

## GENETIC DEFECTS OF THE SKELETAL SYSTEM IN CATTLE

### Défauts génétiques du système squelettique des bovins

### Defectos genéticos del sistema esquelético en el ganado vacuno

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Each body system is susceptible to congenital defects. The skeletal system is involved in many congenital defects and either an isolated part or the entire skeleton may be affected. Although many different skeletal dysplasias have been described in man, relatively few dealt with cattle (8). But, congenital skeletal defects are economically important in cattle.

This report further describes clinical, macroscopic, and microscopic changes together with pathogenetic and genetic data of bovine skeletal defects. It is a sequel to earlier papers on the same subject (1-9).

#### MATERIALS AND METHODS

The cases were collected at Kansas State University as outlined previously (1-3, 5-9). Technicians of the University-owned Artificial Breeding Unit were requested to report anomalous calves born in patrons' herds (dairy and beef). Additional cases were received from cattlemen, practicing veterinarians and from Dykstra Veterinary Hospital.

Between 1964 and 1974, defective calves were purchased or collected. Each calf underwent detailed necropsy. Affected tissues were fixed in 10 % buffered neutral formalin, routinely processed, and stained with hematoxylin and eosin. Bone sections were decalcified in formic acid then routinely prepared for sectioning. Selected cases also were studied with the scanning electron microscope (SEM).

Since 1972, the research on congenital defects has been expanded to include karyotyping. In addition, a research herd of animals heterozygous or homozygous for genetic skeletal defects is maintained.

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RESULTS

*Frequency of congenital skeletal defects*

Out of a total of 1275 congenital defects studied in Kansas, most common were skeletal defects followed by those of the central nervous system and the muscular system (Table 1).

TABLE 1  
BODY SYSTEMS INVOLVED IN BOVINE CONGENITAL DEFECTS IN KANSAS HERDS

Body system	Percent of total
Skeletal system ... ..	37.3
C. N. S. and eye ... ..	30.7
Muscular system ... ..	14.9
Large body cavities ... ..	4.9
Digestive system ... ..	2.8
Reproductive system ... ..	2.4
Skin ... ..	1.9
Circulatory system ... ..	1.9
Other systems ... ..	1.2

The 476 skeletal defects are recorded by affected body region in Table 2. Generalized skeletal defects were chondrodystrophy in Angus and Hereford cattle, osteopetrosis in Angus cattle and persistence of secondary spongiosa in Hereford cattle.

TABLE 2  
SKELETAL REGIONS AFFECTED BY BOVINE SKELETAL DEFECTS IN KANSAS HERDS

Skeletal region affected	Number of calves
Generalized skeletal defects ... ..	95
Facial region ... ..	67
Mandible ... ..	28
Axial skeleton ... ..	232
Appendicular skeleton ... ..	54

Regional skeletal defects included campylognathia, Roman nose, schistopropus and palatoschisis. Tetramelic arthrogryposis and palatoschisis in Hereford and Charolais cattk were frequently seen. Brachygnathia inferior was commoner than agnathia. Some rare syndromes like short spine lethal and atlanto-occipital fusion were seen. Most frequent defects of the axial skeleton were kyphoscoliosis and taillessness.

Several types of appendicular skeletal defects were encountered, including polydactyly, monobrachia, adactyly, ectrodactyly, and syndactyly.

Bovine skeletal defects with genetic cause

The genetic skeletal defects (generalized or regional) we studied in the field and in breeding experiments are given, with pathogenetic comments, in Table 3.

TABLE 3  
BOVINE SKELETAL DEFECTS OF GENETIC ORIGIN IN CATTLE

Type of defect	Pathogenetic feature	Inheritance pattern	Breed affected
Chondroystrophy ...	Defect of cartilage model.	Recessive.	Angus, Hereford, Holstein, Charolais.
Osteopetrosis ...	Bone formation normal, disturbed or absent bone resorption.	Recessive.	Angus.
Syndactyly I ...	Fusion or nondivision of functional digits.	Recessive with incomplete penetrance.	Holstein, Chianina.
Syndactyly II ...	As above and facial defects.	Recessive.	Angus.
Tibial hemimelia ...	Agenesis of tibia and multiple other defects.	Recessive.	Galloway.

