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Prominent Congenital Defects in Nebraska Beef Cattle

This NebGuide describes congenital defects that affect the economic value of beef cattle, and provides recommendations on how to minimize them in breeding programs.

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Congenital defects are abnormalities of structure or function present at birth. They may be caused by genetic or environmental factors, or a combination of both. The causes of many defects remain unknown.

Developmental defects may be lethal, semi-lethal, or compatible with life, causing very little effect or only aesthetic effect.

With the increasing use of artificial insemination (AI) in cattle, defects are no longer rare occurrences and are important collectively to the cattle industry. Congenital defects cause economic losses by increasing perinatal calf mortality, decreasing maternal productivity, and decreasing the value of defective calves and their relatives.

Susceptibility to injurious agents varies with fetal development and decreases with age. Before Day 14 of gestation in cattle (period of pre-attachment), the zygote or embryo is resistant to agents that produce the incidence of congenital malformations (teratogens), but is susceptible to genetic mutations and abnormality of chromosome numbers or structures (chromosomal aberrations). During the embryonic period (Days 14-42), the embryo is highly susceptible to teratogens, but this decreases with embryonic age as critical periods for the various organs are passed. The fetus (Day 42+) becomes increasingly

resistant to teratogenic agents with age, except for the late differentiating structures, such as cerebellum, palate, and urogenital system.

Defects are classified by the body system affected. Approximately two-thirds of genetic defects in calves affect the skeletal system, central nervous system, and eye. These conditions are the most visible to the producer. The muscular system, large body cavities, digestive system, reproductive system, skin, and circulatory system comprise the remaining one-third of the recorded defects.

The majority of genetic defects in cattle are inherited as recessives. Approximately one in 10 animals carry a lethal or sub-lethal allele. They reflect harmful mutations that have become established. They usually result from inbreeding, a consequence of which is the founder effect. This effect is defined as the derivation of large populations of beef cattle from a small genetic pool. One animal, therefore, has had a large and disproportionate influence on the development of the breed. This is caused by concentrated use of one blood line. It has been evident in the early use of exotic European breeds, AI, and inbreeding. If a carrier of a deleterious allele is evident, then through the founder effect it may be responsible for the increased incidence of genetic defects in later generations.

Retrospective studies have identified the founder effect of mannosidosis, dwarfism, syndactyly, polydactyly, hypotrichosis, silver-eyed Angus, and arthrogryposis-cleft palate of Charolais.

Skeletal System

Dwarfism

Dwarfism occurs in all breeds of cattle. It is a defect of the interstitial growth of the epithelial, articular, and basal cranial cartilages. It results in variable shortness of the legs, cranial base, and vertebral columns. It is generally considered a recessive gene effect with varying degrees of expression.

Osteopetrosis

Osteopetrosis (Marble Bone) has been reported in calves of Black and Red Angus, Holstein-Freisian, Simmental, and Herefords, and is characterized by small body size and an undershot jaw with impacted molar teeth. The most common finding is the lack of bone marrow cavities. Calves affected with osteopetrosis are born prematurely at 251 to 272 days gestation and may be mistaken for an abortion problem. This is caused by a simple autosomal recessive gene.

Syndactyly

Syndactyly (one-toe) is defined as a fusion or non-division of functional digits. It is common in Holstein, Chianina, Angus, and Simmental cattle, and is due to a simple autosomal recessive gene. The genetic form expresses the defect in the right front foot first, then the left front foot, followed by the right and left hind feet.

Polydactylism

Polydactylism (three-toes) is a duplication of the number of toes. It is common in Simmental and Holstein cattle. It is a polygenic trait (many genes at different loci) with various forms of expression. Incomplete penetrance of the condition can occur; the foot appears normal and an X-ray is needed to reveal the extra digits. In addition to the genetic form, viral forms have been reported.

Arthrogryposis

Arthrogryposis is the most common skeletal defect observed at the present time. It is defined as permanent abnormal joint fixation and is present at birth. It has been reported in conjunction with cleft palate in Charolais, and is a simple autosomal recessive trait with penetrance. Bluetongue and bovine virus diarrhea (BVD) viruses appear to be common causes of this condition. It is reported in all major breeds.

Central Nervous System Problems

Internal hydrocephalus

Internal hydrocephalus is an accumulation of excessive fluid within the ventricular system of the brain. The animals are generally born dead or die within a few days after birth. It is inherited as a simple autosomal recessive gene in Herefords and Shorthorns, and is also caused by BVD and bluetongue viruses.

