J. Dairy Sci. 100:1–10
https://doi.org/10.3168/jds.2016-12164
© American Dairy Science Association®, 2017.

Genome-wide mapping and estimation of inbreeding depression of semen quality traits in a cattle population

Maja Ferenčaković,* Johann Sölkner,†[1] Miroslav Kapš,* and Ino Curik*
*Department of Animal Science, Faculty of Agriculture, University of Zagreb, Svetosimunska 25, 10000 Zagreb, Croatia
†University of Natural Resources and Life Sciences Vienna, Department of Sustainable Agricultural Systems, Division of Livestock Sciences, Gregor Mendel Str. 33, A-1180 Vienna, Austria

ABSTRACT

Inbreeding depression is known to affect quantitative traits such as male fertility and sperm quality, but the genetic basis for these associations is poorly understood. Most studies have been limited to examining how pedigree- or marker-derived genome-wide autozygosity is associated with quantitative phenotypes. In this study, we analyzed possible associations of genetic features of inbreeding depression with percentage of live spermatozoa and total number of spermatozoa in 19,720 ejaculates obtained from 554 Austrian Fleckvieh bulls during routine artificial insemination programs. Genome-wide inbreeding depression was estimated and genomic regions contributing to inbreeding depression were mapped. Inbreeding depression did affect total number of spermatozoa, and such depression was predicted by pedigree-based inbreeding levels and genome-wide inbreeding levels based on runs of homozygosity (ROH). Genome-wide inbreeding depression did not seem to affect percentage of live spermatozoa. A model incorporating genetic effects of the bull, environmental factors, and additive genetic and ROH status effects of individual single-nucleotide polymorphisms revealed genomic regions significantly associated with ROH status for total number of spermatozoa (4 regions) or percentage of live spermatozoa (5 regions). All but one region contains genes related to spermatogenesis and sperm morphology. These genomic regions contain genes affecting sperm morphogenesis and efficacy. The results highlight that next-generation sequencing may help explain some of the genetic factors contributing to inbreeding depression of sperm quality traits in Fleckvieh bulls.
Key words: inbreeding depression, sperm quality, runs of homozygosity, single nucleotide polymorphism

Received October 17, 2016.
Accepted February 27, 2017.
[1]Corresponding author: johann.soelkner@boku.ac.at

INTRODUCTION

Inbreeding depression, defined as reduction of the population mean for a quantitative trait such as size, fertility, vigor, yield, and fitness caused by inbreeding, has long been known to occur widely in the plant and animal kingdom, including humans (Kristensen and Sorensen, 2005; Bittles and Black, 2010; Leroy, 2014). Despite its importance, the genetic basis of inbreeding depression, such as the gene pathways or numbers of loci involved, remains unclear. Many studies of animal populations have dealt with such depression simply by performing regression of individual performance on individual pedigree inbreeding coefficients (Kristensen and Sorensen, 2005; Leroy, 2014).

Genome sequencing technologies provide an avenue for more sophisticated approaches to understand the genetic basis of inbreeding depression. Runs of homozygosity (**ROH**), regions of the genome without heterozygosity in the diploid state (Gibson et al., 2006), have been used to quantify individual inbreeding in humans (McQuillan et al., 2008), cattle (Sölkner et al., 2010; Ferenčaković et al., 2011; Purfield et al., 2012), and pigs (Bosse et al., 2012), and ROH-based results appear to be more accurate than traditional pedigree-based estimates (Curik et al., 2014) and also more accurate than inbreeding coefficients calculated when deriving the genomic relationship matrix based on variance of genotype values (Van Raden, 2008), see Sölkner et al. (2010). These studies have established ROH inbreeding as an indicator of inbreeding level and they show that this can be calculated with respect to a given reference population, a specific genomic region, or even individual SNP. This opens up several possibilities for exploring the genetic mechanisms of inbreeding depression. In human genetics, the drawback with this approach is the need for extremely large numbers of samples, given that most human populations show low inbreeding levels varying little between individuals (Keller et al., 2012; Howrigan et al., 2016). The higher inbreeding levels and particularly the much higher variability of inbreeding in livestock populations opens up

the opportunity to analyze local effects of inbreeding at reasonable power with much smaller sample sizes.

Using ROH-derived inbreeding coefficients ($\boldsymbol{F_{ROH}}$) and other estimators of molecular inbreeding, Bjelland et al. (2013) identified inbreeding depression of lactation performance and reproductive traits in Holstein cattle. Those authors concluded that only F_{ROH} can distinguish between identical-by-descent (**IBD**) markers and identical-by-state markers. Pryce et al. (2014) used ROH to pinpoint specific genomic regions associated with inbreeding depression of calving interval and milk production. Howard et al. (2015) analyzed milk production and calving interval of Jersey cows in the United States and Australia and found different ROH signals for the 2 genetically linked populations for dairy traits and only one significant signal for calving interval in the US population.

