How Did This Happen?

by Dr. Debra K. Aaron, Professor, Department of Animal and Food Sciences, University of Kentucky

A lamb is born and, much to the producer's dismay, has a defect that will decrease its market value or, even worse, will affect its chances of survival. Maybe the lamb looks normal at birth, but exhibits some deleterious condition shortly thereafter. What happened? Is it a result of genetics or environment or a combination of both?

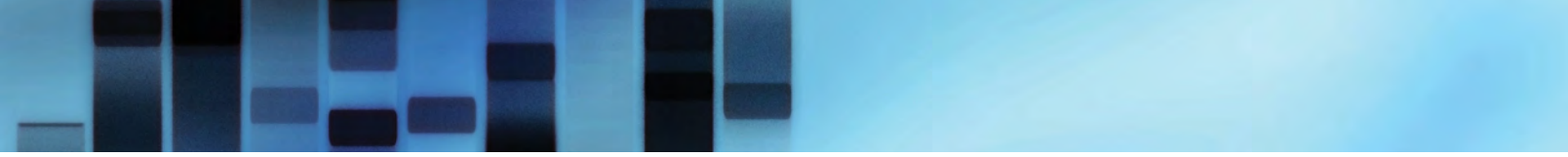
> A defect is defined as any characteristic that impairs the producing ability or the possibility of survival of the animal (Sheep Production Handbook, 2002 Ed., Vol. 7). Included may be sub-lethals that result in severely impaired survivability or lethals that result in death of the animal.

Serious hereditary defects are normally of low incidence as the affected animals are either culled or die before reaching the age of reproduction. Thus, they do not leave offspring. Also, any breed or flock with a high incidence of sub-lethals or lethals would not be acceptable to sheep producers.

Determining the Cause of Defects: Genetics or Environment?

When defects occur in a flock, the producer must objectively determine whether it is the result of genetic, environmental or both genetic and environmental factors. Considering the following questions can aid in determining if the defect is of genetic origin:

1. Were affected lambs all of one breed or by one sire? Generally, a defect is less likely to be hereditary if it occurs in more than one breed or in more than one sire progeny group. The former assumes breeds do not share a common origin (as do composite breeds) or have not been intermingled through crossbreeding; the latter assumes sires are unrelated. Also, genetic defects are uncommon in crossbred populations if no occurrences have been noted in the purebreeds contributing to the cross.
2. If the defect is present among one sire's offspring, are all lambs affected? Are the dams related? If most of the affected lambs are from related ewes, producers should be suspicious of a genetic cause. If it occurs in nearly all lambs sired by a ram and the dams are unrelated, the producer should look for environmental causes.
3. Is the defect present in lambs produced by the mating of closely related individuals; in other words, by inbreeding? Inbreeding results in increased frequency of traits caused by recessive genes. (See "Is Inbreeding Good or Baad?" in Genetically Speaking, *HoofPrint*, Volume 9, Fall 2012.) As will be described later in this article, most, though not all, genetic defects are inherited as simple recessives.
4. Were dams of the affected lambs under similar environmental, nutritional or management conditions during gestation? A genetic cause is unlikely if the affected lambs were born during the same lambing season while lambs born during other lambing seasons, sired by the same ram and from ewes



of similar breeding were free of the defect.

5. Are symptoms similar to those of a defect that has been identified through progeny testing or study of an animal's chromosomes using DNA (genetic) tests? Several abnormalities do not yet have a genetic test. For these, an animal (ram or ewe) can be confirmed as a carrier of the defective gene if and only if it produces an affected offspring. It is important to realize that some environmental factors can produce conditions similar to some genetic defects. However, genetic testing, when available for the defect in question, can confirm whether the defect is due to genetic or environmental causes.

Environmental causes (for example, diet, equipment, housing, management) can be corrected quickly; once the cause is identified, it can be removed and the problem will be solved. Genetic causes require longer-term solutions.

Inheritance Example

Genetic defects are a result of random mutations in the sheep genome (that is, the complete set of the sheep's DNA, including all of its genes). Genes may be missing, in excess, mutated or in the wrong location. Most defects that are currently monitored in the U.S. sheep population are thought to be **autosomal recessive**. This means that two copies of the mutated gene are needed for the animal to be affected. It also means that the gene responsible for the defect may remain hidden in a flock or may occur at a low incidence for a long time.

