# Genetic defects in Farm Animals

## 1- Genetic Defects in Sheep

**By: Kris Sweiter**
**Erica Gacsala**
**Humberto Esquivel**

Genetic defects refer to deformities that exist at birth. They occur sporadically and rarely contribute to major losses of lambs in flocks. There are more than 30 known or suspected genetic defects of sheep. Many are lethal. Others are semi lethal, because the effect is crippling but death is not inevitable, although a proportion do die. Some common genetic defects are entropion (inverted eyelids), cleft palate, parrot mouth (undershot jaw, cryptorchidism (one or both testicles retained in the abdomen), hernias, abdominal impaction, and spider lambs, and even prolapses.

### Causes of Genetic Defects

Chromosomes inherited from parents determine an animal's genetic make-up. There are many genes in each chromosome. Genetic abnormalities occur when genes are missing, in excess, mutated or in the wrong location (translocation). A few genes can directly cause an abnormality, however, these are rare. Usually, these genes are recessive, meaning two must be present to cause an abnormality. Both parents must be carriers of the gene for a calf to be abnormal. In this case, only one of every four offspring will be abnormal. Two will be carriers and one will be normal.

*Certain conditions show that an abnormality is likely to have a genetic origin*:

1. The abnormality is more common in a group of related animals.
2. The symptoms are similar to those of an abnormality identified through test matings. Study of an animal's chromosomes using blood samples can identify several genetic defects.

**Below are some of the most common genetic defects in sheep**.

**Jaw defects** Jaw defects are present in almost all breeds of sheep and are associated with failure of the incisor teeth to properly meet the dental pad. A jaw is undershot if the incisor teeth extend forward past the dental pad; it is overshot if the teeth hit in back of the dental pad (this condition is known as parrot mouth). Cull sheep with either of these genetic defects. If the sire and dam can be identified, remove them from the flock.

**Overbite:** (overshot, parrot mouth, class, overjet, mandibular branchygnathism)
In this condition the upper jaw is longer than the lower jaw. There is a gap between the upper and lower incisors when the mouth is closed. Some lambs that are born with an overbite might self-correct if the bite is no larger than the head of a wooden match. In most breeds of sheep the bites are "set" by the time a lamb is a few months old. An overshot bite will rarely improve after the lamb reaches maturity.

**Underbite:** (undershot, reverse scissors bite, prognathism)
In this condition the lower jaw is longer than the upper jaw. If the upper and lower jaw meets each other edge to edge, the bite is referred to as an even or level bite. If your lamb has either an overbite or an underbite, it will not be able to properly nurse, get enough nutrition, or even eat from the creep feeder with the other lambs as seen below.

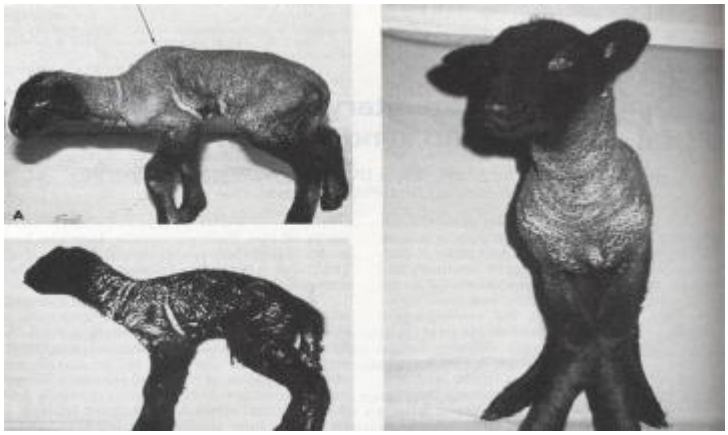
**Rectal prolapse:** Rectal prolapse is a serious defect most commonly associated with the meat-type sheep. It is most common among lambs fed a high-concentrate ration. It is believed that this weakness is due to inheritance. This condition is sometimes corrected by surgery, but affected animals often continue to prolapse after surgery. Cull from the flock breeding sheep in which this occurs.