*Chondrodystrophy.* Dwarfism (chondrodysplasia, bulldog calves, achondroplasia) was economically important in American Angus and Hereford herds. A chondrodystrophy gene was also present in Charolais and in Holstein cattle in Midwest U. S.

*Osteopetrosis.* Osteopetrosis (lack of bone resorption) occurred in both sexes of red and black Angus cattle. Affected calves were stillborn, small, and had short, immobile mandibles. Long bones, although completely solid and lacking bone marrow cavities, fractured easily under lateral pressure.

Gross examination of longitudinal bisections of bones revealed absence of bone marrow cavities and a solid bone within bone appearance. Radiographs disclosed homogenous bone densities without distinction between cortical bone and bone marrow cavity. The cranial cavities reduced in size gave rise to cerebellar coning. The optic nerves were hypoplastic.

Histologic and electronmicroscopical examination of bone tissue disclosed persistence of primitive chondro-osseous tissue within the medullary cavity. Bone resorption was deficient. Osteoclasts were present but appeared to be inactive. Other histological changes included mineralized vascular walls and neurons in various areas of the brain including the hypothalamus which was most severely affected.

Pedigree analyses of the calves affected with osteopetrosis revealed common ancestry in each case. In 200 randomly selected pedigrees of the Angus breed, bull X (known to be free of dwarfism) was a common ancestor of 99.5%. Herds with cases of osteopetrosis had different ancestry from bull X.

Analysis of herd data suggested that osteopetrosis resulted from a simple recessive gene. Preliminary results of a breeding trial at Kansas State University are confirmatory.

In addition, we diagnosed osteopetrosis in a grade Hereford herd and have initiated studies of macroscopic and microscopic features and mode of transmission.

*Syndactyly in Cattle.* Syndactyly in Holstein-Friesian cattle (fusion or non-division of functional digits) resulted from a simple recessive gene with incomplete penetrance.

The external features of syndactyly in Holstein-Friesian cattle include four groups: 1) normal overlaps, despite sy/sy genotype; 2) partial syndactyly, two clearly defined hooves partially fused at the axial-coronary border; 3) intermediate syndactyly, a syndactylous hoof's dual origin indicated by dorso-axial groove; 4) complete syndactyly, a hoof with no indication of its dual origin.

The external fusion pattern includes right-left and front-rear gradients, which were paralleled by the osteological pattern.

The most proximal synostosis involved the carpal and tarsal bones: synostosis of intermediate and carpal bones, synostosis of the first tarsal bone with the fused second and third tarsal bone.

The right metacarpal bone was brachydactylous in most syndactylous animals. The distal trochleas were reduced to a single trochlea with one or two sagittal ridges. Some animals revealed tendency towards syndactyly by enlargement of the small metacarpalia with numbers of proximal sesamoid bones varying. The phalanges revealed horizontal synostosis with second phalanges most frequently fused followed by fused third and first phalanges.

The muscles had adapted to the syndactylous hooves. In the fetlock region, the three digital extensor muscles fused to a wide aponeurotic plate which inserted on phalanx II and III. The superficial and deep flexor tendons usually remained undivided. The vascular and nerve supply were also adapted to the syndactylous condition.

Syndactyly, an anatomical defect, is associated with hyperthermia, a functional defect.

During the 15 year period of our study of syndactylous cattle, nine syndactylous cattle had serious heat adaptation problems and nine, slight discomfort during hot summer weather.

We subjected five syndactylous and five control cattle to standardized, moderate, temperature stress in a climate chamber. All syndactylous cattle developed clinical signs of hyperthermia, including elevated rectal temperatures (41.5° to 45° C), tachycardia, and tachypnea. Two syndactylous cattle developed oliguria and haematuria; one developed polyuria, and three became recumbent and paralyzed after exposure. One cow died without premonitory signs after 96 hours exposure. Blood chemical changes were slight, except for preterminal hyperglycaemia in two cows. Change in total leukocyte count was small. However, relative and absolute eosinopenia, lymphopenia, and neutrophilia occurred in the syndactylous cattle. The only changes observed in the controls were attributable to normal adaptation. Post mortem examination revealed a wide-spread parenchymatous degeneration.

