Congenital defects or diseases are abnormalities of structure or function present at birth. In cattle many of these defects of either genetic, environmental, or unknown cause—or due to environmental-genetic interaction—have been identified. The genetically caused diseases are mostly due to homozygosity of a simple autosomal recessive gene, meaning that the animal has received one defective gene from each of its parents.

It is desirable to recognize congenital defects which are of significance to cattle owners and breeders. Not only is diagnosis important, but methods to control genetically induced defects should be available. The most frequently encountered defects seem to involve bone, brain, muscle and skin. In this chapter the more significant genetic diseases in beef and dairy cattle are reviewed.

**Mulefoot.** The single most common and economically important disease in Holstein, Angus, Chianina, Charolais and Simmental cattle is syndactyly (mulefoot). This is nondivision or fusion of functional toes. It affects most frequently the right front foot followed by the left, then right hind and all four feet. The cow is not adapted to the single hoof and walking is painful and difficult for mulefoot animals. Frequently they die due to stress.

Syndactyly is a simple autosomal recessive gene. It is common in various breeds in the United States and Europe. Other species affected are swine, sheep and dogs. This ranks as the most important genetic disease in cattle in the United States.
Treatment is by control of carrier bulls in artificial insemination and natural breeding.

A method of early detection has been developed at Kansas State University. It combines superovulation, embryo transfer (two embryos for recipient), and early fetal cesarean section at 60 days of gestation—at which time the mulefoot condition can be easily recognized.

Cows may also be tested in this way: The cows are superovulated and then inseminated with semen from an affected syndactylous bull. The embryos are transferred to recipient cows (two per recipient) and then the fetuses are removed by cesarean section at 60 days of gestation. It takes 7 normal fetuses per cow or bull to declare the suspect animal clean (a non-carrier). As outlined above, prevention can be accomplished only by preventing the use of carrier animals in breeding.

Too Many Toes. Polydactyly, more toes than normal, also is a genetic disease—mostly in Simmental and Holstein cattle. It is transmitted as a polygenic (many genes with small effects). Usually
Normal 60-day fetus. Note normal cloven foot (arrows).

A 60-day fetus affected with syndactyly (mulefoot). Note abnormal uncloven front feet (arrows).

Polydactyly (more toes than normal) in a calf. Notice three toes developed instead of normal two toes. Affected calves have difficulty walking.
one finds 3 toes on both front feet. The affected animals have difficulties in walking. Spread of this defect is by carrier bulls. There is no treatment. Prevention and control is only by carefully recording defective calves and removing carrier bulls from service.

Tibial hemimelia is a bone defect in the hind legs of Galloway calves. It is due to a genetic factor, a simple autosomal recessive (with one abnormal gene from each parent). Affected calves lack development of a bone in the hind leg, the tibia. Thus, the calves are unable to rise and nurse. Other defects accompanying this defect are brain and reproductive abnormalities. There is no treatment for

“Snorter” dwarf with typical saddle nose, short legs and bloated belly. Dwarfs are rare today but once were a serious threat to livestock industry.

the condition except to prevent its occurrence by eliminating carrier animals from breeding service.

Besides leg defects there are various other abnormalities including the bone of the head and spinal column. Cleft lip and cleft palate may be hereditary defects in cattle but they need more careful studies.

The entire skeleton is affected in dwarfism, bulldog calves (chondrodysplasia), and marble bone disease (osteope-
"Bulldog" calf born dead. Notice abnormal face and short legs.

Leg bone split in half from calf affected with marble bone disease. Note lack of bone marrow cavity (a).
trosis). All these defects are genetic in origin.

Dwarfism is characterized by small size and a snorting sound while breathing. That led to the condition being referred to as snorter dwarfism. At one time it was the single most important disease in U.S. beef herds. There still are occasional cases. It is inherited as a simple autosomal recessive. Prevention and control is by carefully controlled breeding programs.

**Bulldog calves** are seen in the Holstein breed and various beef breeds. They are small, have head deformities, and frequently are born early. The legs always are very short. Severe abnormalities involve bones of the legs.

Bulldogs have a genetic cause and more studies are needed to clarify the mode of transmission. Spread of the disease is via carrier animals which implies again that control may be accomplished by identifying carrier animals and eliminating them from breeding programs.