**Cryptorchidism:** Rams with one or both testicles retained in the abdomen, or not descended fully into the scrotum are cryptorchids. Cryptorchidism presents itself in one of two forms: 1) unilateral cryptorchidism - normal descent of only one testicle, 2) bilateral cryptorchidism - retention of both testicles. Unilateral cryptorchid lambs are usually capable of breeding, whereas bilateral cryptorchids are sterile. The condition usually is inherited as a simple recessive trait. There seems to be some association between this condition and the polled characteristic found in some fine-wool rams. Purebred breeders should make every effort to eliminate this condition. In spite of the fact that bilateral cryptorchid lambs are sterile, both bilateral and unilateral cryptorchids should be castrated, to reduce the risk of possible future complications. Unilateral cryptorchids should never be used in a breeding program.

**Inverted eyelids:** Inverted eyelid (entropion) is widespread among most breeds of sheep. This trait is highly heritable. Inverted eyelids are a "turning in" of the margin of the eyelid and therefore bringing the eyelashes into direct contact with the cornea. This contact creates an irritation, making it necessary for the animal to blink constantly. As the animal blinks, it is compounding the problem by scraping the eyelashes across a more extensive area of the eye. This extreme irritation

if left unattended, can eventually cause blindness. The condition may be noted at birth and treated at that time. Entropion should never be left to take care of itself. If left untreated, the condition could cause sore watery eyes, infection, ulcers on the cornea and even blindness. Entropion condition requires surgical correction by a veterinarian. One method of treating this condition is to clip a metal suture to the center of the affected eyelid. Gather enough skin under the clip in a vertical direction to hold the lid away from the eye. The clip can be left in place for several days. Mark the affected lambs and do not allow them to enter the breeding flock.

(Normal, Healthy Eyes)



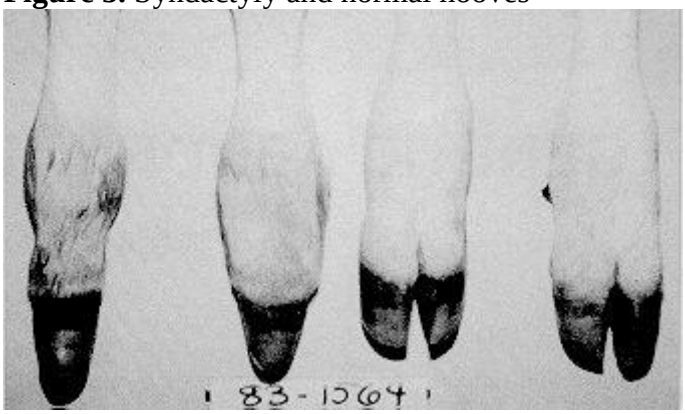
**Spider Lamb Syndrome** (SLS) or ovine hereditary chondrodysplasia is a genetic disorder causing skeletal deformities in young lambs. These defects commonly include abnormally long, bent limbs, twisted spines, shallow bodies, flattened rib cages, and long necks. The syndrome is inherited as a genetic recessive disorder meaning that affected lambs must inherit the mutation from both their parents. Because of this inheritance pattern, the identification of genetic carriers of SLS has been difficult without the use of progeny testing. The presence of SLS in several breeds has prompted breeders to divide pedigrees into two categories, "gray-pedigreed" animals having ancestors that have produced spider lambs, and "white-pedigreed" animals having ancestors that have never produced affected lambs.

# 2- Genetic Defects in Cattle

## Syndactyly (Mulefoot):

Syndactyly refers to the fusion of the two toes of the foot. Caused by **a recessive gene**, mulefoot most often affects the front feet. This condition occurs in the Aberdeen Angus breed.

**Figure 5.** Syndactyly and normal hooves



## Hypotrichosis (Hairlessness)

Hairlessness occurs in several breeds of beef cattle. It expresses itself as complete or partial loss of hair. Calves are often born with no hair but will grow a short curly coat of hair with age. Affected individuals are prone to environmental stress (cold and wet) and skin infections are more prevalent. A recessive gene causes hairlessness.

