

MAKING SENSE OUT OF ALL THE GENETIC DEFECT BUZZ

An examination of a number of recently surfaced bovine genetic defects and what can be done to limit their impact on the beef industry.

By Marty Ropp, ASA Director of Field Services



Unless you have been hiding under a rock lately, you are well aware of the publicity surrounding “new” genetic defects that are significantly affecting both the commercial cattle business and the business of several breed organizations including our own. The word new is in “parenthesis” only because few if any of these genetic mutations are actually recent in origin. Several have very long histories of reports while other mutations have perhaps only occurred in the past 20 years. These abnormalities have become big news very quickly because of the number of influential animals and pedigrees implicated as defect carriers. Adding to the attention, are genomic technology advances that now allow us to confirm cases and build diagnostic tests for identifying defect carrier individuals relatively quickly. In fact, advanced DNA testing is our industry’s number one tool for managing and minimizing the effect of defects on breeding populations. Because of the availability and effectiveness of new age genomic testing, we need not fear these deleterious genes, nor panic and overreact to their presence as breeders of the past were forced to do often wiping out entire valuable lines of seedstock to control the spread of single genes.

As with any significant challenge to our business, there is substantial misinformation and half-truths associated with the hype surrounding genetic abnormalities. Let’s start with some half-truths. It is common to hear that selection for certain traits has brought about these unwanted consequences. The true half of this argument is that intense genetic selection surrounding a single individual or line increases the gene frequency of all the genes of an individual both positive and negative. Selection concentrates genes. Therefore, if a highly used individual in a breed or line happens to be a carrier of an unwanted genetic defect, the frequency of its deleterious gene is increased in addition to the frequency of the desired genes. Using that same logic one could erroneously blame artificial insemination or high-quality genetic evaluation for creating the problem. For example, it is very common to hear that a particular line of Angus cattle has genetic defects because breeders selected too much for marbling. Certainly there is a specific high marbling line that has been proven to carry genetic defects just as there are high marbling lines without defect. More likely, a sire line that was proven to be superior for certain traits, by chance, happened also to be genetic defect carriers.

When these ancestors became extremely popular, the genetic defect frequency increased substantially in that breed population and the rest is history. It is possible, however, to have unwanted genes linked to desirable genes or gene effects. In the case of the halothane gene in the world of pork production. There is some unproven rhetorical evidence that Tibial Hemimelia may be linked to certain “show ring desired” phenotypes that led to unusually rapid increased gene frequency in certain populations, but most of the time they are accidental consequence associated with intense use and or popularity of an individual in a selected line.

I, too, have seen written that line breeding or inbreeding is the culprit. Again, just a half-truth. In fact, inbreeding individuals is one of the fastest ways to discover genetic abnormalities in a population before an individual sire or dam can become highly influential. The truth part is that inbreeding increases the potential for a single recessive gene to be expressed in an affected calf because it increases the likelihood of creating homozygous individuals for a trait, because of individuals represented in recent generations on both sides of the pedigree. Without significant inbreeding, genetic defects can and have hidden in populations for centuries before selection and or circumstances bring their effects to bear. Often by that time the defect has spread to so many lines and individuals its effects on breeding populations are substantial.

Let’s go over the list of some high profile defects with some candor regarding their origins and spread. To date all of these genetic defects are considered to be simple recessives and are therefore inherited in a similar fashion to horns or red coat color which are also single gene autosomal recessives, but with less serious consequences. In depth descriptions and genetic details regarding these and other genetic defects are available at www.simmental.org.

TH — Tibial Hemimelia — Originally identified in Shorthorn and perhaps Galloway lines more than 30 years ago, this defect has recently become common because of increased line selection centered around offending animals. Spread of this defect to other breeds has been accelerated by the interest in Shorthorn genetics for club calf production. There are claims that carrier animals have phenotypes more similar to ideals for the show steer business such as having more hair than defect free relatives, but this has not been substantiated.



*Calf with tibial hemimelia.
Note short malformed hind limbs
and long curly hair.*

PHA — Pulmonary Hypoplasia with Anasarca — Primarily identified with offending animals originating in the Maine Anjou herdbook, this defect has also been concentrated due to carrier lines becoming popular. It too has spread to other populations primarily as a result of breeding for composite club calf production.