**Osteopetrosis (marble-bone-disease)** has been described in black and red Angus calves. Occasional cases have been observed in Simmental and Hereford cattle. The disease is caused by homozygosity of a simple autosomal recessive gene.

This defect is characterized by small body size and weight. Calves are born 3 weeks prematurely and dead. There is a short lower jaw. All bones in the body of a calf affected with marble bone disease are hard and solid. There are no bone marrow cavities.

Spread of the disease occurs by carrier bulls and carrier dams. There is no treatment for this genetically transmitted disease. Control and prevention is only possible by diagnosing affected calves which identifies the sire and dam as carriers. Carrier animals have to be excluded from further use in breeding.

**Muscle defects** are next in importance. Crooked calf disease may affect any breed of dairy or beef cattle. Most commonly it has been diagnosed in the Charolais breed.

This defect is inherited as a simple autosomal recessive. A word of caution: plant poisonings such as lupine (silver or blue lupine) may cause a similar defect. However, a good veterinary examination usually can distinguish the genetic condition from the lupine problem.

Calves affected with the
genetic form of "crooked calf disease" (arthrogryposis) have all four legs in a frozen position (contracture). The legs cannot be moved and may cause problems during birth. Almost always a cleft palate is present. The defect can be spread only by carrier bulls and dams. These animals should be identified and eliminated to prevent occurrence of the disease.

An important muscular disorder is "double muscling" encountered in most U.S. beef breeds. It is considered a genetic defect due to homozygosity of a simple autosomal recessive gene.

External appearance of double muscling varies, the most noticeable being a round outline of the hind quarters. Muscles of the shoulder, neck, back and hind legs are heavy and separated by deep creases. The tail is attached higher than normal. Many double-muscled animals stand in a stretched position. Reproductive organs are small.

Spread of the disease occurs by breeding animals carrying the gene for double muscling. Control and prevention of the disease is by eliminating carrier animals.
Brain Defects

Congenital defects of the central nervous system are common and economically important. There is a lot of room for improvement of diagnosis and further studies are needed to arrive at a better understanding of these defects.

The single most important brain defect is internal hydrocephalus (water-on-the-brain), and occurs most commonly in beef cattle such as Herefords. Hydrocephalus is an inherited defect of simple autosomal recessive nature. It results from excessive fluid accumulation in the brain cavities.

The head may be domed or not. There are eye defects such as small eyes, and muscles are pale. Calves do not get up after birth and are unable to nurse. Some of the calves are soon dead or they die within a few days after birth. Spread of hydrocephalus is through using carrier bulls in herds where carrier dams are present. There is no treatment and control again is possible only by identifying carrier animals and eliminating them from breeding.

Congenital cerebellar disease is important in cattle. A disease afflicting a certain
part of the cerebellum (a brain area deterioration) has been described in calves. It is referred to as abiotrophy and occurs in Holstein, Hereford and Charolais calves.

This appears to be a simple autosomal recessive. Calves lose control over their movements at an age of 6 to 8 months. Spread by carrier animals should be prevented by identifying and eliminating carriers.

Hereditary neuraxial edema, another brain disease, has been seen in Hereford calves. It is considered a simple autosomal recessive. Calves are born unable to get up and nurse. Hand clapping or touching the calves can send them into violent spasms. Calves ultimately die or are destroyed. The spread of neuraxial edema is by carrier animals and only their identification and removal from service prevents and controls this disease.

**Spinal Cord Ills**
Spinal cord diseases are just starting to emerge as genetic problems in calves. Further studies are needed to better understand and control them.

Spastic and paralytic diseases, common in cattle, are of genetic origin. Their spread, control and prevention can be accomplished only after further studies. Spastic paresis is characterized by one or both hind legs stiffening up and being kept off the ground, preventing afflicted animals from moving about.

An important brain and spinal cord disease afflicts Brown Swiss cattle between 6 to 8 months of age. It is referred to as the weaver condition. The disease most likely is transmitted as a simple autosomal recessive.

Calves become sick at about 6 months of age. Weavers then have an unsteady gait, will frequently stumble and fall to one side. The disease progresses during the next few months. Most cases are misdiagnosed as back injuries or arthritis. Weavers finally go down and die or have to be destroyed.

