Cow/Calf Session II

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Congenital Defects of Current Concern in Beef Cattle

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Introduction

Congenital defects are abnormalities of structure, formation of function present at birth. They may affect a single anatomical structure or function, an entire system, parts of several systems, or may involve several body systems, or combine functional and structural defects to form syndromes. Our consultancy program at Kansas State University utilizes a combination of clinical examination, pathology, genetics, research and a central congenital defects file, to assist breeders and the AI industry in the decision making process of breeding and defect monitoring programs. Congenital defects of current concern in beef cattle are reviewed here.

Causes

Many congenital defects have no clearly established cause; others are caused by environmental or genetic factors, or by environmental-genetic interactions.

Environmental Factors

Teratogens are factors identified as causing fetal anomalies. Teratogens usually do not affect the pregnant dam but damage the embryo/fetus primarily during the first 40 days.

Teratogens reported in cattle include toxic plants, viruses, drugs, trace elements, and physical agents such as irradiation, hyperthermia, and undue pressure during rectal examination in early pregnancy. Teratogens follow seasonal patterns or known stressful conditions, may be linked to maternal disease, but do not follow a familial pattern.

Toxic Plants

Ingestion of lupines (*L. sericeus, L. caudatus* and *L. nootkatensis*) between day 40-70 of gestation may cause joint contractures in calves associated with torticollis, scoliosis and/or kyphosis and various degrees of cleft palate.[57,62] Keeler and others at Logan, Utah identified anagyrine as the teratogenic principle in lupines.[57]

Viral Infections

Prenatal viral infections may be teratogenic in cattle. Intrauterine Akabane virus infection caused abortion, premature births, and congenital defects (arthrogryposis and hydranencephaly) in Australia, Japan, Israel, and Kenya.[33,59,60] Bovine virus diarrhea virus induced cerebellar dysplasia, ocular defects, brachygnathia, intrauterine growth retardation, and impaired immunologic competence.[24,39] Experimental transmission of bluetongue virus to pregnant heifers via insects resulted in abortion, stillbirths, and congenital defects such as arthrogryposis, campylognathia, and prognathia with domed cranium.[26,73]

Physical

Rectal palpation between day 35 to 40 gestation has been reported to cause atresia coli.[88]

Genetic Factors

Genetic defects are pathophysiologic results of mutant genes or chromosomal aberrations occurring in any environment. A number of congenital defects are inherited, many as simple autosomal recessives. At least 260 visible traits and defects of cattle have been reported to be controlled by major genes; most are pathologic usually congenital, and are important to the cattle breeding industry.[46] Diagnosis of defects due to genetic factors is necessary before effective control can be established. In Kansas for many years, a systematic program for gathering, recording, interpreting, and communicating information on congenital defects has been maintained.[67]

As a rule, genetic defects determined by mutant gene(s) run in families and occur in typical intergenerational patterns and intergenerational frequencies requiring enumeration of normal and abnormal calves of a bull and identifying familial relationships. Various statistical methods may be used to analyze the data.[67] Breeding trials are necessary to confirm the genetic pattern of transmission of a defect, by conventional methods or by superovulation of homozygous affected or heterozygous animals, embryo transplantation and preterminal cesarean section where applicable.[10,14,37,53,54,65,67]

Chromosomal Defects

Although chromosomal aberrations are diagnostically important in man, their frequency and significance in cattle have yet to be ascertained. The most common chromosomal defects in cattle are 1/29 Robertsonian translocation and 14/20 translocation.[74,81]

Nature and Effect

Congenital defects result in fewer losses than those caused by nutritional deficiencies, infectious agents, or neoplasia. Defects, however, may cause considerable economic losses by increasing perinatal mortality. Furthermore, loss of value of relatives of genetically defective calves can be serious. Additional losses occur when congenital defects are a manifestation of a syndrome that also includes embryonic and fetal mortality. Congenital defects may confuse the diagnosis of other diseases and abortion. Finally, if the defect is genetic, control measures may require extensive and expensive adjustments of breeding programs.