The following is an example of how Dermatosparaxis, a recessive lethal mutation found primarily in White Dorper sheep and their crosses (and described in more detail later in this article), is inherited.

The genetic model is as follows:

NN	Normal
NM	Normal (A Carrier)
MM	Affected

If both members of a gene pair are the same, it is referred to as "homozygous." If they are different, then it is referred to as "heterozygous."

If a heterozygous normal ram (NM, a carrier) is mated to a homozygous normal ewe (NN), the resulting offspring would be 100% phenotypically normal with 50% of them homozygous (NN) and the other 50% heterozygous (NM). This is illustrated by the following example:

	Ewe	
Ram	*N*	*N*
N	NN	NN
M	NM	NM

If two heterozygous (NM; a carrier as noted previously) animals are mated (see example below), there is a 25% chance that the lamb will be affected (MM), a 25% chance it will be homozygous normal (NN), and a 50% chance it will be a carrier (NM). The only time an affected lamb can be produced is when two carriers are mated, and even then the odds are that the lamb will be normal (NN or NM) 75% of the time.

	Ewe	
Ram	*N*	*M*
N	NN	NM
M	NM	MM

Common Genetic Defects in Sheep

There are more than 30 known or suspected genetic defects in sheep. Some common non-lethal defects include jaw defects, cleft palate, hernias, abdominal impaction, prolapses, entropion (inverted eyelid) and cryptorchidism. These defects do not necessarily result in death of affected lambs, but they greatly reduce productivity. Dwarfism is a defect that may be classified as sub-lethal because affected lambs seldom live to maturity. Spider Lamb Syndrome and Dermatosparaxis are two hereditary defects that generally result in death of affected lambs (classified as lethal or sub-lethal) and, thus, cause serious economic loss in affected flocks.

Characteristics of common genetic defects are summarized in Table 1. More detailed descriptions of some of them are provided in the following paragraphs.

Table 1. Some common hereditary defects that affect market value or survival in sheep.

Genetic Abnormality	*Primary Breed(s) of Incidence*	*Lethal or Non-lethal*	*DNA Test Available*
Jaw defects Overbite (parrot mouth) Underbite (monkey mouth)	Most breeds	Non-lethal	No
Cleft palate	Most breeds	Non-lethal	No
Rectal prolapse	Meat-type breeds	Non-lethal	No
Entropion (inverted eyelids)	Most breeds	Non-lethal	No
Cryptorchidism	Most breeds	Non-lethal	No
Dwarfism	Texel and most other breeds	Sub-lethal	No
Spider Lamb Syndrome	Suffolk, Hampshire and other blackfaced breeds	Lethal or sub-lethal	Yes
Dermatosparaxis	White Dorper	Lethal or sub-lethal	Yes

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 **overshot** (Photo 1) if the incisor teeth hit the back of

Photo 1. Sheep with overshot jaw (parrot mouth).

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the dental pad resulting in a gap between the dental pad and lower incisors. This condition is commonly called **parrot mouth**. Some lambs that are born with a slight overbite might self-correct. In most breeds of sheep, however, bites are fixed by the time lambs are a few months old. An overshot bite will rarely improve after the lamb reaches maturity. A jaw is **undershot** if the teeth extend forward past the dental pad. In this condition the lower jaw is longer than the upper jaw. This is commonly called **monkey mouth**. Lambs with jaw defects will not be able to nurse properly or eat from a creep feeder. Growth will be stunted because they are not able to get enough nutrition. Some lambs may die.

Cleft Palate

In sheep, cleft palate or cleft lip is hereditary in nature although maternal nutritional deficiencies and some viral infections during gestation have also been implicated. Affected lambs have difficulty nursing and milk may drip from the nostrils when the newborn attempts to nurse. Respiratory infection due to aspiration of milk is common. Affected lambs usually do not live. In Texel sheep, a syndrome of bilateral cleavage of the lip with accompanying defects of the maxilla (bones that form the upper jaw and palate of the mouth) has been reported to have an autosomal recessive mode of inheritance.

Rectal Prolapse

Rectal prolapse (Photo 2) is a serious defect most commonly associated with meat-type breeds of sheep. It occurs most frequently among lambs fed a high-concentrate diet. The existence of breed and sire differences indicate rectal prolapse is hereditary; however, this condition is largely due to environmental influences. Extremely short docking and heavy feeding, especially under dusty feedlot conditions where coughing may be a complicating factor, are factors known to contribute to this problem. Rectal prolapse can sometimes be surgically corrected, but affected animals often continue to prolapse after surgery. Most affected animals do not survive or are slaughtered. None should ever be retained for breeding purposes.