**Figure 1.** Partial Hypotrichosis

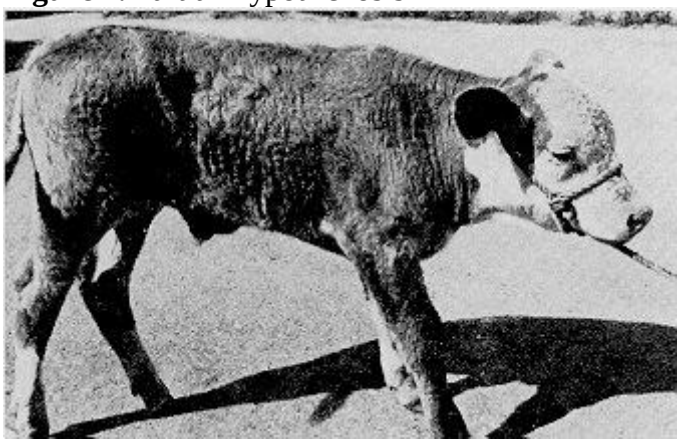
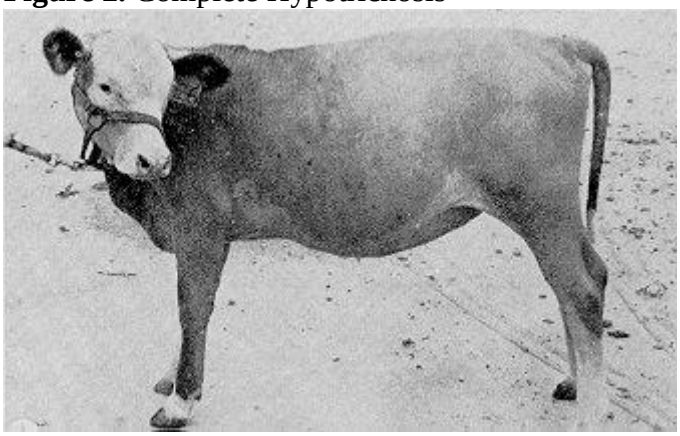


**Figure 2.** Complete Hypotrichosis



# Beef Cattle Handbook

BCH-1900 Product of Extension Beef Cattle Resource Committee

## Congenital Defects in Cattle

R. R. Schalles, Department of Animal Sciences and Industry, Kansas State University
H.W. Leipold, Department of Pathology, Kansas State University
R. L. McCraw, Department of Animal Science, North Carolina State University

There are many undesirable traits that show up in beef cattle. These range from poor performance and structural unsoundness to semi-lethal and lethal diseases. Many of these are caused by varying amounts of genetic influence; others are caused by environmental conditions or an interaction between the animal's genetic makeup and the environment in which it lives. Congenital defects are present in all breeds of cattle. In most herds, they are rather uncommon; however, occasionally the frequency within a herd will be high enough to be of considerable economic importance. Congenital defects are abnormalities of structure or function present at birth and may account for a high percentage of calf losses from just before to just after calving. The frequency of congenital defects ranges from less than 1 to over 3 percent within herds. Cattle breeders should be conscious of potential defects and investigate animals exhibiting unusual characteristics.

### Causes

The cause of many congenital defects is unknown, but some are inherited. The most common inheritance pattern is as a simple recessive trait. The defective calf receives a recessive gene from its sire and one from its dam. A few congenital defects are known to be caused by genes with incomplete dominance and a few are caused by two or more sets of genes.

Genetically caused congenital defects usually run in families. The parents of a genetically defective calf will generally have at least one ancestor in common. When more than one genetically caused defective calf is born in a herd in the same calving season, their dams are usually related (for example, half sisters) and are sired by the same bull. A change in the breeding program is required to correct this situation.

Many congenital defects are caused by environmental factors. These include the level of nutrition, excess or shortages of certain nutrients, toxic plants or other toxic substances, infectious diseases, and extremes in temperature during pregnancy. Most environmentally caused congenital defects will occur during a short period of the calving season, from cows that were managed as a group. After proper diagnosis, a change in management is necessary to correct these conditions.