Frequency

The frequency of congenital defects in calves is not known. From medical records of 137,717 animals in North American Veterinary Clinics, 6,455 had congenital defects; 533 in cattle.[82]

In a Japanese study of 223 calves with congenital defects, involvement by body system was skeletal, 39.1%; circulatory, 12.1%; central nervous, 9.5%; digestive, 8.1%; muscular, 5.6%; defective twinning, 4.4%; female reproductive, 3.9%; urinary and male reproductive, 3.1%, respectively; large body cavity, 3%; and others, 8.1%.[42] Findings from studies in Kansas were similar.[31]

Kansas Genetic Disease Program

Animal breeders and veterinarians are involved daily in improving animal health and production. Accurate diagnosis of diseases and defects, partly or wholly caused by genetic factors, is necessary before effective control measures can be established. To that end in Kansas for many years, a systemic program for gathering, recording, interpreting, and communicating information on congenital and genetic diseases has been maintained.[67]

Control

If a defective calf is submitted, the history should include breed, age of parents, parentage of affected and unaffected control calves, geographic region, season, type of pasture, soil type, exposure to or suspected exposure to teratogenic plants, feeding and management practices, breeding records, maternal medical and vaccination records, disease status of the herd, periods of stress, drugs administered, congenital defects observed previously, and history of any similar congenital defects in neighboring herds. Breeding records may be analyzed for evidence of inbreeding and for characteristic intergenerational transmission patterns and intragenerational frequencies. Necropsy is performed and the defect(s) classified by the body system primarily involved. Serum samples are taken to check for bovine virus diarrhea and other viral antibodies. Samples of brain and other tissues are taken for possible virus isolation. Relevant tissues are histologically examined, and selected cases are submitted for electron microscopic study.

For decisions on breeding programs, the following steps should be taken. Most breed associations follow similar procedures:

1. Blood typing to establish verified parentage, especially where AI sire is involved;
2. Certified statement by a veterinarian or third-party witness;
3. An extended pedigree chart made;
4. Laboratory examination by a pathologist where applicable; and
5. Decision withheld until all reasonable doubt has been eliminated (usually requires two or more thoroughly

documented reports for bulls).

If it is determined that a bull is carrying an undesirable recessive gene, most organizations proceed to label the bull as a heterozygote and remove him from service. If he is not removed, advertisements should carry the statement that he is a carrier of this defect and information concerning the congenital defect.

Breeding trials are done conventionally. However, superovulation of affected cows followed by insemination and embryo transfer (two per recipient) and early cesarean-section at 60 days have been developed for testing sires and females[53,65]

Biochemical testing methods will be more important in the future. Heterozygotes for protoporphyria, dumps, cirtullinemia, α-and β-mannosidosis can be detected by enzyme tests.[1,13,55,56,64,83,87,89] Test methods based on DNA studies will also be employed in the future.[34,90,98]

Diagnosis of Specific Defects

It is difficult to establish a single system suitable for classifying congenital defects. Three principal systems have been used: etiologic, affected embryonic tissue, and principal defective body system involved; the last system is used.

Skeletal Defects

Bone defects are common and may involve a single structure or the entire skeleton. Single, isolated skeletal defects are common and some are economically important.[67]

Craniofacial Defects

Defects involving face and/or cranium may be isolated or combined with defects in other body systems.[52,67,78]

A facial defect of either sex and various breeds has been seen with somewhat increasing frequency; a split face with the tongue and lower jaw located in the center with a nostril on either side (schistoprosopia). Studies to learn more about the nature of this defect and its cause are in progress.

Cleft lip (cheiloschisis) may affect one or both sides. In our studies in Angus and Jersey cattle, unilateral appears to be more frequent. The defect is inherited but not as a simple autosomal recessive.

