Managing Genetic Conditions

Where are these genetic conditions coming from? A word on mutation.

by ***Tonya Amen,*** *American Angus Association*

With the Aug. 12 announcement of a new genetic condition (developmental duplication, or DD), many producers are asking questions regarding how this problem arose. It's a good question, the answer for which also sheds light on how cattlemen can genetically manage their herds to minimize the effects of what is simply a genetic mutation.

So what is a mutation? In simple terms, a mutation is a change in genetic material (or the process by which the change occurs). This change can be as simple as a swapping of one nucleotide (the As, Ts, Gs and Cs that make up our DNA) for another, or it can be more complex, involving large or small insertions or deletions of nucleotides.

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Between 60 and 100 new mutations occur *in every animal,* and half of these (along with any historic mutations they inherited from their ancestors) will be passed on to their offspring. The majority of the time this is not a problem because some mutations cause no visible phenotypic change, and those that do are typically recessive, meaning that an individual must inherit copies from both parents for a change in phenotype to be observed (something that is unlikely when unrelated animals are mated).

However, when related animals are mated, the chance for an individual to receive two copies of a mutated gene increases, and when an animal becomes widely used, as is the case with popular sires used by artificial insemination (AI), these mutations can be spread through the population.

It's important to note that mutation is valuable, if not essential, as it creates new raw genetic material for evolution and thus is the source of all genetic variation. However, in the Angus breed, we've also come to know mutation as the source of genetic conditions such as arthrogryposis multiplex (AM), neuropathic hydrocephalus (NH), contractural arachnodactyly (CA), etc. (For more information on these various conditions, please refer to the fact sheets for each condition, available online at *www.angus.org/angus.aspx.)*

Recessive mutations for simply inherited traits

Cattle are diploid organisms, meaning they have a pair of each type of chromosome, with one inherited from the sire and one inherited from the dam. That means genes on the chromosomes are also inherited in pairs — with one allele, or form of the gene, from the sire and one allele from the dam.

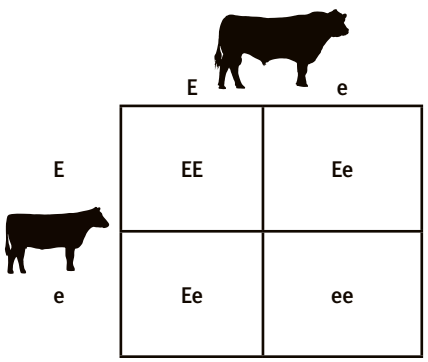
From the days of Mendel, we know that these alleles can have a dominant form and a recessive form. If the dominant form is present, it will always express itself in the phenotype.

A simple recessive phenotype is one in which an animal must have two copies of a recessive allele in order to express the corresponding phenotype.

One of the most common simple recessive phenotypes in cattle is red coat color. So, for the sake of understanding inheritance of simply inherited traits, we'll use it as an example. Black is dominant to red and, thus, an animal must receive the red allele from both parents in order to have red hair.

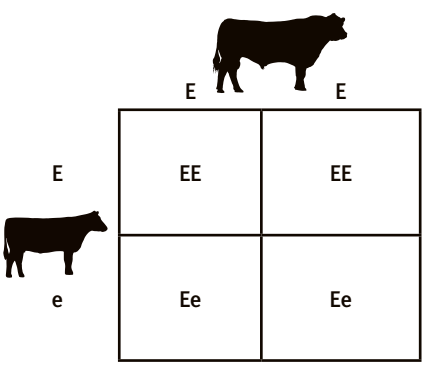
For the two examples below, let's assume that "E" codes for black coat color and "e" codes for red. Along the top of each box are the possible alleles contributed by the bull, the possible alleles from the cow are along the side. An animal is said to be homozygous if it carries two copies of the same allele, or heterozygous if it carries one copy of each variation of the allele.

Example 1: A heterozygous bull (black in color himself, but carries one copy of the red allele) is bred to a heterozygous cow (Ee × Ee).



