Congenital disorders in the cattle population of the Czech Republic

J. Čítek[1], V. Řehout[1], J. Hájková[2]

[1]Department of Genetics, Animal Breeding and Nutrition, Faculty of Agriculture, University of South Bohemia, České Budějovice, Czech Republic
[2]Veterinary Research Institute, Brno, Czech Republic

ABSTRACT: The aim of the paper was to analyse congenital disorders in the Czech cattle population in 1986–2001. The offspring of 474 sires – 215 Czech Simmental, 236 Holstein, and 23 beef – were diagnosed with congenital disorders which were unevenly distributed because only 18 occurred in the progeny of 10 and more sires, in contrast to 88 occurring in the progeny of 1 sire only. Umbilical hernia was the most frequently noted disorder, and 136 sires fathered progeny with limb anomalies. The most frequent gestational accident was schistosomus reflexus, the results suggesting a familial burden. Three sires fathering offspring with the afflicted spinal column and limbs were heterozygous for Complex Vertebral Malformation (CVM) though they had not been reported as such. Foetal defects and stillbirth were quite frequent, and the calves affected were fathered by 56 sires. In rare disorders with a low incidence, an accurate genetic analysis or even simple discrimination between inherited and acquired defects is problematic. It would be our recommendation that those sires with a higher incidence of defects among their offspring should be disqualified from fathering stock bulls, or culled.

Keywords: cattle; health; genetics; congenital defect; inherited disorder; stillbirth

Congenital disorders do not occur frequently. Involving both hereditarily caused defects and malformations suffered during pregnancy, the definition is rather vague from the genetics aspect. The aetiology of the latter is very 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 not therefore possible. 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 subjected to analysis by many authors over the past decades since as early as the 1920s, and there are a number of excellent reviews which summarise their work: Herzog (1992), Agerholm et al. (1993), Ladds (1993), Saperstein (1993), Toombs et al. (1994), Wikse et al. (1994), Healy (1996), Grubbs and Olchowy (1997), Kuhn (1997), and others are excellent sources of information. A comprehensive list of single locus traits is maintained by the Online Mendelian

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Inheritance in Animals (OMIA) database and is accessible on the internet (Nicholas, 2003).

Here, the results of the long-term study are presented, conducted to analyse the congenital disorders in the Czech Simmental, Holstein, and beef cattle in 1986–2001.

MATERIAL AND METHODS

In the Czech Republic, a surveillance programme has been established to check the genetic health of cattle, whereby data on calves with congenital disorders are collected and evaluated. Field veterinarians report the birth of disabled calves, note their diagnosis, and identify the sire and the dam. The checking of approximately one hundred progeny is considered to be sufficient for the evaluation of a sire. In this paper, sires born between 1986 and 2001 were analysed.

The recessive hereditary disorders BLAD and CVM were diagnosed by molecular analysis; CVM according to the patent Genetic Test for the Identification of Carriers of CVM, and BLAD according to the methods of Nagahata et al. (1993).

Robertsonian translocations were diagnosed by conventional cytogenetic investigation. We used a modified method of investigation of live dividing lymphocytes from peripheral blood (Moorhead et al., 1960). At least 20 mitoses per animal were evaluated.

RESULTS AND DISCUSSION

In total, the offspring of 474 sires were diagnosed as being afflicted with a congenital disorder, namely 215 sires of Czech Pied cattle (Czech Simmental), 236 Holstein sires incl. Red Holstein, 1 Hereford, 1 Aberdeen Angus, 2 Belgian Blue, 6 Limousine, 9 Charolais, 3 Piemontese, and 1 Blonde d'Aquitaine (i.e. 23 beef sires). When more than one disorder occurred in the progeny of a sire, each disorder was counted individually and summarized. For example one of the evaluated sires had progeny with micromelia, weak calves, abortion, and perishing of calves, so it has been counted four times. Therefore, the sum of sires with offspring carried disorders is 734, even though the actual number of sires is 474.

The numbers of sires recorded newly into the pedigree books of Czech Simmental and Holstein breeds in the Czech Republic are given in Table 1. As can be seen, around 8.6 per cent of Holstein sires and 6.5 per cent of Czech Simmental sires have born the offspring with congenital disorder.