Short lower jaw (brachygnathia) is seen most commonly in Simmental cattle. Studies using affected dam and sire produced normal and abnormal calves indicating genetic transmission but not as a simple autosomal recessive.[101]

Other craniofacial defects described in cattle (prognathia, agnathia, etc.) need further etiologic and diagnostic evaluation.[67]

Vertebral Column

Several congenital defects involve the spinal column and include short spine lethal, atlanto-occipital fusion, kyphosis (dorsal arching of back), lordosis (ventral), scoliosis (lateral) and torticollis (twisted). The defects may occur alone or in combination (kyphoscoliosis) or are associated with defects of other body systems.[25,67]

A more common defect is perosomus elumbis characterized by partial or complete lack of development of the spinal column caudal to the thoracic area and accompanied by posterior bimelic arthrogryposis. Affected calves are recumbent as they cannot use their hind legs and have to be destroyed. The defect is suspected to be inherited.

Appendicular Skeleton

Defects involving abnormal limb development are usually inherited and economically important. Syndactyly defined as fusion or nondivision of functional digits occurs in various breeds such as Angus, Chianina and Simmental and is inherited as a simple autosomal recessive. It is better defined in Holstein cattle where it is inherited as a simple autosomal recessive with varying degrees of expressivity, incomplete penetrance and pleiotropic effect. The right front leg is usually affected followed by the left front leg. If three legs are affected, they are the right and left front legs and the right rear leg. The most advanced degree of syndactyly usually involves the right front leg. Involvement of all four feet is rare in Holstein cattle whereas it is common in beef cattle.[32,37,67,76]

Angus and Holstein/Angus crossbred calves had similarities between Angus and previously described changes in Holsteins, as well as several striking differences. Angus-Holstein crossbred calves revealed a combination of both patterns. Syndactyly in Angus calves is due to homozygosity of a simple autosomal recessive gene.[67] Furthermore, syndactyly genes are allelic in Holstein and Angus cattle. Bulls suspected to be heterozygous for syndactyly are being tested by superovulating syndactylous cows, embryo transfer and preterminal cesarean section at 60 days gestation. Cows are tested similarly using semen from syndactylous bulls.[10,37,53,65]

Polydactyly, a medical increase in the number of digits, usually affects the fore legs in various cattle breeds.[54] Polydactyly in Simmental cattle is transmitted polygenically requiring a dominant gene at one locus and recessive genes at another.[54]

Tibial hemimelia, lack of development of both tibias, has been described as a simple autosomal recessive in Galloway calves. It is usually associated with meningocele and abnormalities of the pelvic bones and reproductive system.[63]

Systemic Skeletal Defects

Chondrodysplasia ("Bulldog")

A calf affected with chondrodysplasia has short legs, large round head, cleft palate, internal hydrocephalus, and disturbed bone growth. Chondrodysplasia is a defect of in-

terstitial growth of cartilage causing bones to be short and disorganized.[45,51] The mode of inheritance needs to be clarified.

Osteopetrosis

Osteopetrosis, a recessive hereditary defect in black and red Angus and Simmental cattle, is characterized grossly by small body size and weight, brachygnathia inferior with impacted molar teeth, misshapen coronoid and condyloid process, open fontanelle, thickened cranial bones, agenesis or hypoplasia of major foramina of the skull, and lack of bone marrow cavities.[69,77] Calves affected with osteopetrosis are born prematurely and dead at 251 to 272 days (mean 262) gestation and may be misdiagnosed as abortion. Osteopetrosis in Hereford, Dutch Holstein-Friesian and European Simmental calves, had similar morphologic features to those in Angus calves.[67,69]

Osteogenesis Imperfecta

A defect characterized by bone fragility, joint laxity, dentinogenesis imperfecta, and other connective tissue defects. The inheritance is most likely to be polygenic.[67]

Generalized Cartilage Defect

A defect in Angus calves that causes degenerative disease of all major joints soon after birth. Affected calves have thickened joints and soon become recumbent and finally are unable to move and have to be destroyed. They also have a round shortened face. Transmission seems to be polygenic.[52]

Hereditary Angular Limb Deformity

Angular limb deformity in Jersey calves is genetically transmitted as a simple autosomal recessive. It occurs also in Holstein, Simmental, Gelbvieh, Polled Hereford, and Angus breeds.