### Diagnosing the Cause

To determine the cause of defects, the breeder must have good records and know why every calf dies. Breeding records which include sire and dam of each calf and breeding date are needed. Blood typing or DNA typing of the calf and possible parents can be used to help determine parentage. The calf must be alive and at least one month old when the blood sample is obtained for blood typing. Management records should include which cows were in groups during each time period. Most breeders have a list of which cows are in each pasture. A date in and out of the pasture usually will help identify problems. Feed analysis reports, toxic plants present, and herd health and vaccination programs are also of value.

Knowing the cause of death is important in controlling diseases as well as congenital defects. The cause of some deaths will be obvious, others will be much more difficult. If the breeder does not know the cause, they

Figure 1. Dwarfism. Notice short, blocky appearance with deformed nose causing labored breathing (snorter dwarf).

should get help from their local veterinarian or state diagnostic laboratory. The breed associations, AI organizations, and extension personnel usually can help contact diagnostic personnel also. It is very important to act fast. Tissue of a dead calf will deteriorate very fast in warm weather, especially the nervous system. If the calf cannot be delivered to the laboratory within a few hours after death, it should be well cooled before transportation. The best diagnosis can be obtained from a calf delivered alive. Call the diagnostic laboratory ahead so they can give instructions on how to handle the calf and be ready when it arrives. If a defect is diagnosed as genetically caused, the breeder of the animal and the breed association should be notified. The responsibility for control of genetic defects rests primarily with the purebred breeders and breed associations. Good diagnosis from one dead calf may save many others.

## Genetic Defects

1. *Dwarfism.* There are several types of dwarfism caused by both environment and genetics. Each of the three genetically caused dwarfisms discussed here are different traits caused by different sets of genes.
   a. Snorter dwarfism causes short, blocky appearance with deformed bone growth in the nasal passages which causes difficulty in breathing. Inherited as a simple recessive trait (Figure 1).
   b. Long head dwarfism causes small size but does not affect the bone growth in nasal passages. Inherited as a simple recessive trait.
   c. Compress dwarfism is inherited as incomplete dominance. An individual with one compress gene and one normal gene has an extremely compressed body conformation. The individual with two compress genes is a dwarf and the calf dies at or soon after birth.
2. *Water head* (internal hydrocephalus). Excess fluid is present in the brain (Figure 2). Calves are usually born dead or die shortly after birth. Environmental factors can cause the disease, as well as being inherited as a simple recessive trait.
3. *Marble bone* (osteopetrosis). The calves are usually born dead two to four weeks early. Bones are solid

Figure 2. Waterhead (internal hydrocephalus). Calf was born dead. Notice dilation of internal aspects of cross-section of this brain.

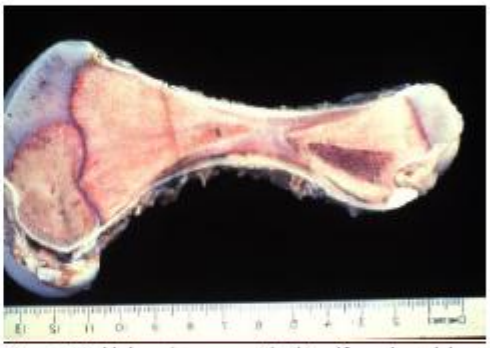


Figure 3. Marble bone (osteopetrosis). This calf was aborted three weeks before its term. Notice thick bone with no bone marrow cavity.

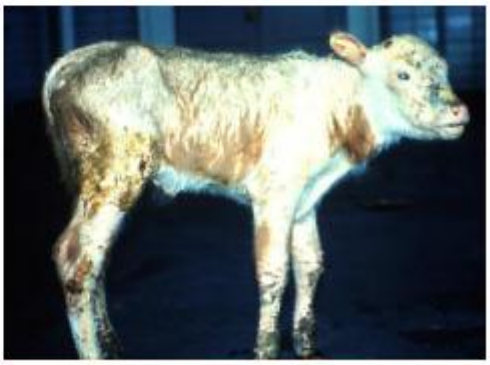


Figure 4. Hairlessness (hypotrichosis) in a calf.

and do not contain marrow, making them very brittle and easily broken (Figure 3). Inherited as a simple recessive trait.