AM — Bovine Arthrogyrposis Multiplex Congenita

— More commonly known as “Curly Calf,” this lethal defect has been identified in a significant number of popular Angus pedigrees. The relatively high gene frequency of AM in Angus lines is primarily due to the popularity and subsequent influence of G A R Precision 1680 and his sire Rito 9J9 of B156 7T26 in the American Angus herdbook. As an unintended consequence of concentrating the positive genes offered by 1680 and 9J9’s descendants, AM was also selected for. The use of CA Future Direction 5321 and his descendents to make SimAngus has been the primary source of this deleterious gene in the Simmental herdbook.



Cases of AM

NH — Neuropathic Hydrocephalus

— This lethal recessive trait has also been associated with the descendants of GAR Precision 1680. Unlike AM, that was passed down from Rito 9J9 of B156 7T26, this sire may be the point of mutation for this form of Hydrocephalus. It is extremely rare to identify the original mutation point for any genetic defect. AM and NH do not appear to be related, but these genes can actually appear together in the same animal as in the case of several Angus sires including CA Future Direction 5321. In the case of animals that carry both defects the probability that progeny of these sires or dams are free from both defects when bred to defect free animals is only 1 in 4. When an individual carries only 1 defect, one half of the progeny will be defect free when mated to clean individuals.

OS — Osteopetrosis — More commonly known as “Marble Bone” this defect can be most commonly found in several Red Angus pedigrees, but has been historically documented in Angus cattle as well. Some question whether the mutation is the same in both breeds but to date no one has confirmed a difference. The two most widely used offenders identified in the Red Angus registry are Buf CRK Romeo L081 and LCHMN Grand Canyon 2419J. With the popularities of these two sires because of the positive traits they offered, the unintended spreading of genetic defects is regrettable and most probably unrelated to other traits.

HT — Hypotrichosis (Hairless Calf) and IE Idiopathic Epilepsy — Both appear to originate from Hereford populations. Though these deleterious genes occur in a very low frequency in the Simmental herdbook, many original US Simmental animals were bred up using Hereford cows as a base. DNA tests are currently available for each of these defects.



Calf with Hypotrichosis.

FC — Fawn Calf — This defect is listed last because very little documented information regarding FC is currently available. It is however found in significant frequency in several popular Angus lines, but to date those offending lines are not publicly confirmed. Currently, no DNA test is available for Fawn Calf. FC is not lethal in all cases but has debilitating effects on calf mobility and growth rate.

DNA tests for these (not including Fawn Calf) and other genetic defects can be completed relatively quickly and are extremely effective for individual screening. This makes dealing with serious genetic defects far easier today than in the past. Many defect free individuals with immense genetic value have already been identified within carrier lines. In some cases, high value carrier parents can still be utilized to make more valuable defect free progeny for the future. Decisions need to be made based on the science available and not the immediate emotion of panic associated with past experience or the fear of short term negative public opinion. Yes, these defects need to be minimized in populations by aggressive selection for free individuals, but the truth is that few if any of these genes will ever be eradicated despite propaganda to the contrary. For example, most breed registries run mis-parentage rates of 3%-6% when randomly DNA tested. Therefore some offending individuals have already passed the genetic defect screening gates because their erroneous pedigrees were not suspected and thus not tested. Though the risk is very low, this is part of the rationale for testing all AI sires and donor females for parentage and why the ASA screens all of the top 50 sires in the breed for available genetic defect tests, it’s just good risk management.

It is crucial that carrier animals are identified so the offending genes can be selected against and not just the individuals or ancestry. The ASA genetic defect policy helps breeders to do just that. It warns of but does not disqualify potential vectors and provides genetic roadmaps to understand which animals need to be tested or create reasonable concern without legislating breeding programs or options. We will combine DNA technologies available today with market pressure to reduce genetic defects to make a better and more defect free product today and in the future. So, make wise, informed decisions regarding genetic defects, use all the tools available to produce genetic packages with increasing value while protecting both your customers and your business.

If you have an individual that you believe to be genetically defective, check out the ASA website www.simmental.org for steps you can take to report the situation and for procedures to collect photographic evidence and DNA for potential diagnosis. Most birth defects are congenital and not truly inherited genetic defects, but reporting abnormalities helps protect our membership and customers from long term challenges.

ST