

## Inbreeding and genetic diversity loss of four cattle beef breeds in Slovakia

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The aim of the paper was to evaluate trends in inbreeding and loss of genetic diversity in four beef cattle breeds (Blonde d'Aquitaine-BA, Charolais-CH, Limousine-LI, Simmental-SM). The highest ratio of inbred animals was found in the SM breed (63.6%) and the lowest in the LI (14.1%). The highest average inbreeding intensity was found in the SM, the lowest in the BA. The amount of genetic diversity in the reference population accounting for diversity loss due to genetic drift and unequal founder contributions was the highest in the SM (6.2%), following the BA (3.5%), LI (1.1%) and CH (0.9%). The proportion of genetic diversity lost due to genetic drift was higher in BA, CH, LI than the loss of genetic diversity due to unequal founders contributions.

**Keywords:** beef cattle, pedigree analysis, inbreeding, genetic diversity

### Introduction

Knowing the level of genetic diversity is the basis for effective breeding work with a population. Genetic diversity has economic and environmental benefits, allowing for undertaking genetic improvement of animals for economically important traits and facilitating adaptation to changing production systems (Melka et al., 2013). Trakovická et al. (2015) pointed out that several genes were determined as population's genetic indices with potential significance of their impact on long-life production traits in endangered Pinzgau cattle. Similar results are important because in several studies were demonstrated (Kasarda and Kadlečík, 2007; McParland et al., 2007; Kadlečík et al., 2012; Tang et al., 2013; Pavlík et al., 2014) improvement of inbreeding intensity that negatively influenced genetic diversity status and economics of some traits. Investigation of genetic diversity requires also to estimate trends and major causes of diversity loss. The issue of animal genetic diversity loss has become crucial and needs an immediate attention to conserve the available animal genetic resources (Melka et al., 2013). Šidlová et al. (2015) studied genomic variability among cattle populations based on runs of homozygosity. Several authors have reported genetic diversity loss (Melka et al., 2013 in Guernsey population, Krupa et al., 2015 in 5 breeds of pigs). Genetic diversity loss can be derived from parameters based on probability of gene origin like effective number of founders, or founder genome equivalent ( $f_{ge}$ ), effective number of

founder genomes, or founder genome equivalent ( $f_{ge}$ ) and effective number of non-founders, or non-founder equivalent ( $f_{ne}$ ) as described Lacy (1995), Cabalero and Toro (2000). Mészáros et al. (2015) pointed out genomic analysis importance for effective management in small and endangered populations.

The aim of the paper was to evaluate trends in inbreeding and loss of genetic diversity in four beef cattle breeds.

### 2 Material and methods

As we already described in previous publication (Kadlečík and Pavlík, 2012), tradition of beef cattle breeding in Slovakia is short. Four new purebred beef breeds (Blonde d'Aquitaine-BA, Charolais-CH, Limousine-LI, Simmental-SM) were imported to Slovakia after 1990. Their further development was organized in the frame of established The Beef Cattle Breeders Association. In this paper were analysed reference populations consisted of (pedigree populations in brackets) 109 (481) Blonde d'Aquitaine (BA) animals, 1,762 (3,955) Charolais (CH), 773 (2,063) Limousine (LI) and 428 (916) Simmental (SM). Reference populations (RP) set up living heifers and cows, registered in Herd Books in the year 2011. The reference population analysis covered living sires in insemination, frozen genetic material deposited in reproduction centres and natural mating, as well.

Inbreeding trends and genetic diversity loss were estimated from genealogic information of animals using the data obtained from The Beef Cattle Breeders

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Association in Slovakia. Pedigree completeness, the parameters of diversity based on probability of identity by descent and based on probability of gene origin were estimated by the software Endog v.4.8 (Gutiérrez and Goyache, 2005).

### 2.1 Pedigree completeness parameters

The pedigree completeness has influence on estimated genetic diversity parameters. Three different measures were used to evaluate pedigree completeness and depth:

- maximum generations traced was estimated as the number of generations between an animal and its earliest ancestor;
- mean complete equivalent generations was computed as the sum over all known ancestors of the terms computed as the sum for  $(1/2)^n$ , where  $n$  is the number of generations separating the individual to each known ancestor (Maignel et al., 1996);
- pedigree completeness index (PCI) described by MacCluer et al. (1983):

$$PCI = 2 C_{sire} C_{dam} / C_{sire} + C_{dam}$$

where:

$C_{sire}$  and  $C_{dam}$  – contributions from the paternal and maternal lines

and

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

where:

- $g_i$  – the proportion of known ancestors in generation  $i$
- $d$  – the number of generations that are taken into account

### 2.1 Genetic diversity and genetic diversity loss

Genetic diversity was evaluated according to three parameters based on the probability of the identity by descent and seven measures based on probability of gene origin.