Central Nervous System

Congenital defects of the central nervous system (CNS) are common and most are recognized by structural changes, some are functional, and others involve both skeletal and central nervous systems. CNS defects are classified into five groups: 1) cerebral defects and malformations involving only or mainly the cerebrum; 2) defects of cerebellum and brain stem, involving ony or mainly the cerebellum and brain stem; 3) spinal cord defects: 4) spastic and paralytic diseases; and 5) storage diseases.[9,15-20,30-31,34-36,40-44,67]

Cerebral Defects

Internal Hydrocephalus

Internal hydrocephalus, a common and important defect, may be caused by genetic or environmental factors.[3-6] The genetic form in Hereford cattle is characterized by small body size and weight; narrow, refined facial features, and cranial doming; caudodorsal-rostroventral angulation of palpebral fissures; microphthalmia; protruding edematous tongue; mesencephalic kinking at the anterior portion of the aqueduct, lateral splaying of the dorsal thalamus, and absence of interthalamic adhesion. The sigmoid configuration of the brain and splaying configuration of the thalamus resembled the cephalic flexure and diencephalon of a 40-day-old bovine embryo.[4] In contrast to genetic internal hydrocephalus, BVD virus-induced hydrocephalus has lesions such as porencephaly.[3]

Muscular System Defects

Congenital defects of muscle are common in cattle and are economically important. Muscular hypertrophy ("Doppellender", double muscling, muscular hyperplasia) is present in most major beef breeds.[67]

Arthrogryposis

Arthrogryposis, defined as permanent joint contracture present at birth, is a worldwide problem in all cattle breeds.[67] It includes more than one etiologic and pathologic entity. Tetramelic arthrogryposis in Charolais calves associated with cleft palate is inherited as a simple autosomal recessive.[75]

Arachnomelia and Arthrogryposis

A lethal syndrome of arachnomelia and arthrogryposis was described by German and Swiss workers in the 1980's in Simmental and Brown Swiss cattle. It occurred in offspring of Brown Swiss imported from the United States and affected calves in Germany and Switzerland traced back to the same common ancestor. The major lesions are in the skeletal and muscular systems and affected calves are born dead or live only a few hours.[58] The legs are long, slender and thin, the muscles smaller than normal and the joints are either hyperflexible or have contractures and bones fragile. There are additional defects such as short lower jaw and kyphoscoliosis. Affected calves usually cause dystocia. The defect has now been reported in the United States.[71]

Duplication

Congenital duplication in calves mainly affect the skeletal and central nervous systems as recently reviewed.[43]

Defects of the Cerebellum and Brain Stem

Arnold-Chiari Malformation

The Arnold-Chiari malformation (ACM) involves caudal herniation of tongue-like processes of cerebellar tissue through the foramen magnum into the cranial cervical spinal canal with caudal displacement and elongation of the medulla oblongata, pons and 4th ventricle.[17,41] Affected calves have spina bifida of the lumbar area and ar-

throgryposis of both hind legs. It has been reported mainly in Angus calves.

