

DOI 10.1515/pjvs-2015-0059

Short communication

# Detection of *Brachyspina* carriers within Polish Holstein-Friesian bulls

A. Ruść, S. Kamiński

University of Warmia and Mazury in Olsztyn, Department of Animal Genetics,  
Oczapowskiego 5, 10-718 Olsztyn, Poland

## Abstract

The aim of this paper was to verify the hypothesis whether carriers of genetic defect *Brachyspina* occur in the Polish Holstein-Friesian Cattle. PCR method was used to screen 78 Polish Holstein-Friesian bulls. Eight bulls were identified as heterozygotes for 3,3 kb deletion in the FANCI gene – the mutation causing *Brachyspina* defect. All carriers were sons of 3 sires: Cleitus Jabot, Sandy-Valley Bolton ET and Coyne-Farms Dorcy ET which were descendants of the US sire Sweet Haven Tradition (HOUSAM 1682485). Systematic screening of young bulls having in the pedigree *Brachyspina* carrier is necessary to prevent spreading of the recessive mutation in the dairy cattle population in Poland.

**Key words:** *Brachyspina*, carrier, Holstein-Friesian cattle, lethal defect

## Introduction

In current livestock breeding practices, especially of dairy cattle, the exchange of genetic material proceeds very rapidly by means of artificial insemination (AI), import of semen and transfer of embryos. The intensity and universality of these practices require the constant monitoring of the active population (especially of AI bulls) to detect potential defects and genetic diseases as well as pathways of their spreading since the use of a small number of bulls in breeding programs results in an increased number of related animals in the entire population of cattle. *Brachyspina* syndrome (OMIA 000151-9913) is the newest genetic defect in Holstein cattle caused by 3,3 kb deletion in the bovine fanconi anemia complementation group 1 (FANCI) gene (Charlier et al. 2012). Recessive homozygotes undergo early-term abortion or are

stillborn (Agerholm et al. 2006, 2007). All so far identified carriers were traced back to a common ancestor, the US Holstein sire Sweet Haven Tradition. The acronym of the disease – BY, is currently used in pedigree files in the most advanced dairy cattle breeding programs ([www.holsteinusa.com](http://www.holsteinusa.com)).

The aim of this paper was to verify the hypothesis whether *Brachyspina* carriers occur also in the Polish Holstein-Friesian Cattle.

## Materials and Methods

The present study includes 78 Polish Holstein-Friesian Black-and-White bulls born between 2005 and 2014 which were the property of domestic insemination companies or individual breeders providing young bulls to these companies. Genomic

---

Correspondence to: S. Kamiński, e-mail: [stachel@uwm.edu.pl](mailto:stachel@uwm.edu.pl), tel.: +48 89 523 47 14

DNA was isolated from ear tissue samples using the NucleoSpin Tissue kit according to the manufacturer's instructions (Macherey-Nagel GmbH & Co. KG, Dueren, Germany). BY carriers were identified by two separate PCR amplification as described earlier (Charlier et al. 2012).

## Results and Discussion

Among 78 bulls investigated, 8 were identified as heterozygotes for 3,3 Kb deletion of FANCI gene. All carriers were sons of 3 sires: Cleitus Jabot, Sandy-Valley Bolton ET and Coyne-Farms Dorcy ET which were descendants of US sire Sweet Haven Tradition (HOUSAM 1682485).

Our preliminary results clearly show that the BY carriers are present in Polish Holstein-Friesian cattle population and should be included in regular screening of AI dairy bulls. It should prevent spreading of the BY recessive mutation within cow population and steadily eliminate the number of new carriers as it was achieved for other genetic defects (BLAD, CVM) screened by our group (Czarnik et al. 2007, Ruś et al. 2013). The high incidence of BY carriers (ca. 10%) reported in this paper does not reflect the actual average frequency of BY in the whole population since animals included in the analysis were chosen by breeders as having in the pedigree data BY-positive ancestor.

Based on the analysis of pedigree of BY carriers which semen was imported to Poland in the last decade, we assume that the number of BY carriers is lower than BLAD and CVM carriers at initial stage of screening program (unpublished data). Data obtained in other countries however, show that the number of BY-positive bulls is still relatively high and can spread rapidly when ignored. For example, Charlier et al. (2012) reported 7,4% BY carriers and VanRaden et al. (2011) – 6%, both for Holstein breed. In the Chinese Holstein, Fang et al. (2013) found 13 carriers within 342 animals (206 bulls and 136 cows).

Since Holstein-Friesian cattle is one of the most inbred dairy cattle breed, closer international integration of national associations of Holstein cattle breeders and coordination of genetic defects discovery programs is necessary to decrease the risk of new recessive disorders and reduce their prevalence.

## Acknowledgements

This work was financially supported by UWM grants No. 0105-0804 and 0105-0316.

## References

- Agerholm JS, McEvoy F, Arnbjerg J (2006) Brachyspina syndrome in a Holstein calf. *J Vet Diagn Invest* 18: 418-422.
- Agerholm JS, Peperkamp K (2007) Familial occurrence of Danish and Dutch cases of the bovine brachyspina syndrome. *BMC Vet Res* 3: 8.
- Charlier C, Agerholm JS, Coppieters W, Karlskov-Mortensen P, Li W, de Jong G, Fasquelle C, Karim L, Cirera S, Cambisano N, Ahariz N, Mullaart E, Georges M, Fredholm M (2012) A Deletion in the Bovine *FANCI* Gene Compromises Fertility by Causing Fetal Death and Brachyspina. *PLoS One* 7: e43085.
- Czarnik U, Grzybowski G, Kamiński S, Prusak B, Zabolewicz T (2007) Effectiveness of a program aimed at the elimination of BLAD-carrier bulls from the Polish Holstein-Friesian cattle. *J Appl Genet* 48: 375-377.
- Fang L, Li Y, Zhang Y, Sun D, Liu L, Zhang Y, Zhang S (2013) Identification of brachyspina syndrome carriers in Chinese Holstein cattle. *J Vet Diagn Invest* 25: 508-510
- OMIA000151-9913. Online Mendelian Inheritance of Animals. <http://omia.angis.org.au/OMIA000151/>
- Ruś A, Hering D, Puckowska P, Barcewicz M, Kamiński S (2013) Screening of Polish Holstein-Friesian bulls towards eradication of Complex Vertebral Malformation (CVM) carriers. *Pol J Vet Sci* 16: 579-581
- VanRaden PM, Olson KM, Null DJ, Hutchison JL (2011) Harmful recessive effects on fertility detected by absence of homozygous haplotypes. *J Dairy Sci* 94: 6153-6161