Altogether, only 18 congenital disorders occurred in the progeny of 10 and more sires (Table 2): 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 cerebralis, and dystocia. Further 12 disorders affected the progeny of 5 to 9 sires, i.e. atresia ani, big foetus, acaudia, aplasia uteri, cheilognathopalatoschisis, palatoschisis, shortening of spinal column, aplasia of uteri and ovaries, cheiloschisis (gnathoschisis), aplasia of genitals, chimerism XX/XY, and shortening of limbs. 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.

Umbilical hernia was the disorder which occurred most frequently in the analysed period: 84 sires, i.e. 17.7% of 474 males with affected progeny fathered offspring with this disorder (Table 2). Herrmann et al. (2001) in German Fleckvieh found an overall incidence of 1.8%, and concluded that the sire and sire line had a significant influence. Even though they could not explain the segregation pattern either by an autosomal recessive or

Table 1. Sires recorded newly in the pedigree books in the Czech Republic in 1986–2001

	Total	Born in the Czech Republic		Imported	
		n	(%)	*n*	(%)
Holstein	2 740	603	22	2 137	78
Czech Simmental	3 307	2 811	85	496	15
Total	6 047	3 414	56	2 633	44

Table 2. Congenital and inherited disorders found in offspring of sires of the Czech cattle population

Group/sum of sires	Diagnosis	Number of sires with offspring carrying the disorder
Hernia/88	hernia umbilical cong.	84
	hernia abdominalis	4
Recessive inherit. disorders/68	BLAD	64
	CVM	4
Chromosomes/23	Robertsonian translocation 1/29	18
	chimerism XX/XY	5
Birth defect/40	schistosomus reflexus	35
	monster	1
	amorphus globosus	2
	haired formation in embryolemma	1
	heart ante apertura thoracis cranialis	1
Head/33	hernia cerebralis	10
	dicephalus	2
	duplicitas anterior, dicephalus	1
	pygopagus, dicephalus	1
	dicephalus bicollis	1
	dicephalus, spondyloschisis	1
	cranioschisis	1
	schisoprosopia	2
	enlargement of neurocranium	1
	short wide head	1
	short head	1
	cerebrocortical necrosis	1
	fissura cerebralis	1
	defect of head, 4 eyes, 2 lower jaws	1
	double muzzle	1
	missing nostrils	1
	double lower jaw	1
	anodontia	1
	defects of teeth	1
	hypoplasia and bilateral deformity of auricles	1
	double auricles, dermal formation in genit. region, male	1
	dermal formation on head	1
Eye/13	cataract lentis bilateralis	1
	anophthalmus	3
	anophthalmus bilateralis	1
	aplasia of left eye	1
	aplasia of right eye	1
	head without eyes, only orbits developed	1
	microphthalmia	3
	microphthalmia bilateralis	1
	blindness	1

Table 2. to be continued

Group/sum of sires	Diagnosis	Number of sires with offspring carrying the disorder
Jaw/35	brachygnathia inferior	17
	brachygnathia superior	12
	brachygnathia et cheiloschisis superior	1
	brachygnathia inferior and superior	2
	brachygnathia	1
	agnathia	2
Fissure of jaw or lip/20	cheilognathopalatoschisis	7
	palatoschisis	7
	cheiloschisis (gnathoschisis), ch. partial	6
Spine and pelvis/23	shortening of spinal column	7
	spondyloschisis	1
	defects of cervical spinal column	2
	defects of pectoral spinal column	1
	defects of spinal column	1
	schistothorax in sternum	1
	defects of lumbar spinal column	1
	spondyloschisis in loin region	1
	deformed spinal column and pelvis	1
	deformed pelvis	1
	hypoplasia of lumbar vertebrae	1
	diastematopyelia (cleft pelvis)	1
	hypoplasia pelvis	1
	cleft neurocanal	1
	kyphosis	1
	scoliosis	1
Perosomus elumbis/3	perosomus elumbis	2
	perosomus elumbis, susp.	1
Cauda/10	acaudia	8
	double cauda	1
	malformation of cauda	1
Abdomen/5	coeloschisis	3
	abdominal fissure	1
	aplasia of abdominal muscles	1
Anus/24	atresia ani and recti	15
	atresia ani	9
Limbs/136	opened hock	50
	contracture of muscles and ligaments of limbs	43
	achondroplasia susp.	3
	achondroplastic shortening of limbs	3
	achondroplasia I.	2
	achondroplasia II.	1
	shortening of limbs	5
	shortening of right forelimb and left hindlimb	1