4. *Hairlessness* (hypotrichosis). Partial to almost complete lack of hair (Figure 4). Hair develops and is lost so an affected animal will vary somewhat in expression from month to month. Inherited as a simple recessive trait.
5. *Rigid joints* (arthrogryposis). Many environmentally caused forms appear but one form is inherited as a simple recessive trait. The joints of all four legs are fixed symmetrically and a cleft palate is present (Figure 5).
6. *Extra toes* (polydactyly). One or both front feet are usually affected, but all four may have the outer dew claw develop into an extra toe (Figure 6). At least two sets of genes are involved in the inheritance of this trait.
7. *Mulefoot* (syndactyly). The two toes are fused together to produce only one toe. The front feet are most often affected, but all four may be affected (Figure 7). These cattle cannot tolerate hot temperatures. Inherited as a simple recessive trait.
8. *Weaver calf* (progressive bovine myeloencephalopathy). Calves start developing a weaving gait at 6-8 months and get progressively worse until death at 12-20 months (Figure 8). Inherited as a simple recessive trait.
9. *Photosensitivity* (protoporphyria). Animals are sensitive to sunlight and develop scabs and open sores when exposed to sunlight (Figure 9). The liver is also affected and the animals may suffer from seizures. Inherited as a simple recessive trait.
10. *Bulldog* (achondroplasia). This trait is inherited as an incomplete dominant. The homozygous may be aborted dead at 6-8 months gestation, and has a compressed skull, nose divided by furrows and shortened upper jaw, giving the bulldog facial appearance. The heterozygous calf is small and heavy-muscled (Figure 10).
11. *Double muscling*. Animals are extremely heavily muscled (Figure 11). However, considerable variation exists in the expression of this trait. Inherited as a simple recessive trait.
12. *Parrot mouth* (brachygnathia inferior). One type of parrot mouth (Figure 12) is inherited as a simple recessive trait. The more common cause of teeth and denture pads not meeting is a quantitative trait caused by several sets of genes. This can cause either an under or over shot jaw with varying degrees of expression.
13. *Cryptorchidism*. One or both testicles fail to descend into the scrotum. Inherited as a sex limited trait and probably involves at least two sets of genes.
14. *Prolonged gestation*. The fetus fails to trigger parturition. Parturition must be induced or the calf removed. The calf is often extremely large and often dies. Inherited as a simple recessive trait.
15. *White eyes* (Oculocutaneous Hypopigmentation).

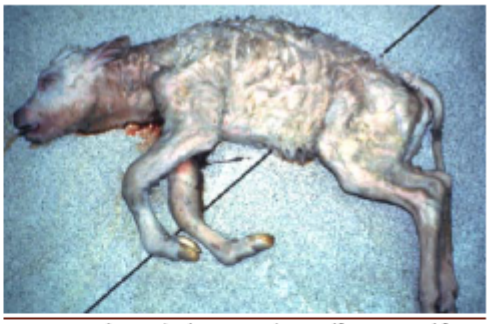


Figure 5. Rigid joints (arthrogryposis) in a calf. Notice rigid front legs. These calves have a cleft palate.

Hair coat is a bleached color and the iris is pale blue around the pupil with tan periphery. Inherited as a simple recessive trait.

Many other genetically caused undesirable traits are known. The beef cattle geneticist at your land grant university will have knowledge of most of them and will be able to help if a problem arises.

Many abnormal conditions are not genetically caused. Two headed calves and calves with extra legs are caused by mistakes in development and not the genetic makeup of the individual or its parents. Freemartin heifers are caused by circulation of the male twin's hormones through the developing female fetus. Some hydrocephalus can be caused by BVD (bovine virus diarrhea) infection during pregnancy. Crippled-calf disease is caused by the cow eating lupines between days 40 and 60 of pregnancy. Flexed pasterns (contracted flexor tendons) is usually caused by a large fetus

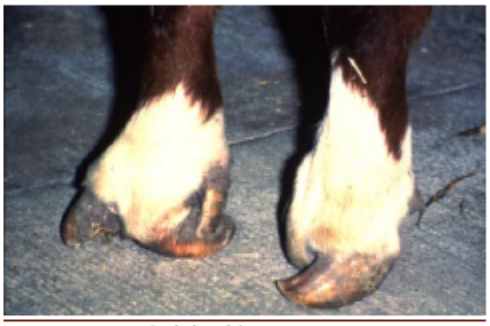


Figure 6. Extra toes (polydactyly).

developing in a small uterus. However, both crippled-calf and flexed pasterns can also be genetically caused, inherited as simple recessive traits.

