## **Managing Genetic Defects**

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**KNOXVILLE, TENN.** The advent of national genetic evaluation by a number of purebred beef breeds has provided the industry a way to identify genetically superior animals in a number of traits within

a breed. This occurrence has provided the opportunity to identify and use a certain few sires within a breed in an attempt to fix a desired phenotype and performance in traits. It is not unusual for a sire that excels genetically in a particular combination of traits to appear in several generations of a pedigree on both sides of an individual. Also, with the additional means of multiplying a given set of genetics by artificial insemination and embryo transfer, a particular sire's genetics or bloodline can become very concentrated in a breed.

The concept of mating individuals within a line and trying to maintain a substantial degree of relationship to an outstanding individual within that line is called linebreeding. This concept has a tendency to fix a number of genes from the superior individual that influence superior performance in certain traits as well as other genes affecting other traits from that superior individual. It also enhances the opportunity for an increase in inbreeding and the likelihood of discovering a genetic abnormality affected by a recessive genetic defect.

Recessive genetic defects occur in all breeds and species, even humans. Recessive genetic defects in cattle are inherited congenital abnormalities. These genetic mutations occur on one of the 29 pairs of autosomal (non-sex coding) chromosomes. Animals that inherit a single defective or mutated gene from one parent and a normal copy of the gene from the other parent are called heterozygotes. Although they are not affected, heterozygotes carry the mutated gene and on average will pass it to half of their offspring. A recessive genetic defect is expressed only when an individual carries recessive mutated genes from both its heterozygous parents (homozygous recessive). Animals that do not carry a copy of the mutated gene are called homozygous normal and do not differ by observation, or phenotypically, from heterozygous animals.

Phenotypic selection against a recessive gene is ineffective since there is no measurable difference between the homozygotes and heterozygotes. It is similar to selecting against the red gene in coat color in cattle. Both the homozygous black cattle and heterozygous black (red gene carriers) are black and not distinguishable. However, when two black animals produce a red calf, we can determine that both parents are heterozygotes (red gene carriers).

In the past, proving suspected carrier bulls free of a recessive gene for a trait required mating the bulls to several known carriers (females that had produced an individual possessing the trait), or using sire-daughter matings. If a defective animal resulted, the bull was deemed a carrier of the recessive trait. Fortunately, advances in molecular genetics have provided new tools to identify specific mutations that cause defects. DNA-based diagnostics identify carriers and help manage various genetic defects. These technologies have led to the identification of genetic mutations that cause a number of genetic defects in various breeds: IE (idiopathic epilepsy), AM (arthrogryposis multiplex or "curly calf syndrome"), OS (osteopetrosis or "marble bone"), NH (neuropathic hydrocephalus) and others.

Genetic defects in the Angus breed have recently caused a tremendous amount of concern as a very popular bull (GAR Precision 1680) has been identified as a carrier of both the AM and NH genes. He has sired many offspring and some composite breeds (Gelbyieh Balancer, Simm-Angus, Chiangus, and others) have used his genetics in their developmental programs. His descendants have a possibility of being carriers for one or both genes. With the new DNA tests available, breeding programs can be managed toward eliminating or reducing the frequency of these recessive genes. Within the Angus breed, known AM carriers are denoted in the pedigree as AMC and animals proven free of the gene are denoted as AMF. Also, known NH carriers are signified by NHC and those free of the NH gene are reported as NHF. The majority of Angus AI sires have been DNA tested for both recessive genes and their results are posted on the Angus Web site (www. angus.org). Several other breeds list known carriers of various genetic defects on their homepage. Not all animals need to be tested, only those that have ancestors in their pedigree that are known or possible carriers would be necessary to eliminate the chance of the occurrence of one of these defects. Because of the mode of inheritance, it is also important to understand that an individual who has a carrier in his pedigree may him/herself not be a carrier.

Sound selection decisions and breeding programs can eliminate the occurrence of AM and NH. Commercial producers that use Angus bulls in their crossbreeding programs have a low probability of producing or losing calves from either genetic defect even if the bull they are using has a carrier in his pedigree. However, if that commercial producer saved heifers for replacements from that bull which had a carrier in his pedigree, they need to make sure that their next bull purchase is not himself a carrier for either genetic defect. The best procedure to eliminate the risk of having these genetic defects is to use only bulls proven to be free of both genes. The elimination of this condition is manageable with proper record keeping in a commercial cow-calf program.

Genetic defects are not new to the industry. All breeds contain some genetic defects and it is extremely rare that they are observed. However, closer matings (both bulls and cows that share common ancestors) increase the likelihood of their occurrences. With the genetic molecular technology available and the ability to easily identify carriers, abnormalities can be eliminated in more quickly.  $\Delta$ 

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