Table 2. to be continued

Group/sum of sires	Diagnosis	Number of sires with offspring carrying the disorder
Limbs/136	shortening of forelimbs	1
	micromelia	2
	hypoplasia of pelvis, limbs and lumbar vertebrae	1
	aplasia of limbs of metatarsus	2
	hypoplasia of forelimbs, partial aplasia of right hindlimb	1
	flexion of forelimbs with ankylotic joints	1
	inflexed joints	2
	flexion of carpal joints	1
	anomaly of limbs	1
	malformation of spinal column and joints of limbs	1
	convoluted forelimbs	1
	anomaly of limbs with ankylosis	3
	ankylosis	3
	released inter-hoof ligament	3
	atrophy of muscles on left forelimb	1
	malformations of hindlimbs	1
	malformation of left forelimb	1
	defects of hoofs	1
	defect of hoofs' keratin	1
Polypodia/6	polypodia	2
	polypodia posterior	2
	polypodia anterior	2
Fingers/6	polydactyly	4
	adactyly of left forelimb	1
	adactyly of forelimbs	1
Paresis/20	spastic paresis	11
	spastic paresis of left hindlimb	4
	spastic paresis of right hindlimb	1
	paraplegia	1
	torticollis	3
Epitheliogen. imperf./3	epitheliogenesis imperfecta	3
Alopecia/1	alopecia	1
Hydrops/42	hydrocephalus congenital	23
	hydrocephalus internus	1
	hydrothorax	1
	ascites	15
	hydrops univ.	2
Female genitals/29	aplasia of genitals	6
	aplasia uteri and ovaries	7
	aplasia uteri	8
	hypoplasia uteri	1
	uterus unicornis	2
	cleft genitals (female)	1
	fissura retrovaginalis, anus vulvovaginalis	1

Table 2. to be continued

Group/sum of sires	Diagnosis	Number of sires with offspring carrying the disorder
Female genitals/29	hypertrophied hymen	1
	hypertrophied clitoris	1
	malformation of vulva	1
Male genitals/9	cryptorchidism	1
	hypoplasia of testes	1
	double scrotum	1
	defect of scrotum	1
	torsion of scrotum	1
	prolapsus of prepuce	1
	defect of sexual differentiation	1
	hermaphroditism of offspring	2
Foetus, calf/56	mummified foetus	4
	foetal death	1
	abortion	12
	stillborn calves	26
	prolonged gravidity, stillbirth	2
	birth mass of 8 to 10 kg	1
	big foetus	9
	thickening of embryolemma	1
Dystocia/10	dystocia	10
Unviable calves/32	unviable calves	19
	perishing of calves	13

by an autosomal dominant monogenic model, they recommended that the use of sires with an occurrence of hernia umbilical in their offspring should be restricted.

BLAD occurred frequently (n = 64) because the evaluation period included the 90's – a period with a generally high occurrence of this monogenic recessive lethal trait. The low incidence of CVM (n = 4) is not surprising, because our data collection stopped in the early 2000's, and hence its culmination was not recorded. The incidence of CVM, BLAD and other genetic disorders in the Czech cattle population has been dealt with in another paper (Čítek et al., 2006). Other authors have different findings but theirs could have been influenced by special circumstances in the population under analysis; for example Agerholm et al. (1993) reported spinal muscular atrophy as the most common disorder in the Danish cattle population, but all calves with the diagnosis belonged to the same families, of American Brown Swiss origin. This stock has not been used in breeding in the Czech Republic.

The Robertsonian translocation 1/29 was reported in the progeny of 18 sires. Czech Simmental sires were affected, but also beef sires, in particular one Blonde d'Aquitaine and one Charolais. Agerholm et al. (1993) reported the Robertsonian translocation in Blonde d'Aquitaine (165 normal, 55 heterozygous) and Limousine breeds (129 normal, 18 heterozygous). In our previous analysis (Čítek et al., 2006), the relative frequency was 0.99% of Simmental and 3.52% of the beef sires analysed. The translocation was found also in 13 females out of 48. In the Czech Republic, an approved prophylactic screening program for Robertsonian translocations has been in place for some years in order to prevent their spread and future fertility damage in the cattle population.

In Table 2, the disorders are grouped according to the type of the affliction or the body part afflicted.

Thus, 42 sires have produced monsters, 31 sires calves with various head defects etc.

Of these groups, afflictions of limbs were the most frequent; 136 sires in total had produced progeny with various anomalies; 50 sires (10.5% of those with affected offspring) had produced progeny with opened hock, and 43 sires progeny with contractures of muscles and ligaments. There has been a relatively high incidence of afflicted limbs in Czech cattle for some time: opened hock is the most persistent, while polydactyly has occurred in the progeny of only four sires, and syndactylism has not been reported at all.