Photo 2. Lamb with rectal proplapse.

Entropion (Inverted Eyelids)

Entropion, which is more commonly known as inverted eyelids, is widespread among many breeds of sheep. This condition is highly heritable; however, the exact mode of inheritance is is believed to be rather complex. It can also be aggravated by environmental conditions.

Inverted eyelids are a "turning in" of the margin of the eyelid. As a result, the eyelashes are in direct contact with the cornea. This contact creates an irritation, making it necessary for the lamb to blink constantly. As the lamb blinks, the problem is compounded; the eyelashes scrape across a more extensive area of the eye. This irritation, if left unattended, can eventually cause blindness.

Entropion may be noted at birth and treated at that time. Treatment is relatively easy and effective. It should never be left to take care of itself. If left untreated, the condition could cause sore watery eyes, ulcers on the cornea and even blindness. Affected lambs should be identified or marked and should not enter the breeding flock.

Cryptorchidism

Rams with one or both testicles retained in the abdomen, or not descended fully into the scrotum, are called cryptorchids. Rams with normal descent of one testicle (unilateral cryptorchidism) are usually capable of breeding. Rams with retention of both testicles (bilateral cryptorchidism) are sterile. Cryptorchidism is 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 (Merino, Rambouillet) rams. Purebred breeders should make every effort to eliminate this condition. In spite of the fact that bilateral cryptorchid lambs are sterile, both unilateral and bilateral cryptorchids should be castrated. Rams with one or both testicles retained should never be used or sold for breeding purposes.

Dwarfism

Dwarfism occurs in many breeds of sheep. It might be classified as a sub-lethal defect because affected animals usually don't survive to reproductive age. In most breeds today, frequency of this defect is very low. There is likely more than one type of dwarfism, but the more distinctive type is that referred to as chondrostrophic or achondroplastic dwarfism, which is characterized by a shortening of the long bones due to premature ossification of the epiphyseal plate. This condition characterized the Ancon sheep, which was propagated as a breed until the late 1800s, is inherited as a simple recessive gene. Most recently (New Zealand Veterinary Journal, 2005), a different type of recessively inherited dwarfism was reported in a commercial flock of Texel sheep.

Spider Lamb Syndrome

Spider Lamb Syndrome (also known as Ovine Hereditary Chondrodysplasia) was first observed in the late 1960s within the Suffolk breed. In the 1980s, this defect spread rapidly among Suffolk flocks and within other blackfaced breeds. Spider Lamb Syndrome causes skeletal deformities in young lambs. These deformities commonly include abnormally long, bent limbs, twisted spines, shallow bodies, flattened rib cages and long necks. Affected lambs have a spider-like appearance. This syndrome is inherited as an autosomal recessive disorder; affected lambs must inherit the mutation from both their parents. Effects may be evident at birth or shortly thereafter. Affected lambs rarely live to reproductive age without intensive care. Those that do survive rarely produce offspring.

Animals known to carry the "spider" mutation, as determined by a DNA test or from having a "spider" offspring, should obviously be culled. Nobody talks about "spider" lambs today as compared to the mid 1980s, but Spider Lamb Syndrome does still play a role in sheep production. Today, sheep producers use DNA testing and monitor breeding programs to keep the frequency of the defective gene low and to prevent Spider Lamb Syndrome from resurfacing as a serious economic threat.

Dermatosparaxis

Dermatosparaxis is a genetic disorder causing a lethal skin condition. It is due to a simple recessive gene, as illustrated previously under "Inheritance Example."

Dermatosparaxis is most prominent in White Dorper sheep but has also been reported in Merino and Border Leicester-Southdown sheep. Affected lambs have abnormal collagen, which causes extreme fragility and tearing of the skin, usually in their inner thighs and under their armpits. The severity of the condition results in death or euthanasia at a very early age; thus, it is considered lethal or sub-lethal.

The abnormal or mutant form of the gene controlling Dermatosparaxis can be detected by DNA testing.

When Is A Flock At Risk?