## Controlling Genetic Diseases

The best control of genetic diseases is to avoid animals that carry these genes. Bulls or semen should be purchased from reputable breeders, produced by parents who are not known to carry undesirable genes. Long

Figure 7. Mulefoot (syndactyly). Notice single toe as compared to the normal front foot.

established inbred lines that have not recently produced genetic undesirables are usually quite safe. Commercial producers who use a crossbreeding system rarely have a problem.

The elite purebred breeder or owner of AI bulls may wish to test for simply inherited traits before bulls or donor cows are heavily used. If the undesirable trait is dominant, no test is needed since the animal would show the trait even if only one dominant gene is present. If the trait is inherited as incomplete dominance, the individual that has only one undesired gene can usually be identified and testing is not needed. Testing is usually useful only when the trait is inherited as a simple recessive trait.

When only one nonlethal, undesired, recessive trait is of concern, the least expensive test is to mate the animals to ones having that undesired trait. For example, if

Figure 8. Weaver calf (progressive bovine myeloencephalopathy)/ Notice unsteady posyure of this animal.

horns are not desired, the polled animal to be tested would be mated to horned animals to produce at least 7 calves (P>.99). If any horned calves are produced, the polled animal has one gene for horns.

If the undesired trait is lethal, the dead animals cannot be used to make the test. If only one lethal trait is of concern, the test should be made using animals that have produced calves with this lethal trait. At least 16 calves should be produced from these matings (P>.99). If a calf is produced that has this lethal trait, the tested animal has one recessive gene for the trait.

If the breeder is concerned about identifying all recessive traits, at least 36 progeny from sire-daughter or mother-son matings should be produced (P>.99). This test is time consuming and expensive and only truly outstanding animals will be able to pay for it. All calves from these matings must be observed very closely and all recessive traits recorded. The tested animal will have a recessive gene for any recessive traits observed.

### What to Do with Carriers

An animal that has one undesirable recessive gene may have thousands of very desirable genes. The animal's desirable genes should be weighed against its undesirable genes. If the desirable genes can be found in other

Figure 9. Photosensitivity (protoporphyria). Notice skin lesions due to sunlight.

animals without the undesirable gene, carriers should be slaughtered and replaced. When the production traits are superior, these animals can be used in a crossbreeding program to produce beef. Heifers should not be kept for breeding. If the individual is extremely superior in production traits, a superior son can be produced that does not carry the undesirable gene. The outstanding carrier animal would be mated to a small group of very outstanding individuals. The best two to four sons produced would be selected and used in test matings to known carrier cows. The best son that does not carry the undesired gene would then be used and all carriers slaughtered. This would take several years and only truly superior individuals could justify such a procedure.

In most cases, the animal that carries the undesirable recessive gene should not be used to produce breeding animals. Daughters should be worked out of the herd and replaced with superior animals that do not carry undesirable genes. Purebred breeders should work with their

breed associations, extension and university personnel, and veterinarian to eliminate and avoid problems.

### Ethical and Legal Considerations

Serious ethical and legal problems are involved in selling known carrier cattle or progeny of known carriers. A seedstock producer in this position should be completely honest with the buyer. It is doubtful that he should sell possible carriers, under any circumstances, to a youngster or to someone who is just getting started in business and may not have the knowledge to understand the consequences of using offspring from known carriers. Selling carriers without informing the buyer will ultimately reduce the confidence that buyers have in the breeder and may eventually reflect negatively on the entire breed.

