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Genetic Defects in Beef Cattle

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Advancements in molecular technology have made it possible to effectively manage genetic defects in beef cattle. This NebGuide discusses the primary defects being monitored by U.S. breed associations.

Genetic Defects in the U.S.

Genetic defects occur due to random mutations in the genome. The defects that are currently being monitored by U.S. breed associations *(Table I)* are autosomal recessive. Autosomal recessive means that two copies of the mutated allele are needed for an animal to be afflicted. Although *Table I* does not describe all the genetic defects in beef cattle, it includes the primary defects being monitored by U.S. breed associations. All of those listed have a simple recessive mode of inheritance.

Inheritance Example

Following is an example of Neuropathic Hydrocephalus (NH), a recessive lethal mutation found in Angus cattle. If a heterozygous bull (Nn) is mated to a homozygous normal female (NN), the resulting offspring would be 100 percent normal phenotypically (i.e., not afflicted) with 50 percent of them homozygous and the other 50 percent heterozygous.

	N	N
N	NN	NN
n	nN	nN

If two heterozygous (carrier) animals are mated (see example below), there is a 25 percent chance that the calf will be afflicted (nn), a 25 percent chance it will be homozygous normal (NN), and a 50 percent chance it will be a carrier (Nn and nN). The only time an afflicted calf can be produced is when two carriers are mated, and even then the odds are that the calf will be normal 75 percent of the time.

	N	n
N	NN	Nn
n	nN	nn

Descriptions of Genetic Defects

Hypotrichosis (Hairlessness). Found in several breeds, this disorder, caused by a recessive gene, can be complete or partial. Calves are born with little or no hair; in most cases, calves will grow a short curly hair coat as they age.

Table I. Genetic defects currently being monitored by U.S. breed associations.

Genetic Abnormality	*Primary Breed(s) of Incidence*	*Lethal or Nonlethal*	*DNA Test Available*
Alpha (α)-Mannosidosis	Red Angus	Lethal	Yes
Arthrogryposis Multiplex (AM)	Angus	Lethal	Yes
Beta (β)-Mannosidosis	Salers	Lethal	Yes
Contractural Arachnodactyly (CA)	Angus	Nonlethal	Yes
Neuropathic Hydrocephalus (NH)	Angus	Lethal	Yes
Hypotrichosis (hairless calf)	Hereford	Nonlethal	No
Idiopathic Epilepsy	Hereford	Nonlethal	Yes
Osteopetrosis	Angus and Red Angus	Lethal	Yes (Red Angus)
Protoporphyria	Limousin	Nonlethal	Yes
Pulmonary Hypoplasia and Anasarca (PHA)	Maine-Anjou and Shorthorn	Lethal	Yes
Tibial Hemimelia (TH)	Shorthorn and Maine-Anjou	Lethal	Yes

Beta (β)-Mannosidosis (Beta-man). This disorder is caused by a recessive gene. Calves born with this disorder do not get up after birth and soon die.

Arthrogryposis Multiplex (Curly Calf). This abnormality, which has been identified in the Angus breed, is caused by a recessive gene. Calves born with curly calf are deformed in the backbone and internal organ arrangement. They rarely survive birth.

Contractural Arachnodactyly (Fawn Calf). This mutation, although rarely fatal, gives rise to calves that are weaker and may have trouble nursing. Calves often have trouble with joint range of motion and do not lay down in a normal calf position. Calves are reported as more slender and taller than their normal siblings.

Neuropathic Hydrocephalus (Water Head). This lethal mutation causes an excess of water to form in the cranium.

Idiopathic Epilepsy. Animals are prone to seizures in which the animal falls to its side with all four limbs extended. Onset of the disease is variable by age, ranging from birth to several months old.

Osteopetrosis (Marble Bone). This lethal disorder is caused by a dense growth of bone, which invades or fills the bone marrow chamber.

Pulmonary Hypoplasia and Anasarca (PHA). Near absence of lungs, extreme fluid accumulation, and lack of lymphatic development; can create early embryonic death.

