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REPORT ON THE INCIDENCE OF HEREDITARY DISORDERS (BLAD, DUMPS) IN THE POLISH POPULATION OF HOLSTEIN-FRIESIAN CATTLE

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ABSTRACT

BLAD (bovine leukocyte adhesion deficiency) and DUMPS (deficiency of uridine monophosphate synthase) are hereditary lethal autosomal recessive disorders that may affect Holstein cattle. The aim of the present study was to evaluate the frequency of BLAD and DUMPS genotypes in the Polish population of dairy cows. In the study 5 732 Polish Holstein-Friesian females were genetically tested for BLAD and DUMPS. Results of the present study indicate that the Polish population of dairy cattle is free from DUMPS as there were not carriers nor sick animals. For BLAD the study confirmed the present of carriers in the population to be at a low value (0.21%). Both BLAD and DUMPS may strongly affect the profitability of a dairy farm, therefore, in order to avoid the economic losses caused by the presence of these genetic disorders in the herd it is important to screen the population and detect carriers as soon as possible.

Key words: BLAD, DUMPS, cattle, gene frequency, microarrays

INTRODUCTION

Genetic disorders are caused by a hereditary change in the structure of DNA that may have a negative impact on vitality. In cattle, the autosomal recessive genetic diseases are the most often breed-specific. There are several specific genetic disorders associated with Holstein cattle, among the most important are bovine leukocyte adhesion deficiency (BLAD) and deficiency of uridine monophosphate synthase (DUMPS).

BLAD

BLAD is a lethal autosomal recessive genetic disease that occurs primarily in Holstein cattle. It spread in Europe in the early 1990s, following the beginning of the use of frozen semen. Its essence is the loss of defence functions of leukocytes. Calves homozygous for this mutation are characterized by a lack of immunity. BLAD carriers show full immunological (defence) efficiency. They are also often of a high breeding value. Among other things, it was shown that mutation-bearing cows produced more milk and protein than their half-sisters with the correct genotype. An improved performance of heterozygotes was one of the reasons for the spread of this mutation in the population [Osten-Sacken 2004].

Animals homozygous for the disease inducting mutation are characterized by a lack of immunity and suffer from frequent, recurrent bacterial infections, ulcers inside and around the mouth, diarrhoea and frequent gastrointestinal inflammations that prevent proper assimilation of feed. The bodyweight of diseased animals usually accounts for around 60% of a normal one [Osten-Sacken 2004]. These symptoms lead to delayed growth and development of calves, which usually die in the first months of life as a result of infections devastating their bodies. Infections are caused by the inability of leukocytes to pass from the bloodstream into infected tissue. This inability is due to the lack of a membrane glycoprotein (the leukocyte integrin beta–2 sub-

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unit; CD18). These glycoproteins – integrins – are vital to cell-cell and cell-substratum adhesion reactions in the body. Such adhesions are centre of anti-inflammatory reaction [Nagahata 2004].

The molecular basis of this disorder was discovered in 1992 [Shuster et al. 1992]. The reason for BLAD is the point mutation in the ITGB2 gene encoding the integrin beta–2 subunit. There is a change of adenine (A) to guanine (G) in the coding sequence of this membrane glycoprotein (thus mutation is called c.383A>G), and consequently the amino acid change from aspartic to glycine (p.Val128Ala) [Daetwyler et al. 2014]. VanRaden et al. [2011] using BovineSNP50 BeadChip confirmed the mapping of this disorder to bovine chromosome 1 (BTA1) [VanRaden et al. 2011] (Table 1).

Bovine leukocyte adhesion deficiency was first described in 1992 in the USA. The oldest carrier of this mutation is believed to be the Osborndale Ivanhoe Friesian Holstein bull born in 1952 [Powell et al. 1996]. In the Polish population of Black-and-White cattle, the main lines that are known to carry the mutation were derived from three famous bulls, the sons of the aforementioned Osborndale Ivanhoe: Penstate Ivanhoe Star, Prowin Jewel and Paclamar Ivanhoe. In Poland, the most well-known sires used for breeding are Puget Sound Sheik, Hannoverhill Stardom and Constantijn, that are descendants of the Prowin Jewel bull line. The genetic information carried by these bulls have been introduced to Polish population mainly through the import of semen [Osten-Sacken 2004].

