Report on the incidence of selected hereditary disorders in the Polish population of Holstein-Friesian cattle

M. Gozdek[12], D. Kamola[1], A. Prostek[1] and T. Sadkowski[1]

[1]Warsaw University of Life Sciences, Department of Physiological Sciences, Nowoursynowska 166, 02-787 Warsaw, Poland
Corresponding Author: m.gozdek@pfhb.pl
[2]Polish Federation of Cattle Breeders and Dairy Farmers, Zurawia 22, 00-515 Warsaw, Poland

Abstract

DUMPS (deficiency of uridine monophosphate synthase) and Haplotypes Holstein (HH1, HH3, HH4, HH5, HH6, HH7) are hereditary lethal autosomal recessive disorders that may affect Holstein cattle. The aim of the research was to determine the frequency of selected unfavorable mutations in the Polish population of Holstein-Friesian cattle. In the study 12 754 Polish Holstein-Friesian females were genetically tested. Results of the present study indicate, that the Polish population of dairy cattle is free from DUMPS. It turns out, that in 2019 the highest number of carriers were HH3 (5.81%), and the least HH7 (0.49%). In 2020, the highest number of carriers was HH5 (6.95%), and the least, similar to 2019, HH7 (0.45%).

Keywords: Cattle, gene frequency, microarrays, embryo mortality.

Introduction

Fertility is one of the most important traits in animal production. Significant decrease in fertility of dairy cattle in Poland has been arising for many years - a decrease in the fertilization and normal pregnancy rate is observed. These problems may result from the accumulation of genetic diseases in the population. Since individual sires generate tens of thousands of progeny via artificial insemination, cattle breeding populations are susceptible to the propagation of recessive diseases. The resulting threat was not immediately recognized because the heterozygous animals (i.e. asymptomatic carriers) do not express the symptoms of disease. Due to the frequent occurrence of genetic defects in combination with high breeding values, they tend to spread throughout the population. The increasing incidence of genetic diseases in cow herds is also the result of increased inbreeding within the entire world's cattle population. There are several specific genetic disorders associated with Holstein cattle. Particular attention should be paid to genetic defects causing embryo mortality. Among the most important defects are HH1, HH3, HH4, HH5, HH6, HH7, and DUMPS (Table 1).

Material and methods

Genetic data used in the present study was collected in the process of routine estimating breeding value (EBV). Poland as a member of EuroGenomics Cooperative, uses their customized arrays. Material for genotyping (12 754 samples) was collected in the years 2019 to 2020. Ear punch samples were collected with the use of AllFlex Tissue Sampling Unit (TSU). DNA extraction was processed with the use of Clean BloodandTissue DNA Kit (CleanNA, Netherlands) according to the producer manual in KingFisher DUO DNA processor (Thermo Scientific, USA). Normalized samples were processed according to Illumina HTS protocol. Beadchips were immediately scanned on Illumina iScan system, scans were analysed using GenomeStudio Software.

Results

Results of the present study indicate that the Polish population of Holstein dairy cattle is free from DUMPS (since 1999 testing of Polish population of Holstein dairy breeding bulls is mandatory). It turns out that in 2019 the number of HH1 carriers was 2,95%, HH3 - 5,81%, HH4 – 2,33%, HH5 – 5,16%, HH6 – 1,83%, HH7 – 0,49% (Holstein-Friesian population). In 2020 the number of carriers was correspondingly HH1 3,11%, HH3 – 4,42%, HH4 – 1,51%, HH5 – 6,95%, HH6 – 1,94%, HH7 – 0,45% (Figure 1).

Discussion

No DUMPS carriers were detected in the present study. Our research results are also in accordance with the results of studies by Patel *et al.* (2006) and Oner *et al.* (2010) who reported no carriers respectively in Indian and Turkey dairy cattle populations. Similarly, Korkmaz Agaoglu *et al*. (2015), Koshchaev *et al.* (2018), Debnath *et al.* (2016) and Citek *et al.* (2006) did not detect any DUMPS carrier in their studies. The number of HH1 carriers in Poland is higher than in Brazil Albertino *et al.* (2022) and the USA

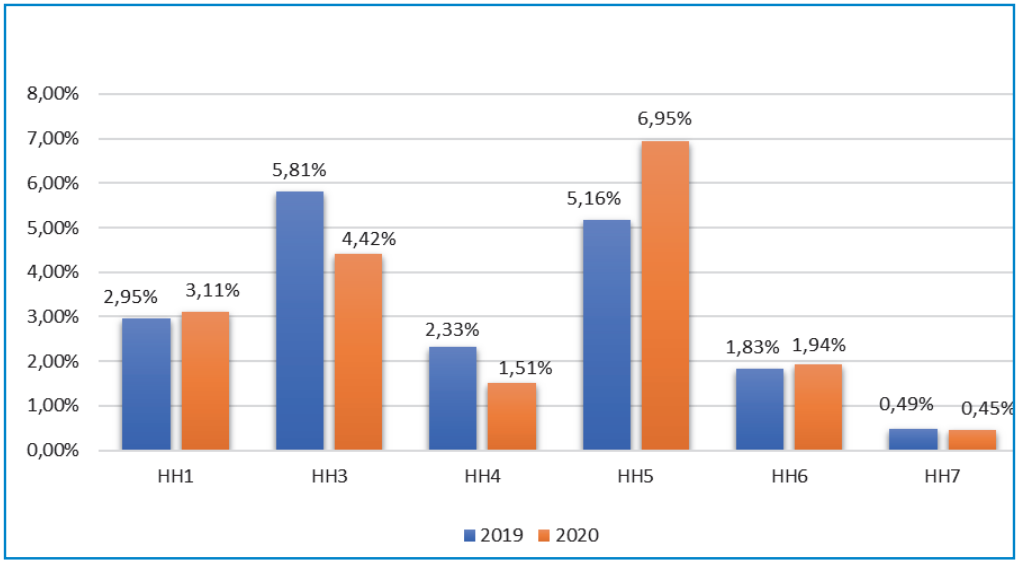

Figure 1. Percentage of carriers of selected hereditary disorders in the Polish population of Holstein-Friesian cattle.