Inbreeding coefficient of an animal ( $F$ ) was calculated according to algorithm of Meuwissen and Luo (1992).

The average relatedness ( $AR$ ) reflects the probability that an allele randomly chosen from the whole population in pedigree belongs to a given animal (Gutiérrez et al., 2009).

The individual increase in inbreeding ( $\Delta F_i$ ) was calculated by means of the classical formula where  $F_i$  is individual coefficient of inbreeding and  $t$  is the complete equivalent generation (Gutiérrez et al., 2009). The trends of inbreeding, coancestry and inbreeding gain were estimated as moving averages on birth years of animals.

Number of founders ( $f$ ) was defined as ancestors with unknown parents, or animals with unknown genetic connections to other animals in pedigrees except its own progenies (Lacy, 1989). In case that all  $f$  would contribute equally the number of founders would be the same as the  $f_e$ .

The effective number of founders ( $f_e$ ) was estimated as the number of equally contributing founders that will produce the same genetic diversity as assessed in the population (Boichard et al., 1997), calculated as:

$$f_e = \left[ \sum_{k=1}^f q_k^2 \right]^{-1}$$

where:

$q_k$  – the probability of gene origin of the  $k$  ancestor

The effective number of ancestors ( $f_a$ ) is the minimal number of ancestors necessary to explain the genetic diversity in the reference population (Boichard et al., 1997), was calculated by formula:

$$f_a = \left[ \sum_{j=1}^a q_j^k \right]^{-1}$$

where:

$q_j$  – the marginal contribution of an ancestor  $j$  which is the genetic contribution made by 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)

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). The  $f_g$  was calculated by the Caballero and Toro (2000) algorithm, as follows:

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

where:

$N_f$  – the number of founders

$p_j$  – the contribution of the founder

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

The ratio  $f_a / f_e$  characterize the role of bottleneck in the population development. 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.

The loss of genetic diversity (GD) was derived from parameters  $f_a$ ,  $f_g$ . Total GD of the reference population was estimated by formula of Lacy (1995):

$$GD = 1 - \frac{1}{2f_g}$$

The genetic diversity loss due to bottleneck and genetic drift in the population was calculated as  $1 - GD$ . The amount of genetic diversity in the reference population considered for the loss of diversity due to unequal founder contributions ( $GD^*$ ) was calculated as (Lacy, 1995):

$$GD^* = 1 - \frac{1}{2f_e}$$

Similarly,  $1 - GD^*$  represented the loss of genetic diversity due to unequal founder contributions. The difference  $GD^* - GD$  measures the loss of diversity by genetic drift accumulated over nonfounder generations and was calculated by Caballero and Toro (2000).

### 3 Results and discussion

#### 3.1 Pedigree completeness

The BA and SM breeds had the most complete pedigrees with  $PCI = 100\%$  in parental generation followed by CH (99.7%) and LI (93.4%). With respect to the generations of ancestors the SM showed the highest quality of pedigrees (Figure 1) with  $PCI = 72.2\%$  in the fourth and 40.4% in the fifth generation. The percentage of known ancestors in the fifth generation of the other three breeds moved from 22.6 to 28.9%. Average values of complete equivalent generations as well as maximum generation traced (Table 1) were comparable among evaluated breeds. Estimated pedigree quality of all four breeds showed that the parameters of genetic diversity should be compared with some caution.

#### 3.2 Genetic diversity and genetic diversity loss

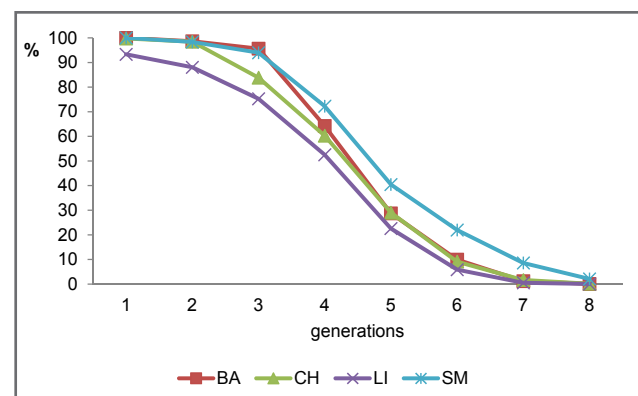
Maintaining of genetic diversity is an important part of the goals in population genetic management. In spite of strong breeder's effort to organize outbreeding as a basic way of animal mating in many breeds a part of populations is inbred. Gutiérrez et al. (2003) found in eight Spanish beef breeds 3.7–48.3% of inbred animals by breeds. In our study the ratio of inbred animals differs by breeds, as well. The highest ratio of inbred animals was found in

