sup> $f_a$ =effective number of ancestors, <sup>5</sup> $f_g$ =effective number of founder genomes, <sup>6</sup>GCI=genetic conservation index, <sup>7</sup>GD=genetic diversity



**Figure 1.** The increases in values of GCI in the generations

The results of effective numbers of founders, ancestors and effective number of founder's genomes in generations are shown in Figure 2. We found the highest effective number of founders (227), effective number of ancestors (152) and effective number of founder genomes in the oldest i.e. 1<sup>st</sup> generation (126.34). The effective number of founders, ancestors and founder's genomes were almost less by half in the each next generation, save for the last generation. In the case of minimum inbreeding, the effective number of founder's genomes is smaller than the effective number of founders and ancestors and smaller than half the effective population size. The degree to which the effective number of founder genomes is smaller is an indication of the degree of random loss of alleles. As alleles are lost every generation, the effective number of founder genomes decreases every generation and is therefore sensitive to depth of pedigree [9], we found the low values of average number of fully traced generation (3.05) and maximum number of generations traced (6.05) both in reference population.



**Figure 2.** Effective number of founders, ancestors and founder genomes in the generations (4-reference population)

#### 4. Conclusions

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. The degree to which the effective number of founder genomes is

smaller is an indication of the degree of random loss of alleles. As alleles are lost every generation, the effective number of founder genomes decreases every generation and is therefore sensitive to depth of pedigree, whereby we found the low values of average number of fully traced generations and maximum number of generations traced. In order to prevent loss of diversity within breed it is needed to monitor and use suitable mating strategy.

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