DUMPS

DUMPS is one of the earliest detected genetic defects in cattle. It is an autosomal, recessive, and embryonic lethal mutation. This disease is an impairment of the production of uridine monophosphate synthase (UMPS) – the enzyme responsible for converting orotic acid into uridine monophosphate (UMP), which is an essential component of pyrimidine nucleotides. Due to the fact that nucleotides are needed in significant amounts during embryonic development, the occurrence of this mutation (recessive homozygote) causes the death of the embryo at an early stage of development.

One of the first studies that brought the topic to light was carried out in the USA. At the University of Illinois a nutritional study was conducted, during which the level of orotic acid in the milk of cows was determined. Some cows were characterized by visible high levels of this acid, leading scientists to believe that the cause of this phenomenon was a low enzyme activity caused by the deficiency in UMP synthase. Different biochemical tests showed that these cows had only 50% of the normal activity synthase uridine monophosphate [Shanks and Robinson 1990]. Deficiency of Uridine Monophosphate Synthase is a disorder of particular interest in Holstein cattle because it is one of the few diseases that causes early embryo mortality. Embryos homozygous for the mutation die around the 40th day of pregnancy during implantation in the uterus [Ghanem et al. 2006].

The vital role of uridine monophosphate synthase in the formation of DNA and RNA has generated interest in the gene encoding this enzyme [Kaminski et al. 2005]. The gene for UMPS is located in the middle of BTA1 (q31-36) and is built of 1 869 base pairs (Table 1). Schwenger et al. [1993] identified the molecular basis of DUMPS, as an effect of a point mutation in the 5th exon of UMPS gene in codon 405. Substitution of cytosine (C) with thymine (T) in the CGA codon (normally coding arginine), creates STOP codon (TGA) [Meydan et al. 2010]. The consequence of this substitution is the premature end of protein translation, which in turn results in a loss of uridine monophosphate synthase activity.

In the USA, testing cattle for DUMPS was officially introduced in 1988. One of the main carriers of the DUMPS mutation was the elite bull Skokie Sensation Ned, born in 1957 [Schwenger et al. 1993]. In recent years, the most important carrier of the DUMPS mutation

Table 1. Basic characteristics of BLAD and DUMPS genetic disorders

Tabela 1. Podstawowe informacje o defektach genetycznych BLAD i DUMPS

Disorder name Nazwa defektu	Number in OMNIA Numer w OMNIA	Species-specific name Nazwa dla gatunku	Gene Gen	Location of the mutation Lokalizacja mutacji		References Literatura
				chromosome chromosom	position pozycja	
BLAD	000595	Bovine Leukocyte Adhesion Deficiency niedobór leukocytalnych cząsteczek adhezyjnych	ITGB2	1	145119004	Kaminski and Rusc [2017]
DUMPS	000262	Deficiency of Uridine Monophosphate Synthase niedobór syntazy monofosforanu urydyny	UMPS	1	69757801	Kaminski and Rusc [2017]

was the American bull called Happy Herd Beautician and it was through him that this anomaly spread worldwide.

In the past there was a concern that DUMPS carriers would appear in the Polish dairy cattle population. In Poland, the first DNA tests used to detect DUMPS carriers were introduced in 1995. Since 1999 testing of Polish population of Holstein (Black-and-White, Red-and-White) dairy breeding bulls is mandatory [Kaminski et al. 2005].

The aim of the present study was to estimate the frequencies of two hereditary disorders (BLAD and DUMPS) genotypes in the Polish population of Holstein-Friesian cattle.

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 Euro Genomics cooperative uses customized EuroGenomics arrays in the version called Eurogenomics MD_POL. Material for genotyping (5732 samples) was collected in the period from 2019 to 2020.

Sampling and transfer. Biopsies samples (ear punch) were collected with the use AllFlex Tissue Sampling Unit (TSU). Samples were kept in room temperatures and imminently transferred to the laboratory by postal services (2–3 days). In the laboratory, samples were coded and frozen in –20°C.

DNA extraction. After unfreezing samples were transferred to 96 well plates, lysis buffer and proteinase K were added. This mix was incubated in 56°C while mixing (600 RPM) on thermomixer overnight. Before the next step, samples were centrifuged (60 s × 3200 g) to remove undigested parts. Lysate was later transferred to deep-well plates and DNA extraction was processed with the use of Clean Blood & Tissue DNA Kit (CleanNA) according to producer manual in KingFisher DUO DNA processor (Thermo Scientific).