Cerebellar hypoplasia

In cerebellar hypoplasia, the cerebellum is small or absent. It has been described as a genetic defect in many breeds, and intrauterine fetal infections with BVD and bluetongue viruses have also been incriminated as causative agents. Affected calves are recumbent with extended limbs, intermediate opisthotonos (head twisted backward), and ataxia.

Mannosidosis

Mannosidosis is a lyosomal storage disease associated with accumulation and storage of the enzyme a-mannosidase. The deficiency of a-mannosidase is characterized by ataxia, incoordination, head tremor, aggression, and reduced growth. Calves may be affected at birth, but clinical signs usually do not appear until they are several weeks or months of age. The most affected cattle die within the first 12 months of life. Heterozygous animals are carriers. This is a simple autosomal recessive condition and can be defined biochemically. It has been reported in Angus and Murray Grey cattle.

Skin Defects

Congenital hypotrichosis

The most common skin defect is congenital hypotrichosis, which is the complete or partial congenital absence of hair. Six forms have been described, varying from complete hairlessness to semi-hairlessness. The semi-hairless types generally occur in Polled Herefords. They have a very thin hair coat at birth. The hair is sparse and patchy, and the skin may be wrinkled and scaly. Histologically, degenerative changes in the hair follicles can be observed. This is a recessive trait. Hairless conditions are also caused by BVD or bluetongue virus.

Ehlers-Danlos Syndrome

Ehlers-Danlos Syndrome is characterized by extreme fragility of the skin and laxity of joints. Collagenous tissues of the body exhibit fragmentation and disorganization of collagen fibers. Affected calves show delayed healing of skin wounds. Minimal trauma gives rise to hematomas due to a deficiency of procollagen peptinase.

Eye Defects

Albinism

Albinism (congenital absence of pigment of the eye) in cattle has been described in numerous breeds. The most recent condition has been partial albinism (heterochromia iridis) in Black Angus cattle. The most distinguishing factor involves the iris color. Angus cattle usually have a dark black iris; the heterochromia iridis calves have a light, usually two-colored iris. This gives a double ring appearance to the iris when viewed closely (an outer faintly brown ring and an inner light blue ring circling the pupil). The pupil always appears constricted in daylight. From a distance the eyes appear white or silver. The fundus of the eye appears albinotic. The animal will show photophobia and may be prone to eye irritation. However, this does not impair the animal's ability to reproduce. This defect is inherited as a simple autosomal recessive.

Ocular dermoids

Ocular dermoids are peculiar defects characterized by solid skin-like masses of tissue on or in various ocular structures. They generally occur in one eye only and their cause is controversial. Although a cosmetic blemish, the main concern is that they are a continual source of irritation and cause visual impairment if large areas of the cornea are involved. Depending on the breed, they show characteristics of both autosomal recessive and polygenic inheritance.

Cardiovascular System

Heart defects are a common problem in cattle and are generally recognized only during necropsy. Most of their causes are unknown.

Ectopia cordis

Ectopia cordis is when the heart is located in the cervical region outside the thoracic cavity, or in the abdominal cavity. Ventricular septic defects may be singular or isolated defects, or may be combined with abnormalities of the large vessels. Sixteen different septal defects have been reported.

Reporting

All congenital defects should be reported to a diagnostic laboratory, veterinarian, Al center, or animal breed association as to the defect found. Complete necropsy should be performed on all animals and an extensive herd history along with a breeding record analysis should be conducted. If the defect is carried by a superior sire and the mode of inheritance is known, then the breeder must decide whether the sire's superior production traits outweigh the obvious defect. If semen is to be sold, all customers should be informed that the animal is a carrier and warned of the possible consequences of using the semen. Carrier bulls should be out-crossed with different lines or breeds in operations where commercial production is the main objective. The bulls should not be used in inbreeding or line breeding programs. Once a clean superior son has been attained, dispose of the carrier bull as soon as possible.

Tests using superovulation, embryo transfer, and preterminal C-section have been conducted to determine genetic defects. Father-daughter test matings can be conducted with a 99% probability by mating the suspected carrier bull to 35 of his daughters.

Accurate diagnosis of defects partially or wholly caused by genetic factors is necessary before control

measures can be established. It is important to recognize that congenital defects are economically significant to the cattle breeding industry.

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