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Complex vertebral malformations in Holstein calves

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Since October 1999 an increasing number of malformed neonatal calves of the Holstein breed have been reported to the Danish bovine genetic disease programme. This programme was established in 1989 to recognize hereditary defects in the Danish cattle population and to reduce the negative influence of such diseases to cattle breeding.

Several calves were submitted for necropsy at either the Danish Veterinary Laboratory or the Royal Veterinary and Agricultural University. Within the first months of year 2000 it became obvious that several calves having similar malformations occurred in a familiar pattern consistent with an autosomal recessively inherited defect. Many calves were progeny of the Danish Holstein bull KOL Nixon (DK234042). KOL Nixon has been used for around 127,000 inseminations in Denmark since 1994. The bull T Burma (DK230104) occurred as grandfather to a number of calves and by pedigree evaluation it became clear that T Burma had to be a carrier of the same defect. More than 340,000 inseminations with semen from T Burma have been performed in Denmark since 1994.

Based on the main lesions in affected calves, the defect is designated complex vertebral malformations (CVM) in Holstein calves.

Breeding trial

Based on these observations a breeding trial was initiated to ensure the inheritance of the defect. Questionnaires were sent to approximately 300 farmers having cows being daughters of T Burma and inseminated with semen of KOL Nixon. Aborted, stillborn or defective calves had to be submitted for laboratory examination. The results will be evaluated within few weeks.

Malformations

Several calves having malformations were submitted for necropsy. Affected calves were necropsied and the skeletal system was radiographed. Virological and serological tests were performed to exclude an infectious etiology. Parentage control was performed by examination of DNA profiles.

Malformations have been observed both in aborted fetuses, prematurely born calves, stillborn calves, and neonatal calves. Cases among older calves have not been observed yet. In general the body weight is reduced. Externally, there are two major findings: In many cases the cervical and/or the thoracic part of the columna seems to be short. Moderate bilateral symmetric contraction of the carpal joints and severe contraction and lateral rotation of the phalango-metacarpal joint (fetlock) are constant findings. Contraction and medial rotation of the phalango-metatarsal joint and slight extension of the tarsus are also common findings.

In most cases an irregular course of the columna around the cervico-thoracic junction is observed. Scoliosis may be observed, and lesions may be present in other regions of the columna. The irregular course is often recognized by inspection and palpation of the ventral aspect of the columna. However, lesions may be minimal and restricted to two or few vertebrae. In such cases the columna may be of almost normal length. Therefore, radiological examination of the columna is recommended to exclude vertebral malformations in suspected cases. The spinal cord is of normal size lying in a focally malformed vertebral canal without compressions.

Using radiology complex vertebral malformations consisting of hemi-vertebrae, fused and malshaped vertebrae and ribs, scoliosis, and synostosis are found at various degrees. This is best demonstrated following removal of the arcus.

In some cases malformations of the heart are present, mostly as a high interventricular septal defect and eccentric hypertrophy of the right ventricle. Malformations of the large vessels may occur. In the lungs fetal atelectasis is present in stillborn calves. Serohemorrhagic fluids are mostly present in the thoracic cavity. A variety of other malformations have been observed, but these are not constant or common findings. Lesions due to dystocia are often found.

Other findings

At present there is no scientific evidence of increased return to service rates in cows inseminated with semen of carrier bulls.

Patho-anatomical diagnosis

A pato-anatomical diagnosis is based on the presence of bilateral symmetric arthrogryposis of the distal joints and malformations of the columna, mainly at the cervico-thoracic junction combined with reduced body weight. However, symmetric contractions of the limbs are common findings in vertebral malformations in calves. Therefore, differential diagnostic problems do exist and in some cases differential between CVM and other malformations is impossible.

The variation in the morphological changes of CVM affected calves has not yet been fully determined. This will be done by combining necropsy findings with genotyping.

Genotyping

A genome scan of CVM affected calves and their parents has been performed, and the localisation of the defective gene has been made. Affected calves are homozygous for a specific allele while carriers are heterozygous for this allele. Further information on the molecular basis of CVM will be released as soon as a patent on the diagnostic method is obtained. The method is commercially available at the Danish Institute of Agricultural Sciences.