Quantification and normalization. DNA concentration was measured according to Illumina protocol (Infinium HD) by the fluorimetric method with the use of Quant-iT PicoGreen dsDNA Assay Kit (Thermo Scientific) in the Fluoroscan (Thermo Scientific) system. Basing on data from Quant-iT protocol, samples were normalized to 70 ng/ul by dilution with the use of TE buffer (Novazym).

Array processing. Normalized samples were processed according Illumina HTS protocol (manual protocol). Beadchips were immediately scanned on Illumina iScan system, scans were analysed using GenomeStudio Software V2011.1 version 1.9. All samples from one type of array were clusterized again together before export with the use of a specific cluster file, created from all samples. Statistical analysis was performed only on data with the call rate over 0.95. The study used probes named: UMPS (IlluminId: UMPS-1_T_R_2276841282) and ITGB2 (IlluminId: ITGB2-1_B_F_2276841212).

RESULTS

During the study, 5 732 cows of the Polish Holstein breed were examined. No sick animals nor carriers were found for DUMPS, 100% of the tested population was homozygous for the wild type genotype (Table 2). However, the study revealed the present of the munition in the ITGB2 gene. While no sick animals were identified, 0.21% of all animals were identified as carriers of the mutation. Most of the tested population (99.79%) was healthy (Table 3).

Table 2. The frequency of DUMPS genotypes and alleles in the examined Polish Holstein-Friesian cattle population

Tabela 2. Częstotliwość genotypów i alleli DUMPS w badanej polskiej populacji bydła holsztyńsko-fryzyjskiego

Genotype, n Genotyp, n			Healthy animals, % Zwierzęta zdrowe, %	Carriers, % Nosiciele, %	Sick animals, % Zwierzęta chore, %	Allele frequency Częstotliwość alleli	
TT	TC	CC				T	C
0	0	5 732	100	0	0	0	1

Table 3. The frequency of BLAD genotypes and alleles in the examined Polish Holstein-Friesian cattle population

Tabela 3. Częstotliwość genotypów i alleli BLAD w badanej polskiej populacji bydła holsztyńsko-fryzyjskiego

Genotype, n Genotyp, n			Healthy animals, % Zwierzęta zdrowe, %	Carriers, % Nosiciele, %	Sick animals, % Zwierzęta chore, %	Allele frequency Częstotliwość alleli	
AA	AG	GG				A	G
5 720	12	0	99.79	0.21	0	0.999	0.001

DISCUSSION

Bovine leukocyte adhesion deficiency (BLAD) carriers in Holstein cattle populations have been reported in many countries such as USA, Germany, France, Poland, Brazil, Japan, Iran, Turkey and India. In the 1990s, BLAD was one of the most widespread genetic diseases. The genetic material derived from these bulls was introduced to Polish population primarily through the import of semen and also as a result of the import of heifer calves in the early 1970s from the USA and Canada [Osten-Sacken 2004]. In 2007, Czarnik et al. [2007] carried out research to determine the frequency of BLAD carriers. The study was carried out in Poland between 1995 and 2006 and included 4 645 young breeding bulls. Due to the implementation of the BLAD control program (1995–1997), a clear decrease in the frequency of BLAD carriers was noted in Poland. The highest frequency of mutant allele carriers was noted at the beginning of this program when it amounted to 7.9%. Regular population surveys have significantly reduced the risk of this disease. Today only sporadic cases of BLAD carriers are reported. In the years 2004–2006 the number of heterozygotes dropped to around 0.8% [Czarnik et al. 2007]. In our study, the frequency of BLAD carriers was calculated to be at the level of 0.21% (the frequency of mutant BLAD allele: 0.1%) which suggest further decrease. The results of the present study are similar to those obtain in other countries such as: China (0.48%) [Li et al. 2011] and Turkey (1.31%, 2%) [Hacihasanoglu and Yardibi 2019, Korkmaz Agaoglu et al. 2015] (Table 4). In Mexico [Riojas-Valdes et al. 2009, Virgen-Méndez et al. 2019], Czech Republic [Citek et al. 2006], Russia [Koshchaev et al. 2018], and India [Debanath et al. 2016] researchers found no BLAD carrier in the studied populations.