Inbreeding negatively affects the reproductive performance of male animals. High levels of inbreeding may be a cause of poor semen quality (Wildt et al., 1982; Margulis and Walsh, 2002; Aurich et al., 2003; van Eldik et al., 2006). Using pedigree data and analyses of sperm quality, Maximini et al. (2011) showed that inbreeding depression reduced fertility of Fleckvieh (i.e., dual-purpose Simental) bulls. In that study, inbreeding affected semen volume, total number of spermatozoa, percentage of live spermatozoa, and sperm motility, all of which serve as quantitative semen quality traits.

Austrian and German Fleckvieh breeding organizations and AI stations, together with research institutions, are very actively pursuing ways of improving male fertility by searching for and acting on mutations causing poor male fertility, either via less viable sperm (Pausch et al., 2014) or embryonic death (Pausch et al., 2015).

To identify genomic regions with effect on inbreeding depression of male sperm quality, we analyzed ROH in a large set of bulls from 3 Austrian AI stations. Using bovine SNP50 Beadchip technology (Illumina, San Diego, CA), we calculated genome-wide autozygosity from ROH of different minimum lengths (Ferenčaković et al., 2013) and compared the ability of ROH and pedigree to predict inbreeding depression of 2 sperm quality traits. We then associated, through autozygosity mapping, both ROH status and genotype of each SNP with sperm quality traits.

MATERIALS AND METHODS

A total of 1,799 Austrian Fleckvieh bulls were genotyped using the bovine SNP50 Beadchip v1 (Illumina), which contains 54,001 SNP (**50k**). Those genotypes and pedigree data on 41,090 animals extending back to the 1930s were provided by ZuchtData EDV-Dienstleistungen GmbH (Vienna, Austria). The pedigree was checked and recoded using CFC software (Sargolzaei et al., 2006). From the pedigree data, the equivalent complete generations and pedigree inbreeding coefficients for full pedigree ($\boldsymbol{F_{PED}}$) and for 5 generations ($\boldsymbol{F_{PED5}}$) were calculated using ENDOG v4.8 (Gutiérrez and Goyache, 2005). The equivalent complete generations, defined as the sum of $(1/2)^n$, where n is the number of generations separating the individual from each known ancestor (see the ENDOG v4.8 User's Guide), represents a measure of pedigree quality that indicates distance to the reference population where all individuals are unrelated. The F_{PED5} was calculated to quantify recent inbreeding, up to 5 generations back, and provide estimates of recent inbreeding depression.

Sperm quality data were obtained from 3 Austrian AI stations: Gleisdorf station in Styria (7,704 ejaculates, 301 bulls sampled from 2000 to 2010), Hohenzell station in Upper Austria (16,671 ejaculates, 309 bulls sampled from 2000 to 2009), and Wieselburg station in Lower Austria (15,514 ejaculates, 293 bulls sampled from 2000 to 2009). All 3 stations keep bulls in tie-stalls and collect semen several times a week, using a dummy or teaser animal and artificial vagina. Staff at the Hohenzell and Wieselburg stations routinely collected ejaculate 2 to 3 times per day from the same bull, whereas workers at the Gleisdorf station collected only 1 ejaculate per day from the same bull. The traits recorded routinely for every ejaculate were semen collector, volume, sperm concentration, percentage of viable spermatozoa, and motility. Motility was not recorded for ejaculates at the Gleisdorf station. Total number of spermatozoa ($\times 10^9$) in ejaculate was calculated from the volume and concentration of spermatozoa.

Taking into account the SNP density of the bovine 50k SNP chip, we performed quality control and determined ROH following the settings proposed by Ferenčaković et al. (2013). Only autosomal SNP that were assigned to a chromosome were used in the analysis. We also excluded SNP for which more than 10% of genotypes were missing and SNP with an Illumina GenCall score $\leq$0.7 or an Illumina GenTrain score $\leq$0.4. Bulls with more than 5% of their genotypes missing were excluded from further analysis. The SNP positions used were from UMD 3.1 (University of Maryland, College Park). The ROH were called if 15 or more consecutive homozygous SNP were present at a density of at least 1 SNP every 100 kb, with gaps of no more than 1,000 kb between them. We defined 4 ROH length categories (in Mb): [2, 4], (4, 8], (8, 16], and >16, see Ferenčaković et al. (2013) for the rationale. In different categories, we allowed different number of heterozygous and missing SNP. One heterozygous SNP was allowed in category >16, whereas in others we did not allow

Download English Version:

https://daneshyari.com/en/article/5542264

Download Persian Version:

https://daneshyari.com/article/5542264

Daneshyari.com