Cerebellar Aplasia or Hypoplasia

Although earlier investigators invariably incriminated hereditary causes in cerebellar disease, intrauterine fetal infection with BVD virus is now incriminated as the cause of cerebellar hypoplasia associated with ocular defects. In light of these findings and evolvement of more precise genetical analyses, the cause of cerebellar defects should be reevaluated.[15,67] Pathological changes in genetic cerebellar hypoplasia seem to differ from those of BVD virus-induced cerebellar defect.[15] Cerebellar abiotrophy occurs in various breeds of beef cattle.[100]

Progressive Ataxia

Progressive ataxia in Charolais cattle occurs at 8-24 months of age primarily as hind leg weakness. It progresses in 1-2 years from slight ataxia involving all four limbs to recumbency. Progressive ataxia is due to defective development of oligodendroglia.[22]

Maple Syrup Urine Disease

A disease of Polled Hereford cattle characterized clinically by extensor spasms and inability to stand, and histologically, by spongiform encephalopathy. The defect is transmitted as an autosomal recessive.[9,35,36]

Spinal Cord Defects

Various developmental spinal cord defects of calves have been reported such as spina bifida and spinal dysraphism. Spina bifida implies a defect of vertebrae with or without accompanying spinal cord involvement. Spinal dysraphism is myelodysplasia of the spinal cord, especially the central canal. Affected calves from birth have hind leg paralysis, severe ataxia, or may be able to stand and have a hopping gait, an extremely wide stance with overextension (goose stepping), and crossing of hind legs while walking or standing.[15,23,44,67]

Spastic and Paralytic Diseases

These diseases include those with clinical evidence of functional CNS involvement and a hereditary basis.

Spastic Paresis

Bovine spastic paresis is characterized by spastic contracture of muscles and extension of stifle and tarsal joints of the affected hindlimb(s). Usually clinical signs indicate a unilateral condition, with the right hind limb being frequently involved; bilateral involvement is rare. Although the disease is assumed to be genetic, the evidence is inconclusive. However, genetic influence(s), as well as environmental factors, may influence its expression.[67]

Spastic Syndrome

Spastic syndrome, a chronic progressive disease, is characterized by sudden spastic muscular contraction of both hind legs and often the back, neck and front legs. Between attacks, muscle function appears to be normal; complete recovery, however, never occurs. Affected cattle of both sexes are 3 to 7 years-old. The mode of inheritance has not been determined; possibly a dominant gene with incomplete penetrance.[91]

Bovine Progressive Degenerative Myeloencephalopathy

Bovine progressive degenerative myeloencephalopathy ("weaver"), a hereditary disease of purebred Brown Swiss cattle, is characterized by hind leg weakness, ataxia, and dysmetria with onset at 5-8 months of age, progressing to recumbency and frequently death from rumen tympany. Lesions are confined to the white matter of the spinal cord and Purkinje cells of the cerebellar cortex. Spinal cord lesions are axon degeneration, loss of axons and myelin, vacuolation of white matter due to large, empty intercellular spaces, and axonal spheroid formation.[27,68,70,94] Since various beef breeds have been established from a Brown Swiss base, this defect and the next may be encountered.

Spinal Muscular Atrophy

Spinal muscular atrophy in Brown Swiss calves is characterized by hind leg weakness at 3 to 4 weeks of age, and finally, severe muscular atrophy, quadriparesis and sternal recumbency. Lesions are degeneration of motor neurons in the ventral horns of the spinal cord and neurogenic muscle atrophy.[28]

Storage Diseases

Lysosomal storage diseases are characterized by accumulation and storage of substances within lysosomes because of specific enzyme deficiency preventing catabolism. Criteria for classifying a disease as an inborn error of lysosomal catabolism are: 1) storage disease; 2) inherited; 3) storage substance, not necessarily homogeneous, should be stored at least initially within lysosomes; and 4) partial or absolute deficiency of a lysosomal enzyme, normally responsible for hydrolyzing the storage material.[55]

Among recorded storage diseases involving the CNS of cattle are GM_1 gangliosidosis, glyconeogenesis, and α- and β-mannosidosis.[1,13,55] Generalized glyconeogenesis type II has been reported in Shorthorn and Brahman beef cattle.