Figure 10. Bulldog calf (achondrodysplasia).

Figure 11. Double muscled calf on pasture. Notice outline of hind legs and deep creases between muscles.

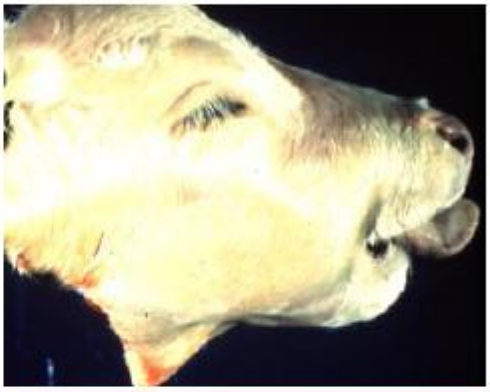


Figure 12. Parrot mouth (brachygnathia inferior). Short lower jaw in a calf.

Authors:
R. R. Schalles, Department of Animal Sciences and Industry, Kansas State University
H.W. Leipold, Department of Pathology, Kansas State University
R. L. McCraw, Department of Animal Science, North Carolina State University

This publication was prepared in cooperation with the Extension Beef Cattle Resource Committee and its member states and produced in an electronic format by the University of Wisconsin-Extension, Cooperative Extension. Issued in furtherance of Cooperative Extension work, ACTS of May 8 and June 30, 1914.

**BCH-1900** Congenital Defects in Cattle

# 3- Genetic Defects in goats

## mucopolysaccharidosis IIID or G-6-S

This defect's full names are mucopolysaccharidosis IIID, or G-6-Sulfase deficiency, and it is usually referred to as G-6-S. It was first identified in 1987 at Michigan State University, and subsequently the researchers tested nearly a thousand goats in Michigan and concluded that about 25% of Nubians carry this gene. All cases are the result of a single mutation, and appear to be confined to Nubians and their crosses; other breeds were tested initially and they do not have this particular defect.

The affected goats lack an enzyme (G-6-S) and this results in a variety of symptoms of varying severity. The main symptom exhibited by affected goats is failure to grow. Sometimes the kid is smaller than normal at birth, and grows slowly. Some breeders have reported kids which grew normally for the first three months and then stopped growing. Other affected goats grow to what appears to be normal size but is in fact small for the particular bloodlines. They lack muscle mass, appear "slab-sided", sometimes with blocky heads. Immune function appears to be compromised, and sometimes they become deaf or blind. The longest-lived goat known to be G-6-S affected died at just under four years of age, and death is usually due to heart failure. Unfortunately affected animals can and do grow up to breed, although they often experience reproductive problems.

## Beta-Mannosidosis

Beta-mannosidosis is a genetic defect of Nubians which is similar to G-6-S in its mode of transmission and in the method recommended for its management. Like G-6-S, it is confined to Nubian goats, it is caused by a simple recessive gene, and a DNA test is available to distinguish normal, carrier, and affected animals. As for G-6-S, the optimal management strategy, the one which offers the best balance between getting rid of the bad gene and saving the good genes, is to use only normal bucks.

Unlike G-6-S, beta-Mannosidosis is rapidly fatal and no affected goats grow up to breed. For that reason the incidence of this defect is lower than the incidence of G-6-S, with only about 13% of the population being carriers.

An affected kid is born with what looks like cerebral palsy. The kid is unable to stand or to hold up its head, and it shakes if it tries to do anything (intentional tremor). If it tries to reach the nipple it shakes violently and fails, but if the nipple is put in its mouth it calms down and is able to suck. It may also have skeletal deformities, and twitching eye movement and be deaf, but these symptoms are more obscure and variable.

The cause of these difficulties is the lack of an enzyme which normally removes certain sugars from the cells. These sugars accumulate in the cells, and the cells of the nervous system show the effects first. Even with the best care the kid will die within a few weeks at the most.

Unlike with G-6-S, it is immediately obvious that the affected kid is defective, although its difficulties might be mistaken for some sort of birth trauma or oxygen deprivation. At least there is no possibility of bad feelings resulting from the inadvertent sale of a defective kid.