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*Short communication*

# Eradication of carriers of Complex Vertebral Malformation (CVM) and *Brachyspina* in Polish Holstein-Friesian bulls

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## Abstract

CVM (*Complex Vertebral Malformations*) and *Brachyspina* (BY) are the most common autosomal recessive genetic defects occurring in the last two decades in Holstein dairy cattle around the world. Beginning from 2004 and 2014, 3035 and 338 Polish Holstein-Friesian bulls were tested to find carriers of CVM and BY, respectively. Among analyzed bulls 191 CVM carriers (6.29%) and 20 BY carriers (5.92%) were identified. No CVM carriers were observed beginning from 2016, whereas only single BY carriers was identified annually for the last 5 years. One bull turned to be double CVM/ BY carrier as a son of also double CVM/BY top Dutch sire (JABOT 90676-4-9). It is shown that CVM and BY defects are practically eradicated from Polish dairy cattle although incidental testing should be continued if new bulls with CVM or BY carriers in sire or dam pedigree will unexpectedly appear.

**Keywords:** Holstein bulls, genetic defect, CVM, *Brachyspina*

## Introduction

CVM (*Complex Vertebral Malformations*) and *Brachyspina* (BY) are autosomal recessive genetic defects which clinically manifest by vertebral anomalies, miscarriages and stillborn in Holstein cattle (OMIA 000001-9913, OMIA 000151-9913). The causal mutation for CVM is substitution G by T in position 559 of SLC35A3 (S*olute Carrier family 35)* cDNA leading to exchange of Val by Phe in position 180 of protein – transporter of UDP-N-acetyloglucosamine (Thomsen et al. 2006). Brachyspina (BY) is caused by 3300 bp deletion of FANCI gene (*Fanconi Anemia Complementation – group I*) resulting in deletion 25-27 exons and creating premature stop codon leading to mRNA degradation *via* NMRD (Charlier et al. 2012). Both mutations were brought into Polish dairy cattle by import of semen of top bulls from USA and Netherlands. In our earlier initial studies we reported first cases of carriers for both defects (Ruść and Kamiński 2013, 2015). Since that time systematic screening for CVM and BY carriers was undertaken in order to eliminate all carriers used in insemination of cows. In this paper, successful effect of this screening is presented.

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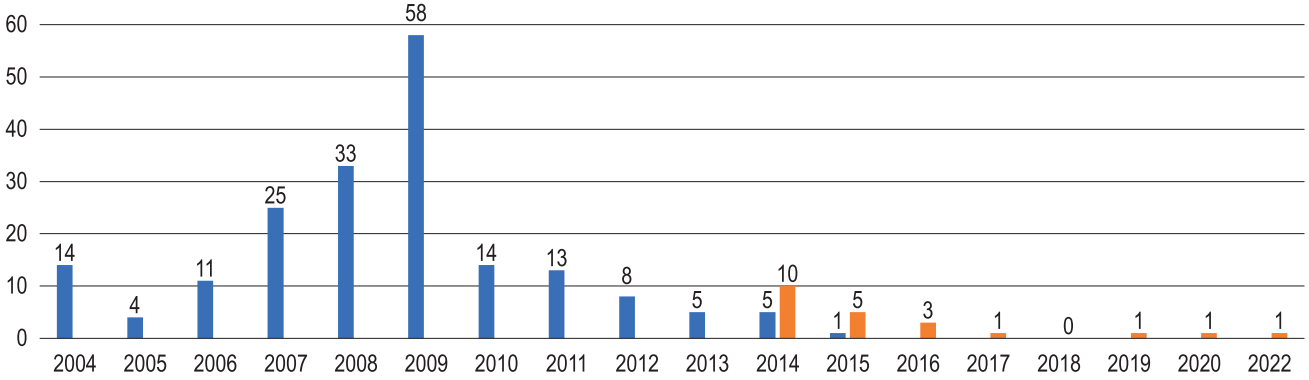


Fig 1. Number of Complex Vertebral Malformations (CVM) carriers (in blue) and *Brachyspina* (BY) carriers (in orange) detected in Polish Holstein-Friesian bulls between 2004-2021 and 2014-2021, respectively.

## Materials and Methods

Beginning from 2004 (CVM) and 2014 (BY), 3035 and 338 Polish Holstein-Friesian bulls were tested to find carriers of CVM and BY, respectively. ½ commercial straw of semen was used to isolate genomic DNA by MasterPure DNA Purification Kit (Promega). CVM carriers were identified by cost-effective and home-developed PCR-SSCP (Ruść and Kamiński 2007) but BY carries were detected by AS-PCR licensed technique (Charlier et al. 2012). For both defects results were authorized by license agreement filed to the University of Warmia and Mazury which approved testing as reliable and legal in pedigree data.

## Results and Discussion

Among analyzed bulls, 191 CVM carriers (6.29%) and 20 BY carriers (5.92%) were identified. The number of CVM carriers was increasing until 2009 and then dropped to 1 carrier in 2015. From 2016 to 2021 no CVM carrier was found (Fig. 1). In case of BY, the number of carriers was much lower but the trend was similar. At the beginning the number of BY carrier was the highest (10 bulls) and then decreased to 1 bull in 2017. The number of carriers reflected the number of bulls included for testing, e.g. in 2008 among 348 bulls, 33 were CVM carriers and in 2009, 58 CVM carriers were found in 568 bulls tested (complete data not shown). The general rule of screening program was to test each young bull which is considered to enter insemination center. Eliminating CVM and BY carriers at this moment automatically reduced the number of new carriers since these bulls were excluded as a sire of sire in breeding program. New carriers may come from the dams which are not tested for CVM and BY. That is why single BY carrier appeared in the last 5 years. This path for new carrier will soon disappear since dams will be exclusively the daughters of already tested sires, and being free for CVM and BY. Interestingly, one bull turned to be double CVM/ BY carrier (JAMEL PL000607299543, born in 1997 in Poland) as a son of also double CVM/BY top Dutch sire (JABOT 90676-4-9). JABOT was a father of 329 daughters held in 245 herds as well as 288 bulls used in insemination in Poland in the late 1990-ties (data retrieved from SYMLEK – database recording pedigree and milk performance traits in Poland). The results show that CVM and BY defects are practically eradicated from Polish dairy cattle although the testing should be continued if new bulls with CVM or BY carriers in sire or dam pedigree will unexpectedly appear. Results presented in the paper show that causal mutations for CVM and BY can be eliminated in relatively short time. Countries which have no own system for early monitoring of inherited defects among new born calves are exposed for unaware spreading of carriers until the owner of bull or breeding organization decide to release the information of testing result. The population of Holstein-Friesian cows in Poland is approximately 2,3 million (www.pfhb.pl), therefore it is believed that the eradication of CVM and BY carriers should lead to reducing losses in fertility and profitability of dairy cattle production in the future.

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