As can be seen in Table 2, some of the listed disorders might have occurred in the same incident, and many have been linked already by the processing of the data. In some cases, the grouping was dubious, because the description of veterinarians was not precise enough. For example, sometimes the disorder was described only as "inflexed joints" or "polypodia", and sometimes more precisely as "flexion of carpal joints" or "polypodia posterior". However, the description was at times even more vague; for example "anomaly of limbs" or "defects of spinal column". This imprecision was caused by a number of factors: non-uniform diagnosis by veterinarians, varying degrees of the care taken, non-uniform terminology, and a further significant factor was the very length of time over which the data had been collected. The interpretation of congenital disorders was complicated by ambiguity, and a degree of unification of diagnostics and terminology would simplify the analysis. Fortunately, in most cases the diagnoses were obvious.

Schistosomus reflexus appeared relatively often; 35 sires had bred afflicted foetuses. This is a malformation with important clinical implications, is always a challenge for veterinarians and breeders, and is often baffling for less experienced obstetricians (Saperstein, 1993).

Our results suggest a familial burden for schistosomus reflexus, because, of the 35 sires, 20 belonged to three lines only. This fits with the hypothesis of autosomal recessive heredity suggested by other authors. Eleven sires had produced other defects along with schistosomus reflexus, but with broad variability. For example, one sire fathering a calf with schistosomus reflexus passed on atresia ani et recti, and also brachygnathia inferior and cryptorchidism to three calves. In another sire, amorphus globosus was reported in one calf, and schistosomus reflexus in the other. Nevertheless, the data are insufficient for reasonable speculation on the genetic connections.

Various congenital defects of the head were reported quite often. As can be seen in Table 2, they concerned the head itself, the eyes, brachygnathia, and schisis. The relatively high occurrence of duplications such as dicephalus, schisoprosopia, four eyes and two lower jaws, double muzzle, double lower jaw, and double auricles, is in concordance with Hiraga and Dennis (1993). They give congenital duplication as a relatively common defect in cattle, the incidence being lower in sheep and pigs, and rare in goats. Cranial duplications prevail in cattle, in contrast to caudal duplication in sheep and pigs. In our analysis we found only sporadic cases of genital fissures. The authors mentioned above regard the differences among species as interesting, and suggest that they may explain why congenital duplications appear more frequently in cattle than in other domestic animals, even though the cause is not known.

Agnathia was not accompanied by defects of auricles, as usually happens in man. One sire who produced agnathia had also bred descendants with flexions of limbs, muscular atrophy and epitheliogenesis imperfecta.

Brachygnathia occurred quite often, as 35 sires produced calves with the defect. Cheilognathopalatoschisis, cheiloschisis, and palatoschisis were also relatively frequent.

The occurrence of serious inherited eye diseases could recently be controlled in other species by genotyping of the respective gene (Bechyňová et al., 2008).

The occurrence of spinal column affliction in the period before CVM was identified prompts the question whether the defects are caused by this recessive inherited disorder or not. The suspect sires also produced progeny showing contractures of muscles, inflexed joints and paresis, and so we compared the list of sires with progeny showing these defects with the list of known carriers of the recessive allele for CVM. We also included the fathers of dams mothering the affected calves. Altogether, 57 sires were involved in the search, and only 19 were found in the register for CVM because the sires involved in the paper were old, and not all were used during the period when the test for CVM was a requirement. Of these, three were found to be carriers of the recessive allele, though in this research they were not reported originally as being CVM carriers. The first of these sires produced calves with contracture

of muscles and ligaments of the limbs, the second with the deformed spinal column and pelvis. The third produced progeny with contracture of muscles and ligaments of the limbs, shortened limbs and achondroplasia, and also calves with brachygnathia superior, aplasia of the left eye and hypoplasia uteri. With heterozygous dams, the sires could father the calves homozygous for CVM. Therefore, the reason for the defects mentioned (i.e. spinal column affliction, contractures of muscles, inflexed joints and paresis) in the 80's and 90's may have been CVM in some cases. Of course, the testing of numerous old sires by molecular methods is economically unimaginable and not necessary from the breeders' point of view; the testing of sires more recently used in breeding, and thus the elimination of heterozygous carriers is highly necessary.