Tibial Hemimelia (TH). Calves will show abdominal hernia, severe distortion of rear leg structure, and exposure of brain and spinal tissue. Cryptorchidism is also possible.

Protoporphyria. Extreme sensitivity to sunlight.

Alpha (α)-Mannosidosis. Lethal nervous disease of Angus and Angus-derived cattle. Cattle afflicted with Alpha-Mannosidosis usually fail to thrive and develop a progressive incoordination and an aggressive disposition when disturbed. Affected animals usually die before reaching sexual maturity.

At Risk Populations

Breeds in which the defect was discovered are at risk, but genetic defects are not limited to purebred cattle. Composites (crossbreds) that contain a breed from an at risk population are also at risk. For instance, SimAngus cattle have the potential to carry genetic defects found in Angus cattle. At risk animals are those that have a known carrier in their pedigree. The probability that an animal is 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 II* illustrates this. Inbreeding increases the likelihood that recessive alleles can pair and, in the case of genetic defects, produce afflicted calves. For the commercial producer, crossbreeding can help mitigate the risk of genetic defects.

Table II. Probability of an animal receiving a defect allele from a known carrier ancestor.

Relationship of Known Carrier to Animal	*Number of Generations of Separation*	*Probability Animal is a Carrier*
Parent	1	50%
Grandparent	2	25%
Great-grandparent	3	12.5%
Great-great-grandparent	4	6.25%

Use of Carrier Animals in Breeding Programs

The question commercial producers have to answer is: "Can I safely use a carrier bull?" This requires knowledge of your cowherd. If there is any risk that your cows are also carriers, you should avoid mating them to a bull that is a carrier of the same defect as the cows. However, if you are confident your cows are clean (i.e., non-carriers), you could choose a bull, based on EPDs and Indexes, that best fits your breeding objectives, carrier or not. Still, if comparable non-carrier bulls are available, you can avoid future problems by avoiding carriers. Realize that in this scenario, there is a 50 percent chance that the resulting calves will be carriers, and if replacement females are retained, they should not be mated to a carrier bull. If you have carrier females in your herd, there are three primary options:

1. Cull all carrier females. If they are sold through a sale barn, full disclosure of their defect status should be provided. It is false to think that the seedstock sector is helping the commercial sector by putting numerous carrier breeding females on the market without providing knowledge of their defect status to potential buyers.
2. Use carrier females as recipient cows in an embryo transfer program.
3. Continue to breed the carrier females and routinely test their calves.

If you think you might have an afflicted calf, it is important to take a DNA sample from that calf and have it tested. It is also important to be able to correctly identify the animal's parentage so that the carrier sire can be identified. This may require genetic-based parentage testing. DNA samples can be garnered via hair follicles, blood, semen, nasal swabs, and ear notches. Check with the lab that will perform the test to determine the preferred sample type. Details on how to collect DNA samples can be viewed at *http://beef.unl.edu/learning/dnacollection.shtml*

Commercial Testing Labs

Two primary commercial labs offer tests for some of the genetic defects previously described.

- Merial Igenity
  http://us.igenity.com/
- Pfizer Animal Genetics
  http://www.pfizeranimalgenetics.com/default.aspx

Summary

With advancements in molecular technology, genetic defects can be effectively managed in beef cattle populations. Instead of purging entire lines of cattle, carrier animals can be identified and either culled or used in certain circumstances with confidence. Numerous genetic defects are currently being monitored by U.S. beef cattle breed associations and others likely will be discovered in the future.

Several defects do not have a genetic test. For these, an animal can be confirmed as a carrier if and only if it produces an afflicted calf. It is important to realize that some environmental factors can produce phenotypes similar to the genetic defects described. However, genetic testing can confirm whether the defect is due to environmental or genetic causes, assuming a test exists for the defect in question.

Commercial producers need to understand the risk of defects in their cowherd by knowing the pedigree of sires they have used and their defect status. Crossbreeding has many benefits and can help mitigate the risk of producing afflicted calves by decreasing the level of inbreeding. Seedstock producers should practice full disclosure when selling carrier animals, either as breeding animals or as intended culls.

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