the SM breed (63.6%) and the lowest in the LI (14.1%). The highest average inbreeding intensity we found in the SM, the lowest in the BA. Figure 2 shows tendencies in three parameters based on the probability of identity by descent within breeds. Since 1990 in populations of three breeds (BA, LI and SM) average relatedness dominated. It led to increasing of inbreeding rate gain and more inbred animals were born. Intensity of inbreeding in the LI after 2003 has positive tendency in spite decreasing tendency of average relationship. The CH had similar tendency between 1990 and 2000 years. Since 1996 inbreeding intensity has increased and after 2000 year was over AR. Results in inbreeding rate and average relatedness tendencies are significantly dependence on sire selection strategy within all evaluated breeds.

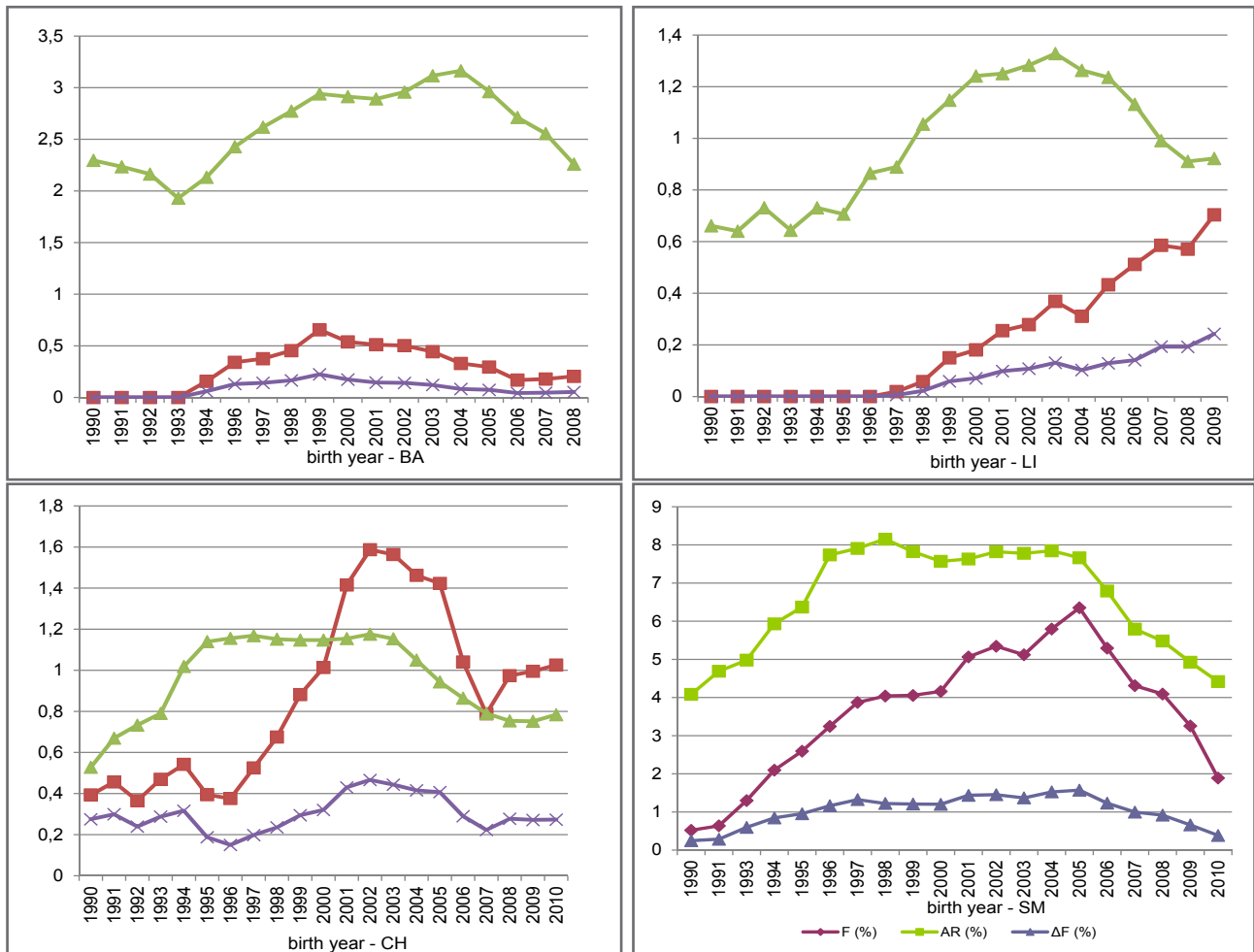
Genetic diversity within populations after a small number of generations can be measured using parameters derived from the probabilities of gene origin (Boichard et al., 1997). Results of parameters based on probability of gene origin are shown in Table 2. The highest number of founders was found in the CH (1224) followed by LI (778) and SM (233), while the lowest number of founders was found in the BA (180). CH and LI breeds had higher effective number of founders than BA and SM. The similar tendency was observed in effective number of ancestors. The  $f_e / f$  ratio was the highest in the BA (0.44) indicating more balanced founder contributions compared to CH, LI and SM despite that BA overall reference population size was the smallest. Disequilibrium of founder contributions in all evaluated breeds indicates perhaps excessive use of some sires in mating programmes as parents of the next generations. The  $f_a / f_e$  ratio in all four breeds is comparably higher than published Melka et al. (2013). It indicate that BA, CH, LI and SM breeds had not passed through such substantial bottlenecks as was described in Guernsey population. The  $f_g / f_e$  ratio measures the impact of genetic drift excluding the effect of founder contributions on