Recently, β-mannosidosis was diagnosed in neonatal Saler calves with death occurring within 48 hours after birth. Affected calves have hydrocephalus and swollen olive green kidneys. Microscopically, vaculoation is observed in neurons, cells of the proximal convoluted tubules of the kidneys and other tissues.[1,13,56]

Metabolic Diseases

Protoporphyria in Limousin cattle is caused by homozygosity of a simple autosomal recessive gene. Heterozygote cattle may be detected by breeding trials, determining ferrocheletase activity in fibroblast tissue cultures, and quantitation of free protoporphyrin in circulating red blood cells.[86,87] Protoporphyric cattle may be affected with ataxia and convulsions.

Clinical signs and lesions were most severe in young animals. Gross, microscopic and ultrastructural lesions have been studied. Liver lesions consisted of portal fibroplasia, bile ductule hyperplasia, parenchymal cell swelling, and pigment accumulation. Maltese cross-like crystals are visible under polarized light. Ultrastructurally, there are large secondary lysosomes comprised of electron-dense granules associated with lipid droplets in hepatocytes. Phagocytic cells in the dermis also contain large heterogeneous secondary lysosomes.[99]

Ocular Defects

Ocular defects are either single or multiple and restricted to the eye, or observed in conjunction with defects in other organs, or associated with pigment deficiencies. Frequency of ocular defects such as anophthalmia and microphthalmia was estimated in six U.S. breeds to range from a maximum of 1 in 50,000 births.[67] Ocular dermoids in Hereford cattle are genetically transmitted, characterized by autosomal recessive and polygenic inheritance.[11]

Skin

Developmental defects involving skin, adnexa and pigment are common in cattle and may be generalized or localized.[49,50,67]

Albinism may be classified as partial, incomplete, and complete.[7,21,50] In partial albinism, the iris is blue and white centrally and brown peripherally, and the coat color is usually characteristic for the breed or more dilute. Incomplete albinos have colobomas of the nontapetal fundus and hypoplasia of the tapetum fibrosum.[67]

Oculocutaneous hypopigmentation identified in Angus cattle is inherited as a simple autosomal recessive trait. The hair over the entire body surface is brown. The skin surface, particularly obvious in glabrous skin around eyelids, ear openings, muzzle, anal and reproductive orifices, is grayish-brown. Affected cattle are photophobic. The iris is light, usually bicolored, with a double-ringed appearance.[21,93] Normal coat color in cattle has been reviewed.[79]

Hypotrichosis

Hypotrichosis in horned and polled Hereford cattle, ranges from slight to severely affected.[12,48,80,84,85] The skin is thin and pliable with only a few hairs per unit area on the lateral and ventral neck, face, ears, thorax, flank, rump, and forehead. Hair over eyelids, around prepuce, umbilicus, and switch of tail is thin, wavy, and silky. Degenerative changes in Huxley's layer were reported as the key to development of hypotrichotic lesions in Hereford cattle.[12,48] Hypotrichotic Hereford calves have histopathological changes in the anagen phase of the hair cycle, characterized by trichohyalin macrodroplets in Huxley's layer.[12,48]

Absence of an arginine-converting-enzyme was suggested as the cause of hypotrichosis in Hereford cattle. However, citrulline is present in follicular tissue of Hereford cattle with hypotrichosis.[85]

Epitheliogenesis Imperfecta

A common skin defect of either sex is epitheliogenesis imperfecta, a simple autosomal recessive lethal trait. Calves have large epithelial defects distal to the carpal and tarsal joints, and one or more defective claws. The muzzle, nostrils, tongue, hard palate, and cheeks also have epithelial defects.[47]

Cross-Related Congenital Hypotrichosis

In cross-related congenital hypotrichosis, the hair is short, curly, dilute-color, malformed, and sometimes sparse. That hair coat causes physical impairment. Because the disease is associated with crossbreeding programs utilizing certain breeds of cattle, it is possible that the causes may be due to color dilution mutants present in European cattle breeds.[8,79]

Congenital Anemia, Dyskeratosis and Progressive Alopecia

A genetic syndrome of congenital anemia, dyskeratosis and progressive alopecia has been reported recently in Polled Hereford cattle.[92] The defect is most likley inherited as a simple autosomal recessive trait.