We recently diagnosed syndactyly in Chianina cattle. Further research is needed to determine its importance in that breed. The anatomical pattern is identical to that of Holstein-Friesians. Although we also diagnosed syndactyly in Hereford calves, we did not determine its cause.

*Syndactyly and facial hypoplasia in Angus calves.* A new autosomal recessive lethal we discovered in a purebred Angus herd affected all four feet and the face. The osteological defect differed considerably from syndactyly in Holstein calves.

*Tibial hemimelia.* Tibial hemimelia diagnosed in Galloway calves left both hind legs without tibias. Associated lesions were encephalocele and abdominal hernia.

The calves were related and the pattern of occurrence was consistent with simple recessive inheritance.

## DISCUSSION

Reportedly, 0.2 to 3.6 percent of all calves born are affected with congenital defects (1-8). Congenital defects in cattle result in economic losses from decreased reproductive capacity, increased perinatal losses and reduced value of related calves when defects are genetic. Many congenital defects follow a simple pattern of Mendelian inheritance. Others are caused by environmental factors, and still others have no clearly established cause. Various parts of the body may be affected with a congenital defect. Some may be single isolated defects; however, it becomes increasingly obvious, that many defects occur together as part of a syndrome. Frequencies of defective anatomical structures vary with geographic region, breeds, and other factors. However, in most studies in cattle, the most frequently encountered congenital defects are those of the central nervous, muscular, and skeletal systems (8).

## SUMMARY

A study of congenital defects in cattle revealed that about one-third of the defects involved the skeletal system, either the entire skeleton or skeletal parts. Most congenital defects occurred as syndromes rather than as single entities.

Skeletal dysplasias encountered were chondrodystrophy, persistence of secondary spongiosa, and osteopetrosis. Chondrodystrophy affected Hereford and Angus calves. Rare types of chondrodystrophy were encountered in Holstein-Friesian and Charolais calves.

The commonest genetic diseases of localized skeletal regions were syndactyly in Holstein-Friesians, syndactyly with facial hypoplasia in Angus calves, and tibial hemimelia in Galloway cattle.

## RESUMEN

El estudio de los defectos congénitos del ganado revela que aproximadamente un tercio de estos defectos tiene que ver con el sistema esquelético, bien en su totalidad bien en alguna de sus partes. La mayoría de los defectos congénitos aparece como síndromes más que como entidades individuales.

Las displasias esqueléticas encontradas fueron condrodistrofias, persistencia de la esponjosa secundaria y osteopetrosis. La condrodistrofia afectaba a los terneros Hereford y Angus. Fueron encontrados tipos poco frecuentes de contro-

Los trastornos genéticos más frecuentes localizados en zonas del esqueleto

fueron sindactilia en la raza Holstein-Friesian, sindactilia con hipoplasia facial distrofia en terneros de las razas Charolesa y Holstein-Friesian. en terneros Angus y hemimelia tibial en ganado Galloway.

## RESUME

Una étude des défauts congénitaux dans le bétail révéla qu'environ un tiers des défauts affectaient système squelettique, soit le squelette entier soit des parties du squelette. La plupart des défauts congénitaux se présentèrent comme des syndromes plutôt que comme de simples entités.

Des dysplasies squelettiques trouvées étaient des chondrodystrophie, persistance de spondiosa secondaire et de osteopetrosis. Les chondrodystrophies affectèrent des veaux d'Hereford et d'Angus. Des sortes rares de chondrodystrophies furent trouvés dans des veaux Holstein-Friesians et des veaux Charolais.

Les maladies génétiques les plus communes de régions squelettiques localisées étaient syndactylement dans les Holstein-Friesians, syndactylement avec hypoplasia faciale dans les veaux d'Angus, et hemimelia tibiale dans le bétail Galloway.

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