J. Dairy Sci. 98:2738–2745
http://dx.doi.org/10.3168/jds.2014-8601

A limited number of Y chromosome lineages is present in North American Holsteins

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ABSTRACT

Holsteins are the most numerous dairy cattle breed in North America and the breed has undergone intensive selection for improving milk production and conformation. Theoretically, this intensive selection could lead to a reduction of the effective population size and reduced genetic diversity. The objective of this study was to investigate the effective population size of the Holstein Y chromosome and the effects of limited Y chromosome lineages on male reproduction and the future of the breed. Paternal pedigree information of 62,897 Holstein bulls born between 1950 and 2013 in North America and 220,872 bulls evaluated by multiple-trait across-country genetic evaluations of Interbull (Uppsala, Sweden) were collected and analyzed. The results indicated that the number of Y chromosome lineages in Holsteins has undergone a dramatic decrease during the past 50 years because of artificial selection and the application of artificial insemination (AI) technology. All current Holstein AI bulls in North America are the descendants of only 2 ancestors (Hulleman and Neptune H) born in 1880. These 2 ancestral Y-lineages are continued through 3 dominant pedigrees from the 1960s; namely, Pawnee Farm Arlinda Chief, Round Oak Rag Apple Elevation, and Penstate Ivanhoe Star, with a contribution of 48.78, 51.06, and 0.16% to the Holstein bull population in the 2010s, respectively. The Y-lineage of Penstate Ivanhoe Star is almost eliminated from the breed. The genetic variations in the 2 ancestral Y-lineages were evaluated among 257 bulls by determining the copy number variations (CNV) of 3 Y-linked gene families: *PRAMEY*, *HSFY*, and *ZNF280BY*, which are spread along the majority (95%) of the bovine Y chromosome male-specific region (MSY). No significant difference was found between the 2 ancestral Y-lineages, although large CNV were observed within each lineage. This study suggests minimal genetic diversity on the Y chromosome in Holsteins and provides a starting point for investigating the effect of the extremely limited number of Y-lineages on male reproduction and other traits important for the future of the Holstein breed.

Key words: Y chromosome, copy number variation, paternal pedigree, Holstein, effective population size

INTRODUCTION

Holstein cattle, renowned for their milk production and recognized by their distinctive black (or red)-and-white markings, are the largest dairy cattle population in the world. In 2012, there were ~26 million Holstein cows registered by the World Holstein-Friesian Federation. Although the origin of Holsteins can be traced to the development of cattle in the north region of the Netherlands over the last 2 millennia (Lush et al., 1936), the history of Holsteins in North America is very short, only about 150 yr. The first group of Holsteins was brought to the United States in the 1850s (Lush et al., 1936). The breed quickly gained the attention of American cattle breeders and dairymen because of its reputation for high yields of quality milk. From then on, 8,800 Holsteins were imported to the United States before importation ceased in the early 1900s due to an outbreak of foot-and-mouth disease in mainland Europe (http://www.holsteinusa.com/holstein_breed/breedhistory.html). Obviously, the imported animals were sufficient for initiation of an American breeding program. To date, there are ~9.2 million milk cows in the United States and ~90% of these are Holstein, supplying ~86,000 million kg of milk (USDA, 2010). The average milk production per cow per year is 9,455 kg in the United States, which is significantly higher than that in other countries (WHFF, 2012).

The Holstein breeders in the United States established the first Holstein-Friesian herd book (**HHB**) association in 1871 (Lush et al., 1936), which is now the world's largest Holstein association with more than 19 million registered Holsteins (http://www.holsteinusa.com/holstein_breed/breedhistory.html). In the late 1940s, modern AI was established [Perry, 1968; Wolf Foundation Prize in Agriculture in 1981 (http://www.wolffund.org.il/index.php?dir=site&page=winners&cs

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Received July 10, 2014.
Accepted December 22, 2014.
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=255); Foote, 2002], and it soon became routine practice to replace the natural mating system, which allowed the use of superior, proven bulls by Holstein breeders across the country in the late 1960s. In addition to AI technology, genetic and reproductive approaches, such as BLUP and multiple ovulation and embryo transfer (MOET), were applied to select animals that have a high genetic potential. Since 2009, genomic selection has been applied to all bulls that enter the AI program in the United States and Canada (Hayes et al., 2009).

