

HEALTH IN CONTEMPORARY CATTLE BREEDING

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ABSTRACT

The paper deals with the genetic health of cattle. The studies done in the Czech cattle population are summarized. The strict measures taken for BLAD in the 90's have been efficient, as the frequency of positive sires has decreased rapidly. The diverse approach to Complex Vertebral Malformation (CVM) resulted in different population dynamics. The use of proven heterozygous carriers of lethal alleles in breeding is not absolutely banned, so the rate of heterozygous sires decreases, but persists still considerably, and the occurrence of CVM females remains high. Recessive alleles of bovine citrullinaemia, DUMPS, glycogen storage disease V and II, and factor XI deficiency were not found. The occurrence of congenital defects was analysed. The XX/XY chimerism was found in 0.50 % of Holstein-Friesian sires and in 0.74 % of Czech Simmental sires. By testing for aneuploidy, 5 Charolais sires were found to be carriers of a redundant sex chromosome (61, XXY; 61, XXX). Autosomal aneuploidies were not found. Robertsonian translocations were occurred in 0.82 % of Czech Simmental sires and in 3.57 % of beef sires, whilst Holsteins were not affected. The estimate of genetic variance of stillbirth caused by sires was 1.90 %, and heritability - 0.078, but the sizeable influence of sire's line was observed. Despite of the low genetic variance, it is recommended that sires with higher incidence of fertility damages should not be used for breeding. In the period of genomics, molecular biology and molecular cytogenetics, the opportunity arises to solve the problem of genetic health principally.

Key words: cattle; health; congenital; chromosomal abnormalities

INTRODUCTION

Fundamentally, the core of the problem is to find the animals that genetically connect high production and good health. The issue is rather intricated, and needs systemic approach.

Firstly, there is some contradiction between breeding for high performance and general state of health. The trait named as fitness, adaptability, and constitution has complex character, and polygene genetic background. It is the main criterion in the natural selection, when the heterozygous animals are preferred. On the contrary, the artificial selection on particular performance traits is mostly directional, homozygous animals are preferred. Thus, it results in lower adaptability, especially when in the breeding process the unsatisfactory attention to health is paid. Then the pressure on the high performance can be resulted in the worse health.

Secondly, the genetics is not clarified sufficiently. In many cases of congenital defects, e.g. even the detection of genetic or environmental aetiology is difficult. The traits with quantitative character, such as reproduction, have low heritability, some characteristics are difficult measurable.

However, in the age of genomics, molecular biology and molecular cytogenetics, the opportunity arises to solve the problem principally.

The paper summarizes the studies done on the Czech cattle population, and focused on the various aspects of the genetic health.

RESULTS AND DISCUSSION

Recessively inherited diseases

Relatively plain are the heritable diseases, lethal and sub-lethal factors caused by monogenic factors.

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The heredity is mostly recessive. In domestic animals, the occurrence of the defect is often breed-specific. In some cases, in different breeds different mutation in various regions of the same gene causes the defect and myostatin could be given as an example. The origin of the breeds is the reason, when they are bred closely over a long period.

Recently, Bovine Leukocyte Adhesion Deficiency and Complex Vertebral Malformation (CVM) in Holstein cattle have become a serious threat. The role of widely used superior breeding males in spreading of the undesirable recessives was described repeatedly. Here, different dealing with a problem of recessive inherited diseases is demonstrated.

In the Czech Republic, the strict measures for BLAD were taken in the 90's. Every Holstein sire had to be genotyped, and the mating of heterozygous carriers has been absolutely prohibited. During the period of 1993-1998, the occurrence of heterozygous animals in Czech Holsteins exceeded 10 %, whilst in other countries, similar or even higher incidence has been found (Shuster *et al.*, 1992; Jorgensen *et al.*, 1993). Obviously, the measures have been efficient, as the frequency of positive sires has decreased rapidly (Tab. 1), the high occurrence of positive heifers in 2002-2004 was detected by testing females suspected because of a heterozygous parent. More recently positive findings in males are sporadic, but continuous testing is necessary, because in commercial herds, the eradication process is not short-term, as the prevalence of heterozygous animals depreciates by only 50 % each generation. The mating of heterozygous sires in the population may still result in births of affected calves.