The frequency observed in the present study (0.21%) is lower than the values described in studies conducted in some countries, for instance in Iran, where the frequency of heterozygotes was evaluated to be at the level of 3.33% [Norouzy et al. 2005], India with 3.23% of animal being carriers [Patel et al. 2007] and Turkey with 2.2% of carriers [Akyüz and Ertuğrul 2006].

No DUMPS carriers were detected in the present study. This is in agreement with the results presented by

Table 4. Frequencies of BLAD alleles and genotypes in different countries

Tabela 4. Częstotliwość alleli i genotypów BLAD w różnych krajach

Breed Rasa	n	Genotype Genotyp			Allele frequency Częstotliwość alleli		Carriers, % Nosiciele, %	Country Kraj	References Literatura
		AA	AG	GG	A	G			
Holstein	300	296	4	0	0.9933	0.0067	1.31	Turkey	Hacihasanoglu and Yardibi [2019]
Holstein	500	490	10	0	0.99	0.01	2	Turkey	Korkmaz Agaoglu et al. [2015]
Holstein	408	408	0	0	1	0	0	Mexico	Virgen-Méndez et al. [2019]
Holstein	61	61	0	0	1	0	0	Mexico	Riojas-Valdes et al. [2009]
Holstein	50	50	0	0	1	0	0	India	Debnath et al. [2016]
Holstein	73	73	0	0	1	0	0	Russia	Koshchaev et al. [2018]
Holstein	615	612	3	0	0.9976	0.0024	0.48	China	Li et al. [2011]
Holstein	406	406	0	0	1	0	0	Czech Republic	Citek et al. [2006]

Table 5. Frequencies of DUMPS alleles and genotypes in different countries

Tabela 5. Częstotliwość alleli i genotypów DUMPS w różnych krajach

Breed Rasa	n	Genotype Genotyp			Allele frequency Częstotliwość alleli		Country Kraj	References Literatura
		TT	TC	CC	T	C		
Holstein	50	0	0	50	0	1	India	Debnath et al. [2016]
Holstein	642	0	0	642	0	1	India	Patel et al. [2006]
Holstein	500	0	0	500	0	1	Turkey	Korkmaz Agaoglu et al. [2015]
Holstein	170	0	0	170	0	1	Turkey	Oner et al. [2010]
Holstein	73	0	0	73	0	1	Russia	Koshchaev et al. [2018]
Holstein	406	0	0	406	0	1	Czech Republic	Citek et al. [2006]

Kaminski et al. [2005], who studied a population of 2 209 bulls and found no carriers of this mutation. 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. DUMPS carriers have only been reported in the USA 1.2% (1990) and Argentine for 0.96% of bulls and 0.11% of tested cows (1996) [Avanus and Altinel 2017]. These results may suggest that years of appropriate breeding programs eliminated the mutant allel from the cattle population in many countries (Table 5).

Currently, BLAD and DUMPS attendance rates of Holstein-Fresian cattle in various countries around the world are being studied. The latest research shows that the frequency of carriers is lower and lower, and the number of sick animals is practically zero. In India, the frequency of BLAD carriers was found to be 4% [Ignetious et al. 2020] and in the West Java Province in Indonesia population was found to be free from sick animals and BLAD carriers [Nasrulloh et al. 2020]. In India also was found no animal carrier for DUMPS . The genotype frequency of normal individuals and the gene frequency of normal allele were found to be one [Ignetious et al. 2017].

CONCLUSIONS

The results of the present study demonstrate that carriers of BLAD mutation are present in the Polish Holstein-Friesian population, although at a low frequency. No carriers of DUMS mutation were detected. Having analysed results accumulated through many years one may conclude that molecular genetic methods significantly help to control the population and maintain the frequency of alleles responsible for genetic disorders in animal populations at least at a low level. Due to the regular use of genetic testing all around the world, the incidences of genetic disorders in the Polish population of Holstein-Friesian cows have decreased or have been completely eliminated.

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REFERENCES

Akyüz, B., Ertuğrul, O. (2006). Detection of bovine leukocyte adhesion deficiency (BLAD) in Turkish native and Holstein cattle. Acta. Vet. Hung., 54(2), 173–178. DOI: 10.1556/AVet.54.2006.2.4.

Avanus, K., Altinel, A. (2017). Inherited diseases of Holstein cattle: Story so far in Turkey. J. Istanbul Vet. Sci., 1(2), 40–46. DOI: 10.30704/http-www-jivs-net.324403.

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. DOI: 10.17221/5553-VETMED.