Sheep breeds in which the defect was discovered are at risk (for example Spider Lamb Syndrome in Suffolks and Dermatosparaxis in White Dorpers), but genetic defects are not limited to purebred sheep. Crosses between "at risk" breeds are also at jeopardy. "At risk" animals are those that have known carriers in their pedigrees. The probability that an animal will be a carrier, given that it has a known carrier in its pedigree, is a function of how far back the known carrier is in the pedigree. The probability is $(1/2)^n$ where n is the number of generations between the animal in question and the known carrier. Table 2 shows the probabilities of a lamb being a carrier given several different pedigree relationships. As noted earlier, inbreeding increases the likelihood that recessive alleles can pair, and in the case of genetic defects, produce affected lambs. (See "Is Inbreeding Good or Baad?" in Genetically Speaking, *HoofPrint* Vol. 9 Fall 2012.) For the commercial sheep producer, crossbreeding can help reduce the risk of genetic defects.

Table 2. Probability of a lamb receiving a defective gene from a known carrier ancestor.

Relationship of known carrier to lamb	*Number of generations of separation*	*Probability lamb is a carrier**
Parent	1	50%
Grandparent	2	25%
Great-grandparent	3	12.5%
Great-great-grandparent	4	6.25%

* Calculated as $(1/2)^n$; n = number of generations between lamb and known carrier.

Use of Carrier Sheep in Flock Breeding Programs

Can sheep producers safely use carrier rams in breeding programs? For purebred or seedstock producers the answer is "no." For commercial producers, the answer is "maybe." It depends on the flock. If there is any risk that ewes in the flock are also carriers, a producer should avoid mating them to a ram that is a carrier of the same defect as the ewes. However, if a producer is confident his or her ewes are "clean," that is, non-carriers, he or she could choose a ram, based on EBVs and production traits, that best fits the breeding objectives, carrier or not. Still, if comparable non-carrier rams are available, future problems can be prevented by not using carriers. Remember that mating carrier to carrier results in a 50% chance that the resulting lambs will be carriers, and if ewe lambs are kept as replacements, they should not be mated to a carrier ram. Also, mating carrier to carrier results in a 25% chance lambs will have the defect in question.

If a flock contains carrier females, there are two primary options:

1. Cull all carrier females. If they are sold through a sale barn, full disclosure of their defect status should be provided.

2. Continue to breed the carrier females and routinely test their lambs, assuming a DNA test is available.

Producers should practice no inbreeding within the flock if there have been defective lambs. Generally, inbreeding should be avoided in all but truly superior flocks (See "Is Inbreeding Good or Baad?" in Genetically Speaking, *HoofPrint,* Volume 9, Fall 2012).

Methods of Eliminating Undesirable Recessive Traits

If producers cull both affected animals and known carriers of the mutant gene, selection pressure is being exercised against the genetic defect. However, the mutant gene will theoretically never be eliminated from the population. Consider a trait like fleece pigmentation. A simple recessive gene for pigmented or colored fleece is found in most fine wool flocks. Although the incidence of this trait is decreasing due to selection pressure against it, the trait has been around for hundreds of years. It will likely continue to occur, but in lower frequency for many more years. With traits, such as fleece pigmentation or color, which are associated with only limited economic loss, producers can tolerate a low, continuing incidence. However, in cases of lethal or sub-lethal traits, it is important to try and eliminate them from the population. If pedigree information is available and both sire and dam of affected animals are culled, the frequency of the gene and incidence of the trait can be quickly reduced.

It may be desirable to have rams genotyped for autosomal recessive traits before they are used extensively in flocks. Where DNA tests are available, as for the spider mutation or Dermatosparaxis, this method can be used to determine whether an animal carries the mutant gene.

Details on DNA testing can be found in "To Test or Not to Test," Genetically Speaking, *HoofPrint,* Volume 10, Winter 2013.

Summary

Genetic defects can never be completely eradicated. Mutations occur in every generation and new genetic defects are likely to be discovered in the future. However, with advancements in molecular genetic technology, genetic defects can be effectively managed in sheep breeds and flocks. Carrier animals can be identified and either culled or used in certain circumstances with confidence.

Commercial sheep producers need to understand the risk of defects in their flocks by knowing the pedigrees of sires they have used and their defect status. Crossbreeding has many benefits and can help mitigate the risk of producing affected lambs by decreasing the level of inbreeding. Purebred (seedstock) producers should practice full disclosure when selling carrier rams and ewes either as breeding animals or as intended culls.

Dr. Debra K. Aaron, PhD, *professor in the UK Dept. of Animal Sciences, teaches animal science and genetics. Her research interests are in sheep and genetic breeding.*