The Impact of Cytoplasmic Inheritance on Sperm Quality in Fleckvieh Bulls

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Summary

Detrimental impact of certain mitogenome mutations on sperm quality traits, and consequently on male fertility is well documented in humans. With a quantitative genetic mixed model, we analysed the impact of cytoplasmic effects, maternal lineages treated as random effect, on sperm quality traits in 554 Austrian Fleckvieh bulls. We have observed that 2% of the phenotypic variance for transformed total number of spermatozoa is due to cytoplasmic (maternal lineage) effects. Regarding percent of viable live spermatozoa, no cytoplasmic effects were detected. However, the observed effects still need to be further evaluated from three perspectives, the analysis of the mitogenome polymorphism effects and the impact of the mitogenome effects on the realised fertility as well as on the whole production economically.

Key words

cattle, cytoplasmic inheritance, Fleckvieh, maternal lineage, semen quality

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Introduction

Cytoplasmic inheritance is one of the biological mechanisms that explain influence from a dam on its offspring, i.e. maternal effect (Legates, 1972; Hohenboken, 1985). Genetic mechanism of the cytoplasmic inheritance is mitogenome (mtDNA), a small circular molecule that in mammals is almost exclusively inherited from maternal lineage (Hutchison et al., 1974). In cattle, mtDNA is a closed loop of 16 338 bp that codes for 37 genes from which 13 are responsible for coding essential proteins for the oxidative phosphorylation (Anderson et al., 1982), crucial biochemical pathway that generates adenosine triphosphate (ATP). Cytoplasmic effects on the milk production (Kennedy, 1986; Boettcher et al., 1996; Boettcher and Gibson, 1997) and growth (Pun et al., 2012) traits were widely studied in cattle populations. It was found that such effects account from 1% to 5% of phenotypic variation (Mezzadra et al., 2005). Garmyn et al., (2011) have analysed the impact of cytoplasmic effects on several fertility traits such as scrotal circumference (SC), percent motility (MOT), percent primary abnormalities (PRIMA), percent secondary abnormalities (SECA) and percent of total abnormalities (TOTA) in Angus bulls. In Garmyn et al., (2011) the observed cytoplasmic effects, expressed as a proportion of phenotypic variance accounted, were negligible for SC, SECA and TOTA (<0.2%) and small for MOT (1.3%) and PRIMA (2.3%). Holyoake et al. (2001) reported the impact of mtDNA polymorphism on the human sperm quality and consequently male fertility. As shown by Sampson et al. (2001) some point mutations or multiple deletions in mtDNA molecule are associated with asthenozoospermia and oligoasthenozoospermia. Furthermore, sperms are shown to be prone to developing deletions of mtDNA that are associated with decline of motility and fertility, and sperm quality traits can be influenced by the mtDNA haplogroups (Kumar and Sangeetha, 2009).

We are not aware of a study that evaluated cytoplasmic effects regarding the sperm quality traits in Fleckvieh cattle. The aim of this study was to estimate the magnitude of cytoplasmic (maternal lineage) effects on the transformed total number of spermatozoa (TTS) and percent of viable live spermatozoa (VLS) in 554 Austrian Fleckvieh bulls.

Material and methods

Sperm quality data for 554 Austrian Fleckvieh bulls were obtained from three Austrian artificial insemination stations: Gleisdorf station in Styria; Hohenzell station in Upper Austria; and Wieselburg station in Lower Austria. All three stations keep bulls in tie-stalls and collect semen several times a week. The traits recorded routinely for every ejaculate were semen collector, volume, sperm concentration, percentage of viable spermatozoa, and motility. Motility was not recorded at the Gleisdorf station, so was not further analysed. In our analyses sperm quality was represented by two traits, TTS and VLS (%). To obtain TTS we first calculated the total number of spermatozoa (x 10^9) in ejaculate by multiplying the volume with the concentration of spermatozoa. Following, we used the TRANSREG procedure with the BOXCOX option (SAS Institute, 2011; Box and Cox, 1964) to transform on a normal distribution scale the TTS variable. The calculated transformation was determined by the formula: TTS = (total number of spermatozoa$^{0.3}$ - 1) / 0.3. After data editing and exclusion of all spurious records our analyses were based on 19,720 ejaculates (Table 1). For detailed description on data editing see Ferenčaković et al. (2017).