A worldwide increase of Complex Vertebral Malformation (CVM) in the Holstein population has been recorded since 2001. The Czech Holstein could not differ of course (Table 1), as the rate of imported sires is approximately 80 %, mainly from the United States, Canada, Germany and the Netherlands. Restrictive measures have been undertaken here too. The CVM status of every sire must be declared, but the use of proven heterozygous carriers of lethal alleles in breeding is not banned, if the breeder agrees. The rate of heterozygous sires decreased, but still persists considerably. The occurrence of CV females remains higher, and similarly in another analysis (Čítek *et al.*, 2006), the frequency of heterozygous females was 19 %. Interestingly, no case of heterozygosity for BLAD and CVM in any individual has been recorded. On the other hand, the probability of contemporary presence is not high taking into account the low number of animals positively genotyped for BLAD since 2001 (n = 14).

Obviously, the diverse approach to the diseases mentioned results in different population dynamics. Of

course, the controlled mating of proven non-carriers on the daughters of heterozygous sires can eliminate the birth of affected calves, but the more thoroughgoing breeding would be salutary.

The incidence of other recessives is markedly lower. This we can confirm by our analyses of bovine citrullinaemia, deficiency of uridine-5'-monophosphate synthase (DUMPS), glycogen storage disease V (deficiency of muscle glycogen phosphorylase, or myophosphorylase, *PYGM gene*), glycogen storage disease type II (GSD II, generalised glycogenosis, GAA gene, exons 7, 9, and 13), and factor XI deficiency (exons 9 and 12), the recessive alleles were not revealed.

Congenital defects

The congenital defects do not occur often. Involving both hereditarily caused defects and malformations suffered during pregnancy, the definition is rather vague from the genetic point of view. The aetiology of the latter is too often unclear, as many teratogens can be involved. Even the discrimination between acquired and inherited defects can be troublesome, as specific and thorough knowledge is sometimes, regrettably, missing.

Some of the afflictions caused by changes in genetic matter occur only rarely, and a thorough analysis is therefore impossible. However, some, particularly those with a relatively frequent incidence, have been quite well explored. Even in such cases the genetic background can be uncertain, so it is sometimes doubtful whether the condition is monogenic or polygenic, and many disorders have been assigned as definitely caused by one-gene on the basis of data insufficient for such certainty. Similarly, a resolution in favour of dominant or recessive inheritance can be dubious. Hence, because of the number and range of factors in operation in dealing with congenital disorders, any interpretation of the data must be prudent.

Congenital disorders have been a subject to analysis by many authors over the past decades from as early as the 1920s, and there are a number of excellent reviews which summarise their work. A comprehensive list of single locus traits is maintained by the Online Mendelian Inheritance in Animals (OMIA) database and is accessible on the internet.

In a survey done in the Czech cattle, around 8.6 % of Holstein sires and 6.5 % of Czech Simmental sires have born the offspring with congenital disorder (Čítek *et al.*, 2009a).

Summarizing the results, only 18 congenital disorders occurred in the progeny of 10 and more sires, sc. hernia umbilical cong., BLAD, opened hock, contracture of muscles and ligaments of limbs, schistosomus reflexus, stillborn calves, hydrocephalus

congenital, unviable calves, Robertsonian translocation, brachygnathia inferior, atresia ani and recti, ascites, perishing of calves, brachygnathia superior, abortion, spastic paresis, hernia cerebialis, and dystocia. A further 12 disorders affected the progeny of 5 to 9 sires, i.e. different types of atresia, aplasia, cleft, chimerism etc. The remainder of the congenital disorders identified, i.e. 121 (80 % of 151 described disorders) were diagnosed in the progeny of 4 and fewer sires; 88 disorders in the progeny of 1 sire only. Thus, the problem of congenital disorders is rather diversiform and difficult to analyse.

Of particular defects, umbilical hernia was the disorder which occurred most frequently. In literature (Herrmann *et al.*, 2001), the influence of the sire and sire line is reported to be significant. Afflictions of limbs were considerably frequent, namely opened hock, and contractures of muscles and ligaments. Quite often schistosomus reflexus and various defects of the head were present. In our analysis, we found only sporadic cases of genital fissures.

Hydrops was found to be relatively common and also hydrocephalus, which has already been referred to as being among the most frequent cranial afflictions. Of defects in female genitals, aplasia was the most common. Male genitals were affected only rarely, cryptorchidism for example, appeared only once.