Three-fourths of the calves from this mating (those that are EE or Ee) will be black and one-fourth (those that are ee) will be red. However, from a genotypic standpoint, two-thirds of the black animals will carry the red allele and have the ability to pass it on to future generations.

Example 2: A homozygous black bull is bred to a heterozygous cow (EE × Ee).



In this example, 100% of the calves will be black. However, 50% will carry the red allele and be able to pass it on to future generations.

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How does this relate to genetic conditions?

Let's assume that instead of coat color, the "E" and "e" alleles are associated with a recessive genetic condition. "E" codes for unaffected or normal phenotype, and "e" is the recessive, or affected, allele.

Example 1 shows the only mating strategy that will lead to calves affected by the various genetic conditions. In order to observe affected calves, both parents must be carriers. Another important result of mating strategy 1 is that two-thirds of the unaffected offspring of this mating will be carriers of the recessive allele.

Example 2 shows one strategy for having only unaffected calves. It shows the logic behind advice to commercial cattlemen to buy bulls tested to be free of genetic conditions that could have a negative effect on production and profitability. Calves born to those bulls, even if mated to carrier cows, should not display the recessive mutation.

In terminal situations, where all calves are sold for harvest, some may find this to be an acceptable strategy. However, if any of the resulting offspring are to be kept as breeding animals, it's important to recognize that one-half of them will be carriers.

How are recessive alleles spread?

In most cases, recessive mutations will go unrecognized. They may occur in animals that aren't used for breeding. Or, if they do occur in breeding animals, most of the time they would remain confined to a single herd and, assuming inbreeding is avoided, two carriers would never be mated. However, if an animal carrying a deleterious recessive mutation happens to become popular and widely used in the population, inevitably, two animals that carry the recessive mutation will be mated. It may take a while to recognize the recessive condition because oftentimes these conditions may cause reabsorption early in pregnancy. If abnormal calves are carried to term and born, they may be dismissed as just a "freak" occurrence.

Things to remember with simply inherited traits:

- ►Between 60 and 100 new mutations occur in every animal.
- ►Mutation can be valuable, providing the genetic material for evolution and genetic variation.
- ►To express a simple recessive phenotype, an animal must carry two copies of the recessive allele.
- ►Mating a heterozygote (Ee) to a heterozygote (Ee) will result in 25% of the calves being EE, 50% being Ee and 25% being ee. Only ee calves will show the recessive phenotype.
- ►Mating a homozygote (EE) to a heterozygote (Ee) will result in 50% of the calves being EE and 50% being Ee, with none of the calves expressing the recessive phenotype.
- ►The majority of registered-Angus animals are not potential carriers for any of the currently recognized genetic conditions.
- ►A potential carrier, having a known carrier in its pedigree and not tested free of the gene, will be designated as such on its registration paper.
- ►The only way to know if a "potential" carrier is actually a carrier is to conduct a DNA test.
- ►In the Angus registry, animals tested for a genetic condition are designated as carriers or free under the animal's birthdate on the registration paper.

How to tell if an unaffected animal is a carrier?

The majority of registered-Angus animals are not potential carriers for any of the currently recognized genetic conditions. However, if an animal has a known carrier in its pedigree, you will find a statement alerting you to this on its registration papers. Look for the wording "this animal has one or more ancestors who are carriers for x", where "x" is a specific genetic condition. According to current policy (this will be true for DD seven months after commercial availability of a test), if an animal has a carrier or carriers in its pedigree, the only way to know for certain whether the animal in question is free or a carrier is to have it genetically tested. Testing for all genetic conditions recognized by the Association is available through Angus Genetics Inc. (AGI).

In many cases, potential carriers may have been tested for the proper genetic conditions before they are sold. Look for GCC or GCF (where GC stands for genetic condition, C designates a carrier, and F designates a non-carrier) on the registration papers right below the animal's birthdate. If the animal is GCF you can breed it with no fear of having an affected calf. If the animal is GCC, you'll need to adopt a management strategy to avoid affected offspring and to deal with potential carriers entering your breeding herd. AJ

Editor's Note: *Tonya Amen is genetic service director for the American Angus Association.*