A half-century of intensive selection has significantly accelerated genetic improvement in milk production, along with a reduction in generation interval in recent years due to genomic selection. However, the intensive selection of bulls combined with worldwide use of a small number of elite sires has also led to a reduction in effective population size and reduced genetic diversity in Holsteins (Sargolzaei et al., 2007; Kim and Kirkpatrick, 2009).

In the present study, our focus was on the genetic diversity of Holstein bulls, particularly on the Y chromosome, as the Y is present in the male genome only. Two important features set the Y chromosome apart from the rest of the genome: (1) absence of homologous recombination on the male-specific region of the Y chromosome (**MSY**) during meiosis, and (2) male-limited transmission. Absence of recombination leads to the degeneration of Y chromosome gene or sequence during evolution, whereas male-limited transmission provides an ideal genomic niche to enhance male fitness because male-beneficial mutations on the Y chromosome are always transmitted through the sex (Rice, 1996; Bachtrog, 2013). One of the consequences of male-limited transmission is the reduction of the effective Y population size (Wilson Sayres et al., 2014). In an ideal breeding population in which the female-to-male breeding sex ratio is 1, there are only one-fourth as many Y-linked genes as autosomal genes and only one-third as many as X-linked genes (Tucker and Lundrigan, 1995). In most farm-animal breeding programs, such as in Holsteins, the female-to-male breeding sex ratio is significantly greater than 1 and the effective population size of Y chromosomes relative to X and autosomes is greatly reduced. The potential effects of a reduced effective population size of Y chromosomes in Holsteins and other livestock species have not been addressed. Genes on the mammalian Y chromosome are believed to be essential for maleness, spermatogenesis, and male fertility. Recent studies on the bovine Y chromosome gene copy number variations (**CNV**; Hamilton et al., 2009, 2011, 2012; Yue et al., 2013, 2014) have revealed that CNV of several multicopy gene families, including ***PRAMEY*** (preferentially expressed antigen in melanoma, Y-linked), ***HSFY*** (heat-shock transcription factor, Y-linked), and ***ZNF280BY*** (zinc finger protein 280B, Y-linked), are associated with fertility traits in Holstein bulls (Yue et al., 2013, 2014), indicating that the bovine Y chromosome has important influences on bull fertility.

The objective of this study was to investigate the effective population size of the Holstein Y chromosome and the effects of limited Y chromosome lineages on male reproduction and traits important for the future of the breed in North America.

MATERIALS AND METHODS

Retrieval of Paternal Pedigree Information of Holstein Bulls in North America

Holstein bull registration numbers, names, and birth dates in the United States and Canada that were enrolled in the National Association of Animal Breeders were retrieved following the August 2013 national genetic evaluations of the Council on Dairy Cattle Breeding (**CDCB**; https://www.cdcb.us/eval.htm). Paternal pedigree information of 62,987 bulls was available from CDCB files beginning from the 1940s. Beyond the 1940s, the paternal pedigrees were manually checked based upon online family tree searches available at http://www.holsteinusa.com/. In cases in which the sire information was missing from online family trees, manual inspection of a physical HHB collection maintained by Lawrence Specht (The Pennsylvania State University, University Park) was used to complete the pedigree analysis.

Determination of Founders and Ancestors for Holstein Bulls

We used founders and ancestors to specify the contribution of an individual Y chromosome in the paternal pedigree. Bulls that were born in the 1960s, when the national genetic evaluation program was implemented and AI technology was widely adopted, are referred to as AI-era "founders." For "ancestors," we refer to those sires that were born in the Netherlands and were imported to the United States before the 1900s, or sires that were traced back to 1880 based on the available HHB records.

In addition, we retrieved pedigrees of 220,872 Holstein-Friesian bulls from 35 different countries, evaluated by the International Genetic Evaluation Service through the multiple-trait across-country evaluation (MACE) of Interbull (http://www.interbull.org/ib/interbull) as of December 3, 2013, which is henceforth referred to as the Interbull database. As the paternal pedigree information was not available for most of the