Czarnik, U., Grzybowski, G., Kaminski, S., Prusak, B., Zabolewicz, T. (2007). Effectiveness of a program aimed at the elimination of BLAD-carrier bulls from Polish Holstein-Friesian cattle. J. Appl. Genet., 48, 375–377. DOI: 10.1007/BF03195235.

Daetwyler, H.D., Capitan, A., Pausch, H., Stothard, P., van Binsbergen, R., Brøndum, R.F., Liao, X., Djari, A., Rodriguez, S.C., Grohs, C., Esquerré, D., Bouchez, O., Rossignol, M.N. et al. (2014). Whole-genome sequencing of 234 bulls facilitates mapping of monogenic and complex traits in cattle. Nat. Genet., 46, 858–65. DOI: 10.1038/ng.3034.

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.

Ghanem, M.E., Nakao, T., Nishibori, M. (2006). Deficiency of uridine monophosphate synthase (DUMPS) and X-chromosome deletion in fetal mummification in cattle. Anim. Reprod. Sci., 91, 45–54. DOI: 10.1016/j.anireprosci.2005.03.008.

Hacihasanoglu, C., Yardibi, H. (2019). Detection of allele and genotype frequencies of bovine leukocyte adhesion deficiency, factor XI deficiency and complex vertebral malformation disease genes in Holstein cattle. Ankara Üniv. Vet. Fak. Derg, 66, 311–315. DOI: 10.33988/auvfd.436199.

Ignetious, S., Joshi, S., Aich, R., Macwan, S. (2017). Genetic screening of deficiency of uridine monophosphate synthase in Holstein Freisian crossbred cattle. Ind. J. Vet. Sci. Biotech., 13(2), 50–53. DOI: 10.21887/ijvsbt.v13i02.10051.

Ignetious, S., Joshi, S., Aich, R., Macwan, S. (2020). Genetic studies on bovine leukocyte adhesion deficiency in Holstein Friesian crossbred cattle. J. Entomol. Zool. Stud., 8(3), 1656–1659.

Kaminski, S., Rusc, A. (2017). Haplotypy obniżające płodność Bydła holsztyńsko-fryzyjskiego [Haplotypes deteriorating fertility of Holstein-Friesian cattle]. Prz. Hod., 1, 6–8. [in Polish].

Kaminski, S., Grzybowski, G., Prusak, B., Rusc, A. (2005). No incidence of DUMPS carriers in Polish dairy cattle. J. Appl. Genet., 46, 395–7.

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. DOI: 10.3906/vet-1412-13.

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.

Li, J., Wang, H., Zhang, Y., Hou, M., Zhong, J., Zhang, Y. (2011). Identification of BLAD and citrullinemia carriers in Chinese Holstein cattle. Anim. Sci. Pap. Rep., 29(1), 37–42.

Meydan, H., Yildiz, M.A., Agerholm, J.S. (2010). Screening for bovine leukocyte adhesion deficiency, deficiency of uridine monophosphate synthase, complex vertebral malformation, bovine citrullinaemia, and factor XI deficiency in Holstein cows reared in Turkey. Acta Vet. Scand., 52, 56. DOI: 10.1186/1751-0147-52-56.

Nagahata, H. (2004). Bovine leukocyte adhesion deficiency (BLAD): a review. J. Vet. Med. Sci., 66, 1475–82. DOI: 10.1292/jvms.66.1475.

Nasrulloh, M.F., Wulandari, A.S., Indriawati, I., Margawati, E.T., Volkandari, S.D. (2020). Detection of the CD18 gene mutation as a marker of BLAD genetic disorder of Holstein-Friesian cattle in West Java. Livest. Anim. Res.,18(2), 116–123. DOI: 10.20961/lar.v18i2.42933.

Norouzy, A., Nassiry, M.R., Shahrody, F.E., Javadmanesh, A., Abadi, M.R.M., Sulimova, G.E. (2005). Identification of bovine leucocyte adhesion deficiency (BLAD) carriers in Holstein and Brown Swiss AI bulls in Iran. Genetica, 41(12), 1697–1701. DOI: 10.1007/s11177-006-0014-7.

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. DOI: 10.3923/ajava.2010.60.65.

Osten-Sacken, A. (2004). Defekty genetyczne u bydła [Genetic disorders in cattle]. Prz. Hod., 5, 9–12. [in Polish].