Table 1. Basic characteristics of selected genetic disorders.

Disorder name	Species-specific name	Gene	Type of mutation	Year first reported
HH1	Haplotype HH1	APAF1	C>T	2012
HH3	Haplotype HH3	SMC2	T>C	2013
HH4	Haplotype HH4	GART	A>C	2013
HH5	Haplotype HH5	TFB1M	Deletion of 138kbp	2016
HH6	Haplotype HH6	SDE2	A>G	2018
HH7	Haplotype HH7	CENPU	Deletion of 4bp	2020
DUMPS	Deficiency of Uridine Monophosphate Synthase	UMPS	C>T	1993

Table 2. Percentage of carriers of selected genetic diseases in different countries.

Disorder name	N	Healthy	Carriers	% carriers	Country	References
DUMPS	500	500	0	0	Turkey	Korkmaz Agaoglu *et al.*, (2015)
DUMPS	73	73	0	0	Russia	Koshchaev *et al.* (2018)
HH1	248	248	0	0	Brazil	Albertino *et al.* (2022)
HH1	5729	5619	110	1,92	USA	Cole *et al.* (2016)
HH3	14 000	13 286	714	5,1	Germany	Schütz *et al.* (2016)
HH3	17 869	17 869	527	2,95	USA	Cole *et al.* (2016)
HH4	1218	1173	45	0,37	USA	Cole *et al.* (2016)
HH5	2100	1985	115	5,5	Germany	Schütz *et al.* (2016)

Cole *et al.* (2016) at a similar time. Our research results are also in accordance with the results of studies by Schütz *et al.* (2016) (the number of HH3 and HH5 carriers over 5%). Research by Cole *et al.* (2016) shows a lower percentage of HH4 carriers than in Poland in 2019-2020. There are no data available on the number of HH6 and HH7 carriers in other countries. The percentage of carriers of selected genetic diseases in individual countries is presented in Table 2.

Conclusion

In recent years, several causal mutations have been discovered in dairy cattle, most of which negatively affect fertility - causing increased embryo mortality. Most of these diseases are inherited autosomal recessively. From a breeding point of view, it is precisely recessive mutations that have highly negative effects. That kind of diseases occur only in animals that inherit two mutant alleles (homozygotes). Animals with a single altered allele (heterozygote) do not get sick, but can pass the mutations on to

their offspring. This makes it much more difficult to diagnose the disease at an early stage and to take preventive measures, e.g. to avoid using these pieces for matings. Therefore it is important to screen the population and identify carries to avoid economic losses due to these genetic disorders in the herd. Thanks to the regular application of genetic tests around the world, the population can be controlled.

Acknowledgement

The authors would like to thank the Breeders for providing the data for the study.

References

Albertino, L. G., Albuquerque, A., Ferreira, J. F., Oliveira, J., Borges, A. S., Patelli, T., and Oliveira-Filho, J. P. 2022. Allele Frequency of APAF1 Mutation in Holstein Cattle in Brazil. Front Vet. Sci. 9: 822224.

Citek, J., Rehout, V., Hajkova, J., Pavkova, J. 2006. Monitoring of the genetic health of cattle in the Czech Republic. Vet. Med. Czech. 51(6): 333–339.

Cole J.B., Null D.J., VanRaden P.M. 2016. Phenotypic and genetic effects of recessive haplotypes on yield, longevity, and fertility. Journal of Dairy Science. 99(9): 7274-7288.

Debnath, A., Kumar, A., Maan, S., Kumar, V., Joshi, V.G., Trilok N., Sangwan M.L. 2016. Molecular screening of crossbred cow bulls for important genetic disorders. Haryana Vet. 55(1): 93–96.

Korkmaz Agaoglu, O., Agaoglu, A.R., Saatci, M. 2015. Estimating allele frequencies of some hereditary diseases in Holstein cattle reared in Burdur Province, Turkey. Turk. J. Vet. Anim. Sci. 39: 338–342.

Koshchaev, A.G., Shchukina, V., Garkovenko, A.V., Ilnitskaya, E.V., Radchenko, V.V., Bakharev, A.A., Khrabrova, L.A. 2018. Allelic variation of marker genes of hereditary diseases and economically important traits in dairy breeding cattle population. J. Pharm. Sci. Res. 10(6): 1566–1572.

Oner, Y., Keskin, A., Elmaci, C. 2010. Identification of BLAD, DUMPS, Citrullinamia and Factor XI Deficiency in Holstein Cattle in Turkey. Asian J. Anim. Vet. Adv. 5(1): 60–56.

Patel, R.K., Singh, K.M., Soni, K.J., Chauhan, J.B., Sambasiva Rao, K.R.S., 2006. Lack of carriers of citrullinaemia and DUMPS in Indian Holstein cattle. J. App. Genet. 47: 239–242.

Schütz, E, Wehrhahn, C, Wanjek, M, Bortfeld, R, Wemheuer, WE, *et al*. 2016. The Holstein Friesian Lethal Haplotype 5 (HH5) Results from a Complete Deletion of TBF1M and Cholesterol Deficiency (CDH) from an ERV-(LTR) Insertion into the Coding Region of APOB. PLOS ONE 11(6): e0157618.