Spread of the weaver condition is via carrier animals. There is no treatment and control and prevention is by adjusting breeding programs.

Storage diseases of genetic nature are beginning to emerge in cattle. Material accumulates inside cells mainly in the brain.

One such disease has been identified in Angus cattle in the United States. Called mannosidosis, it is transmitted as a simple auto-
somal recessive. Angus calves may be affected at birth but usually come down with the disease between 6 to 8 months of age. They quit growing, have difficulties walking, and die within the first 12 months of life.

This is due to an enzyme deficiency. Since the spread is by carrier bulls and dams, the enzyme deficiency can be used to detect carriers. Carriers have half the normal enzyme activity. This fact can be used to identify carriers and thus control and prevent the disease.

Eye, Skin Defects

The most common eye defects in cattle are lack of eyes or small eyes. This may be a genetic problem but needs much more study. A defect which is polygenic (many genes involved) accounts for the transmission and spread of feather-eyes (dermoid) in dairy and beef cattle; skin on the eye surface leads to partial or complete blindness. Feather-
Calf affected with anophthalmia (lack of eye development). Note lack of eye.

Feather-eye (dermoid) in Hereford calf. Note skin covering front of eye, causing blindness.

Albinotic color deficiency in Angus heifer. Note dilute coat color instead of deep rich black.

Albinotic color deficiency in Angus calf. Note white iris color.
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eye, as well as any other defect of polygenic nature, can be controlled only by carefully keeping records of affected calves on a breed basis, thus identifying possible transmitters of the defect.

Genetic defects of the skin have not received enough attention and study. However, a few are important and are presented here.

Albinism has been diagnosed in various breeds of cattle. It always is genetic, usually a simple autosomal recessive.

A new albinotic color deficiency has been identified recently. It is inherited as a simple autosomal recessive.

Hereford calf affected with moderate degree of hairlessness (hypotrichosis).

The defect imparts to Angus cattle a brown hair instead of the typical black. The eyes are white, thus causing the animal pain and reduction of vision in bright light. Spread is again due to normal carrier animals which must be identified for control.

Skin fragility has occurred in various beef breeds. It is passed on as a simple autosomal recessive. The skin is very fragile and tears away from even slight injury. Hematomas (accumulation of blood) and infections are com-
mon. Spread is by carrier animals.

A common skin defect is *epitheliogenesis imperfecta* in Holstein, Shorthorn and Angus calves. A simple autosomal recessive accounts for this defect. Areas of skin turn up missing on the legs and head, allowing infections to gain entrance. The calf usually dies in the first 3 weeks of life.

Use of carrier bulls and dams spreads this defect. Since the carrier animals are normal, only occurrence of the defect reveals the carrier status. Their removal from breeding prevents further cases.

Protoporphyria in Limousin cattle is really a metabolic disease but the main sign is sunburn. It is inherited as a simple autosomal recessive. Inflammation of the skin occurs on the head, neck and back. Skin of the ears and the muzzle are affected. Normal appearing carrier cattle may be detected by an enzyme test, and control instituted with this test.

Hypotrichosis (severe loss of hair) in Hereford calves is a genetic disease caused by homozygosity of a simple autosomal recessive gene. Hypotrichosis varies from mild to severe. Affected calves suffer from adverse weather conditions and also are more susceptible to fungal and bacterial skin diseases.

The disease is seen throughout the United States and is spread by normal unaffected carrier animals. Control and prevention require identifying the normal carriers and eliminating them.

**Internal organs** such as the heart do not escape genetic defects. Much more research on these defects is needed to help establish their nature and significance.

The most important reproductive system defect in the United States is rectovaginal constriction (RVC) in Jersey cattle. RVC affects the anus and part of the reproductive system in Jersey heifers. These areas are inelastic and constricted, leading to difficulties during artificial breeding and problems with birth, usually leading to cesarean section. In addition, RVC cows frequently develop severe udder edema followed by mastitis. RVC is of considerable economic significance. Spread occurs by using normal and affected carrier bulls.

Identification by diagnosing affected offspring helps to control the disease. The bulls identified as transmitters (carriers) may be removed from service.