**Table 1** Size of pedigree and reference populations and pedigree completeness

Parameters	BA	CH	LI	SM
Pedigree population	481	3,955	2,063	916
Reference population	109	1,762	773	428
Maximum generation traced	5.39	5.29	4.95	6.30
Mean complete equivalent generations	3.98	3.82	3.38	4.38



**Figure 1** Reference population pedigree completeness index by the generations of the ancestors in Blonde d'Aquitaine-BA, Charolais-CH, Limousine-LI, Simmental-SM breedso



**Figure 2** Trends of average inbreeding, coancestry and inbreeding intensity gain in Blonde d'Aquitaine-BA, Charolais-CH, Limousine-LI, Simmental-SM breeds

genetic diversity. Impact of genetic drift was higher in the BA (0.16) than in LI (0.18), CH (0.22) and SM (0.27). Estimated measures based on gene origin in assessed breeds are comparable with results of 9 Spanish beef cattle breeds (Gutiérrez et al., 2003), Canadian Holstein and Jersey cattle (Stachowicz et al., 2011) and Guersey breed (Melka et al., 2012). Overall, genetic diversity has been lost in BA, CH, LI and SM breeds since 1990 due to

unequal contributions of founders and random genetic drift. Genetic diversity was more affected in the BA and LI populations by genetic drift than unequal founders' contributions. Number of ancestors explaining 50% of genetic diversity was small, comparing with 10–415 ancestors in Spanish beef breeds (Gutiérrez et al., 2003) but similar results presented Stachowicz et al. (2011) and Melka et al. (2012).

**Table 2** Parameters based on gene origin for the reference populations by breeds

Parameters	BA	CH	LI	SM
Total number of founders, $f$	180	1224	778	233
Effective number of founders, $f_e$	80	232	226	30
Effective number of ancestors, $f_a$	19	72	54	13
Founder genome equivalent, $f_g$	13	51	41	8
$f_e/f$ ratio	0.44	0.18	0.29	0.13
$f_a/f_e$ ratio	0.23	0.31	0.23	0.43
$f_g/f_e$ ratio	0.16	0.22	0.18	0.27
Number of ancestors explaining 50% of gene pool	7	27	23	4

**Table 3** Genetic diversity loss due to unequal founder contributions and random genetic drift ( $1 - GD$ ), unequal founder contributions ( $1 - GD^*$ ), random genetic drift ( $GD^* - GD$ ) by breeds

Parameters	BA	CH	LI	SM
GD	0.964	0.990	0.988	0.937
1 - GD	0.035	0.009	0.011	0.062
GD*	0.988	0.995	0.995	0.966
1 - GD*	0.012	0.004	0.004	0.033
GD* - GD	0.023	0.005	0.007	0.029

Measures of genetic diversity loss can be estimated from  $f_e$ ,  $f_g$  (Table 3). The amount of genetic diversity in the reference population accounting for diversity loss due to genetic drift and unequal founder contributions was the highest in the SM (6.2%), following the BA (3.5%), LI (1.1%) and CH (0.9%). The proportion of genetic diversity loss due to genetic drift was higher in this study (BA, CH, LI) than the loss of genetic diversity due to unequal founder contributions. Similar results published Melka et al. (2012), Stachovicz et al. (2011). However, Tang et al. (2013) reported that the main cause of genetic diversity loss in three Chinese swine breeds was unequal genetic founders contributions.

### Conclusions

The genealogic analyses of Blonde d'Aquitaine, Charolais, Limousine and Simmental indicated accumulation of inbreeding intensity and relationship among animals since 1990. All assessed breeds are small size populations also from the genetic point of view. Loss of genetic diversity is evident and was more caused by the genetic drift than unbalanced contributions of founders. Application of optimum contributions mating and increasing of population size in all assessed breeds could help maintain genetic diversity.

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