Table 1. Descriptive statistics for total number of spermatozoa (TS), transformed total number of spermatozoa (TTS) and percent of viable live spermatozoa (VLS) in Fleckvieh bulls from three AI stations

Trait	N	Mean	SD	Min	Max
TS (10^9)	19,720	7.852	3.624	1.103	22.554
TTS	19,720	2.712	0.874	0.089	5.158
VLS (%)	19,720	66.4	5.8	35.0	90.0

Pedigree data on 41,090 animals extending back to the 1930s were provided by Zuchtdata EDV-Dienstleistungen GmbH. The pedigree was checked and recoded using CFC software (Sargolzaei et al., 2006) and pedigree inbreeding coefficients for full pedigree (F_{PED}) were calculated using ENDOG v4.8 (Gutiérrez and Goyache, 2005). Pedigree structure is shown in details in Table 2. After elimination (tree cut) of non-informative animals, a total of 5,990 animals were involved in the pedigree. The proportion of non–base animals was 96.3%. Among them, 69.8% had both parents known. Small proportion of animals (3.7%) was considered as the base population. The average number of progeny per sire was almost four (3.7). Dams had on average 1.6 progenies. Maternal lineages were identified by tracing female paths to the last female ancestor in the herd. MaGelLAn 1.0 (maternal genealogy lineage analyzer) software, *mag_stat module*, was used to assign the maternal lineage founder to each bull (Ristov et al., 2016). In the analysis, all maternal lines were considered.

Semen quality traits, TTS and VLS, ($y_{ijklmnop}$) were modelled using the single-trait animal model. The mixed animal model showing the best fit for analyzed traits is presented in scalar notation by the following equation [1]:

$$y_{ijklmnoprs} = \mu + b_1(x_{ijklmnoprs} - \bar{x}) + age_i + season_j + year_k + collection\_interval_l + semen\_colector_m + station_n + cyt_{or} + p_{pr} + a_r + e_{ijklmnoprs} \quad [1],$$

where effect of age class (age_i), season ($season_j$), year ($year_k$), collection interval ($collection\_interval_l$), semen collector ($semen\_collector_m$), and AI station ($station_n$) were considered as fixed class effects. F_{PED} ($x_{ijklmnoprs}$) was used as covariate and was modelled as linear regression. Maternal lineage (cyt_{or}), permanent environment (p_{pr}), and direct additive genetic effect (a_r) were included in the model as random effects. Similar model, although without maternal lineage and permanent environment effects, was used in Ferenčaković et al. (2017). Covariance components were estimated by Residual Maximum Likelihood (REML) method using VCE-6 program package (Groeneveld et al., 2008).

Table 2. The pedigree structure of the Fleckvieh population used in the analysis

Item	Number
Animals with records	554
Non–base animals	5,766
- both parents known	4,024
- only sire known	1,618
- only dam known	124
Base animals	224
Proportion of base animals (%)	3.7
Average number of progenies per sire	3.7
Average number of progenies per dam	1.6
Maternal lineages for analysed bulls	369
Total number of animals	5,990

Results and discussion

Cytoplasmic effects accounted for 2% of the phenotypic variance for TTS, while the proportion of explained phenotypic variance by maternal lineage was not observed for VLS (Table 3).

Table 3. Estimated genetic parameters with standard errors for transformed total number of spermatozoa and percent of viable spermatozoa

Trait	h^2	cyt^2	p^2
Transformed total number of spermatozoa	0.15±0.05	0.02±0.01	0.14±0.04
Percent of viable spermatozoa	0.10±0.04	0.00±0.00	0.18±0.03

h^2 – heritability, cyt^2 – ratio for maternal lineage, p^2 – ratio for permanent environment effect

Results are different from those obtained by Garmyn et al. (2011) who, by using somewhat different model, observed small cytoplasmic effects on the MOT (1.3%). Sperm motility is crucial factor for male fertility and the energy for movement is coming from mitochondria in sperm midpiece. Ruiz-Pesini et al. (1998) showed relationships between sperm motility and mitochondrial chain enzyme activities as well as between the number of mitochondria surrounding the midpiece and motility.

It is important to note that obtained impact of cytoplasmic effects on sperm quality is underestimated as bulls used in this analysis have already been preselected for the semen quality. For example, bulls with extremely low semen quality or serious defects were already excluded. In addition, we do not expect that all maternal lineages have the same mtDNA so it will be of interest to further extend our analysis and determine haplotypes or mutations contributing to the observed variability. Also, the cattle reproduction is very complex trait and it would be of interest to estimate the correlation between TTS variation and the realised fertility such as it is, for example, non-return rate of the bulls.

Conclusion

We have observed the impact of cytoplasmic (maternal lineage) effects on the transformed total number of spermatozoa, but not on the percent of viable live spermatozoa in Fleckvieh bulls from three Austrian AI stations. The phenotypic variance that was assigned to cytoplasmic effect was 2%. Although the observed effect could be considered as small, it still need to be further evaluated from three perspectives: (1) the analysis of the mitogenome polymorphism effects on sperm quality traits, (2) the impact of the effects on the effective fertility (for example non-return rate), (3) the economic impact on whole production.

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