Cardiovascular System

Most congenital cardiac defects in cattle have been reported as single cases.[29,40,67] In a 14-year study of defective calves 36 were affected with 78 congenital cardiac defects: ectopia cordis cervicalis 10, common aortic trunk 3, dextraposed aorta 8, duplicated major trunks 1, hypoplastic aorta 2, interventricular septal defect 11, interatrial septal defect 2, left ventricular hypoplasia 10, patent ductus arteriosus 5, patent foramen ovale 5, right ventricular hypoplasia 10, cor triloculare biatrium 1, endocardial fibroelastosis with calcification 3, and valvular hematomas 7.[29]

Digestive

Studies in Germany indicated that atresia coli can be caused by unskilled rectal palpation of the amniotic sac between day 35 to 40 gestation.[88] However, recent studies postulated a genetic cause for atresia of the colon in Holstein calves.[96]

Large Body Cavities

Although scrotal, inguinal, and in particular, umbilical hernias are considered common in cattle, little is known

about their cause.

From 323,961 cases submitted to 12 U.S. and Canadian Veterinary College hospital clinics, 1,315 were diagnosed with congenital umbilical hernia; 705 with congenital inguinal hernia, and 57 with congenital scrotal hernia. Several breeds of cattle are at risk for one or both types of hernia.[38] Introduction of American Holsteins into Northern Germany has led to increased frequency of umbilical hernia.[95]

Immunodeficiencies of Cattle

Chediak-Higashi syndrome, an inherited disease of Hereford and Brangus cattle, is associated with a defect in cell structure resulting in abnormally large lysosomes. Clinical signs are photophobia, partial albinism, tendency to bleed, and recurrent pyogenic infections.[7]

Other Metabolic Defects

Congenital porphyria, transmitted as a simple autosomal recessive, has been reported in various breeds of cattle.[67] Deficiency of uridine-monophosphate synthase (dumps) is involved in embryonic mortality in Holstein cattle. The heterozygote status of dumps can be determined by biochemical tests.[89] Recently, leukocyte adhesion deficiency has been described in Holstein cattle as a cause of calf mortality.[90] The significance of these defects in beef cattle has not been established.

Reproductive System Defects

Various defects afflict the reproductive system. Rectovaginal constriction (RVC) in Jersey cattle is characterized by inelastic constriction at the junction of the anus, rectum, vestibule, and vulva. Rectal examination and artifical insemination are difficult. The male has anal stenosis. Affected cows have dystocia and are prone to develop periparturient udder edema, frequently followed by severe mastitis.[2,61,66,97]

Studies of RVC-affected, carrier and normal Jersey cattle identified differences in electromyograms from the external anal sphincter muscle. Immunohistochemical studies revealed type II collagen within the connective tissue of the external anal sphincter muscle in RVC-affected and carrier cattle.[97]

Genetics and breed dynamics of rectovaginal constriction were investigated in Jersey cattle using data from farmer-owned herds and experimental matings supported simple autosomal recessive inheritance. Computer analyses with a model estimated a slow decline in the frequency of the gene for rectovaginal constriction. Practical dynamics of the disorder in a breed registering 50,000 females and 2,000 males annually are given for current conditions and after 500 generations of selection.[72] Similar defects may be encountered in beef cattle or Jersey crosses.

Conclusions

Animal breeders and veterinarians are involved daily in improving animal health and production, their goal being to produce quality animals. Accurate diagnosis of diseases and defects caused by genetic factors is necessary before control measures can be effectively established. Such diagnoses involve understanding hereditary patterns of disease. Many different congenital defects (genetic, environmental, or unknown cause, or due to environmental-genetic interaction) have been identified in cattle. It is important to recognize that congenital defects are economically significant. Not only is diagnosis important but methods to control genetically-induced defects should be understood. Most breed associations have programs for controlling undesirable traits and genetic defects.

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