Patel, R.K., Singh, K.M., Soni, K.J., Chauhan, J.B., Sambasiva Rao, K.R.S. (2007). Low incidence of bovine leukocyte adhesion deficiency (BLAD) carriers in Indian cattle and buffalo breeds. J. App. Genet., 48(2), 153–155. DOI: 10.1007/BF03194673.

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. DOI: 10.1007/BF03194629.

Powell, R.L., Norman, H.D., Cowan, C.M. (1996). Relationship of bovine leukocyte adhesion deficiency with genetic merit for performance traits. J. Dairy Sci., 79, 895–899. DOI: 10.3168/jds.S0022-0302(96)76438-X.

Riojas-Valdes, V.M., Carballo-Garcia, B., Rodriguez-Tovar, L.E., Garza-Zermeno, M.V., Ramirez-Romero, R., Zarate-Ramos, J., Avalos-Ramirez, R., Davalos-Aranda, G. (2009). Absence of bovine leukocyte adhesion deficiency (BLAD) in Holstein cattle from Mexico. J. Anim. Vet. Adv., 8(9), 1870–1872.

Schwenger, B., Schober, S., Simon, D. (1993). DUMPS Cattle Carry a Point Mutation in the Uridine Monophosphate Synthase Gene. Genomics, 16, 241–244. DOI: 10.1006/geno.1993.1165.

Shanks, R.D., Robinson, J.L. (1990). Deficiency of Uridine Monophosphate Synthase Among Holstein Cattle. Cornell Vet., 80, 119–122.

Shuster, D.E., Bosworth, B.T., Kehrli, M.E. (1992). Sequence of the Bovine CD18-Encoding cDNA – Comparison with the Human and Murine Glycoproteins. Gene, 114, 267–271. DOI: 10.1016/0378-1119(92)90586-E.

VanRaden, P.M., Olson, K.M., Null, D.J., Hutchison, J.L. (2011). Harmful recessive effects on fertility detected by absence of homozygous haplotypes. J. Dairy Sci., 94,6153-61. DOI: 10.3168/jds.2011-4624.

Virgen-Méndez, A., Ayala-Valdovinos, M.A., Galindo-García, J., Sánchez-Chiprés, D.R., Lemus-Flores, C., Duifhuis-Rivera, T. (2019). Carrier frequency of autosomal recessive disorders (BC, BLAD, FXID and CVM) in Holstein cows in Jalisco, Mexico. Pesq. Vet. Bras., 39(7), 481–484. DOI: 10.1590/1678-5150-pvb-6202.

Gozdek, M., Kolenda, M., Kamola, D., Sitkowska, B. (2020). Report on the incidence of hereditary disorders (BLAD, DUMPS) in the Polish population of Holstein-Friesian cattle. Acta Sci. Pol. Zootechnica, 19(3), 15–22. DOI: 10.21005/asp.2020.19.3.02

WYSTĘPOWANIE CHORÓB GENETYCZNYCH (BLAD, DUMPS) W POLSKIEJ POPULACJI BYDŁA RASY HOLSZTYŃSKO-FRYZYJSKIEJ

STRESZCZENIE

BLAD (niedobór leukocytalnych cząsteczek adhezyjnych) oraz DUMPS (niedobór syntazy monofosforanu urydyny) są dziedzicznymi recesywnymi chorobami autosomalnymi, które mogą wpływać na zwiększenie śmiertelności w populacji bydła. Celem niniejszej pracy była ocena częstości genotypów BLAD i DUMPS w polskiej populacji krów mlecznych. W badaniu uwzględniono 5732 krowy rasy polskiej holsztyńsko-fryzyjskiej. Wyniki wskazały, że polska populacja bydła rasy holsztyńsko-fryzyjskiej jest wolna od recesywnej mutacji DUMPS, ponieważ nie wykryto nosicieli oraz zwierząt chorych. W przypadku choroby BLAD wykazano niską obecność nosicieli w populacji (0.21%). Zarówno BLAD, jak i DUMPS mogą silnie wpłynąć na rentowność gospodarstw mlecznych, dlatego w celu uniknięcia strat ekonomicznych, spowodowanych obecnością tych chorób genetycznych w stadzie, należy regularnie monitorować populację w celu szybkiego wykrycia nosicieli.

Słowa kluczowe: BLAD, DUMPS, bydło, frekwencja genów, mikromacierze

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