Even though the genetic analysis of rarely occurring disorders is difficult, sires fathering affected offspring should not be widely used in artificial insemination programmes, but disqualified for fathering of stock bulls or even definitively culled. In this context, it was unfortunate decision, when the control

over the congenital diseases in the Czech Republic was abolished.

Chromosomal abnormalities

Aneuploidy and chimerism can occur more or less frequently, and for this reason cytogenetic analysis was applied in the Czech Republic by the veterinary survey.

The XX/XY chimerism was found in 0.50 % of Holstein-Friesian sires, and in 0.74 % of Czech Simmental sires (Čítek *et al.*, 2009b). By testing for aneuploidy, 3 Charolais sires and 2 heifers were found to be carriers of a redundant sex chromosome (61, XXY; 61, XXX). Autosomal aneuploidies were not found. This does not mean they do not occur. In the group of animals in the analysis, autosomal aneuploidies were evidently lethal early in gestation, and the less harmful gonosomal aneuploidies allowed survival.

The search for Robertsonian translocations showed a relative frequency of 0.82 % in Czech Simmental sires and 3.57 % in beef sires. The Holstein-Friesian sires and heifers were not affected. Our results are well consistent with other reports, e.g. Seguin *et al.* (2000), who did not find any centric fusion in American Holstein-Friesian sires, but did find chimeric animals. Also, Nel *et al.* (1991) note the lack of chromosomal aberrations in pure dairy breeds found by teams across the world, when the chimerism found in Holstein-Friesians cannot be considered to be a primary chromosome defect.

As the chimeric sires have reportedly decreased reproductive performance, and the presence of Robertsonian translocations has been confirmed, the early

Table 1: BLAD and CVM heterozygotes in the Czech Republic 1993-2005 (Čítek *et al.*, 2008)

Year of testing	BLAD				CVM			
	sires		heifers		sires		heifers	
	tested	positive	tested	positive	tested	positive	tested	positive
2005	101	1	8	0	85	7	20	4
2004	161	3	10	2	72	6	13	5
2003	153	0	9	2	108	12	13	5
2002	89	2	9	2	35	7	3	0
2001	83*	2	-	0	10*	2	-	0
2000	69*	1	-	3		not tested		
1999	14	0	2	0		not tested		
1993-98	568	79	121	13		not tested		

* number of tested sires and heifers together

cytogenetic screening of sires seems to be advisable; but again, the systematic examination was abolished.

Polygenic health traits

The field reveals more sub-traits, and it is hard to say, which is more important or should be preferred. In the Czech Republic, the breeding values for the own sire's fertility are assessed, for the fertility of its daughters, the breeding value for the delivery difficultness, and for the longevity. In a broad term, also the body formation could be considered to be an indicator of health.

As mentioned in the introduction, the heritability of fertility is low. In our analysis e.g., the population genetic analysis resulted in the estimate of genetic variance of stillbirth caused by sires as $\sigma_s^2 = 1.90\%$. Then the calculated value of stillbirth heritability was $h^2 = 0.078$ (Čítek *et al.*, 2011). However, in our other analyses, the sizeable influence of sire's line was found. Despite of the low genetic variance, it is recommended that sires with a higher incidence of fertility damages should not be used for breeding.

Breeding for the easy delivery must be carried out advisedly, because birth mass and growth capacity correlate positively, and focusing on the course of the birth results in a reduced body mass. In such cases, genome analysis may be promising. Though many analyses have been carried out already on calving difficulty, stillbirth and birth weight, further extensive studies for the assessment of their reasons, and quantification of genetic parameters are necessary. Recently, quantitative trait loci affecting the traits mentioned have been established with promising results. The QTL affecting dystocia, conformation, and economic merit were described as being related to calf size or birth weight. Grosz and Mac Neil (2001) revealed the QTL influencing the birth weight with no significant effect on growth since birth to weaning. Therefore, potentially the incidence and degree of dystocia can be reduced without compromise of subsequent growth performance. The microarray approach is promising also in other health indicators with polygenic heredity.

CONCLUSION

The occurrence of inherited disorders is sporadic. However, a massive spread in farm animals could have a serious affect on the economy. Identification of the molecular basis for these diseases enables the quick recovery of the population. Permanent attention must be paid to the congenital defects and to the health indicators with polygenic heredity.

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