Leipold et al. (1993) suspected perosomus elumbis to be an inherited defect. The three cases identified in this paper do not allow for the proper genetic analysis (of the three, one was Holstein, one Red Holstein, and one Limousine). The authors mentioned above regarded acaudia to be common in cattle, so eight cases in our analysis did not represent an unusual degree of occurrence.

Epitheliogenesis imperfecta (EI) was reported for the descendants of three sires. Agerholm et al. (1993) detected EI in Hereford calves born of inbreeding, and thus they suggested the involvement of autosomal recessive heredity.

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. Generally, cryptorchidism in cattle is classified as a rare event. According to Amann and Veeramachaneni (2007), cryptorchidism is more common in companion animals, pigs, or humans than in cattle or sheep. Godfrey and Dodson (2005) found that the incidence of testicular hypoplasia increased under tropical conditions. There was a tendency for bulls with testicular hypoplasia or cryptorchidism to have a higher inbreeding coefficient than bulls with normal testes. St Jean et al. (1992) found the breed to be a risk factor for cryptorchidism, but noted its rarity in bovine males; at 1.7 cases per 1 000 bulls, it is in accord with our own findings. In the population analysed in the paper we attribute this positive finding also to the influence of uncompromising selection over the last few decades, as unilateral or bilateral hypoplasia of testes or cryptorchidism has been an unquestioned reason for culling.

We found defects of foetus and stillbirth to be quite frequent; altogether 56 sires were found to have fathered affected calves. Together with unviable calves (n = 32) and dystocia (n = 10), this is a substantial problem of cattle health genetics. Kornmatitsuk et al. (2004) found a continuous increase in the incidence of stillbirth in Swedish Holstein heifers to a current average of 11%. Their results indicated that the aetiology of stillbirth varied depending on the sire used, and was associated with dystocia or low viability of calves. Berglund et al. (2003) concluded that the cause of stillbirth with non-infectious aetiology was likely to be multifactorial and that difficult calving may explain only about a half of the stillbirths; as many as one third of the calves seemed clinically normal with no obvious reason for death. The calving difficulty could be influenced by various non-genetic factors such as season, parity of the dam, sex of calf, short or long gestation length, age at the first calving, and prolonged preceding calving interval (Fiedlerová et al., 2008). Similarly, Mészáros et al. (2008) found a significant influence of the age at the first parturition on the cows' culling. Bureš et al. (2008) recommended the pelvic area measurements and calf birth weight as potential selection criteria to reduce the risk of difficult parturition. Thus, the breeders have an important management tool to influence the course of the parturition. The incidence of stillbirth is a serious factor in the dairy cattle economics (Meyer et al., 2001a), and although some genetic analyses have been carried out already for calving difficulty, stillbirth and birth weight (Meyer et al., 2001b; Eriksson et al., 2004, etc.), further extensive studies for assessment of the genetic parameters are necessary.

Of sires producing stillborn calves, 12 also fathered unviable calves, 5 aborted calves, 4 dystocia, and 9 perishing calves. Other defects were found associated with stillbirth, among them contractures of limbs (n = 4), shortening of the spinal column etc. In seven sires, schistosomus reflexus accompanied defects associated with reproduction such as mummified foetus, big foetus, stillbirth, prolonged gravidity, abortus, cleft genitals, cryptorchidism, and double scrotum.

Since the genetic background of stillbirth and dystocia seems to be unquestioned, sires with a

high prevalence should be a primary target for disqualification from the fathering of stock bulls or culling.

CONCLUSIONS

In conclusion, our analysis found a broad spectrum of congenital disorders over the time span of fifteen years. The frequency was very varied; a few defects occurred in the progeny of tens of sires, but the most typical incidence of disorder was lower than four. The genetic analysis of rarely occurring disorders is difficult when over a long period only a few cases are reported, and even then the discrimination between inherited and acquired defects is troublesome. Nevertheless, sires fathering affected offspring should not be used widely in artificial insemination programmes, but disqualified for the fathering of stock bulls or even definitively culled, as the possible spread of inherited recessive disorders may result in a negative impact on the economics of the cattle industry.

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Corresponding Author

Doc. ing. Jindřich Čítek, CSc., Department of Genetics, Animal Breeding and Nutrition, Faculty of Agriculture, University of South Bohemia, Studentská 13, 370 05 České Budějovice, Czech Republic
Tel. +420 387 772 591, fax +420 387 772 593, e-mail: citek@zf.jcu.cz