Common ancestor

Based on the familiar occurrence of defective calves in a pattern consistent with an autosomal recessively inherited defect, the former elite US Holstein sire 7H543 Carlin-M Ivanhoe Bell was found to be a likely common ancestor. Semen of this sire was available and genotyping has confirmed that 7H543 Carlin-M Ivanhoe Bell was a carrier of CVM. Further work will be done to identify other important carriers of CVM used in international breeding.

At present the Danish elite sire T Klassy (DK236398) has also been identified as carrier. This bull has been used for approximately 100,000 inseminations in Denmark of which 80,000 was made within the last 12 months.

It is obvious that CVM will be a worldwide problem in Holsteins just as bovine leukocyte adhesion deficiency (BLAD) was few years ago. It will be possible to reduce the negative influence on the breed by testing bulls used for breeding and thereby prevent the occurrence of malformed calves. The consequences for the Holstein breed will probably be temporary.

Further information

A detailed description of this malformation and other results obtained during this study will be published in international scientific journals as soon as possible.

We are aware of the severity of this defect for the Holstein breed and the great need for testing of breeding animals. We will make every effort to help breeding associations in resolving this problem.

Questions regarding specific aspects can be forwarded to the relevant co-author.

Fig. 1 A case of complex vertebral malformation in a Holstein calf. Notice short neck and contration of the distal joints.

Fig. 2 Hind limbs of an affected calf. Notice symmetric contraction and rotation of the fetlock and phalanges.

Fig. 3 Macerated specimen showing multiple malformations of vertebral corpora. Ventral aspect, cervico-thoracic junction.

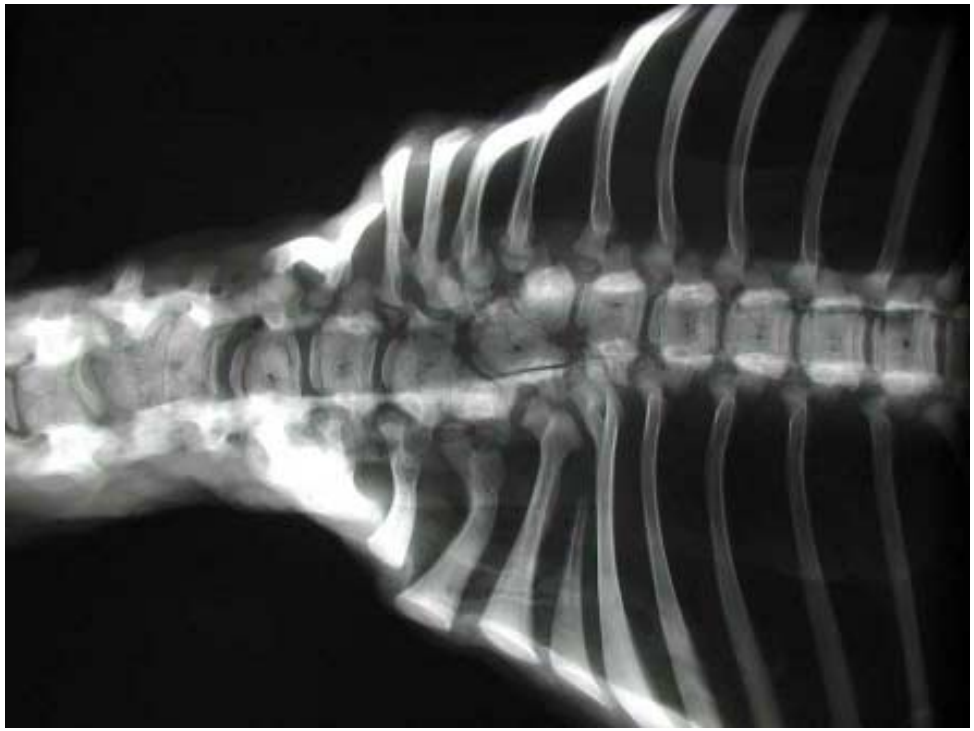

Fig. 4 Radiografh showing malformed vertebrae. Arcus and spinal cord have been removed.