

Tibial Hemimelia Threatens SimGenetics

Dr. Jerry Lipsey Executive Vice President

An examination of factors involving genetic abnormalities in beef cattle

Perhaps every organism, from one as simple as a single-cell amoeba to one as complicated as a beef cow, has genetic abnormalities. If a mutation occurs in groups of genes that control quantitative traits such as back fat or frame score, we may not observe much or any phenotypic response. However, if a mutation occurs at a gene that alone controls a trait (this would be a qualitative trait) such as hair color or dwarf vs. normal body development, the response is often very apparent and sometimes devastating.

This means that not all mutations are bad. For example, horned genes mutating to polled could be considered useful and consequently, fortunate. The mutations that bring about seriously debilitating to lethal results are those that concern us most. Under the worst-case scenario, what if an influential sire such as Abricot (reg. #58,311) had a deleterious mutated gene?

Just for a quick reminder, Abricot sired Mr. Clean, who sired Single Tree Smoke, who sired Landridge Black Jet, who sired Circle S Leachman 600U. Well you get the idea.

Significant Factors

There are several important issues that play a role in managing genetic abnormalities:

1. The impact of the mutated gene (benign or lethal?)
2. The potential reproductive influence of the animal with the mutated gene (a cow vs. a bull, or an influential A.I. sire vs. a bull in a commercial herd)
3. The ease or difficulty identifying the impact of the mutation (as obvious as polydactyly to as invisible as mannosidosis (a sugar metabolism abnormality))

A genetic abnormality can be as unthreatening as a change in hair color, or a nuisance such as near-sightedness. Could cows be near-sighted? Anyway, it's the serious abnormalities that we must document and monitor. Fullblood Simmental imported from Europe have been documented carriers of Cleft Palate, Cleft Lip, Syndactyly, Polydactyly, Spastic Paresis and several other abnormalities. Since all the other cattle in our database (purebreds to percentages) contain genes from other breeds, it is logical that many of the documented genetic abnormalities that exist in cattle "have been unknowingly captured" by our members. Fortunately, we have been vigilant to report and cull parents and offspring with obvious defects and/or poor performance.

Defect of the Tibia

Tibial Hemimelia (TH) a defect of the tibia (a bone in the lower hind leg) has been known for many decades. Calves with this condition (this is a simple autosomal recessive like horned, where both genes on the paired chromosomes must be in the recessive form to show the characteristic) are unable to stand, nurse, walk and consequently, do not survive. Apparently, this is most common among the British breeds, Galloway and Shorthorn, for example.



Femur from calf affected by tibial hemimelia. The distal epiphysis is large and rounded without normal development of the condyles and intercondylar fossa.



A Shorthorn calf with tibial hemimelia. Note the short, malformed hind limbs and long, curly hair.

Recently, the American Shorthorn Association announced that they have documented several sires that are carriers of TH. ASA staff has had conversations with breeders who say some Maine Anjou and Chianina sires (apparently with Shorthorn blood) are also carriers. Several of these Shorthorn, Maine Anjou and Chianina sires have progeny both submitted and registered in the ASA database. Fortunately, there is DNA marker technology to ascertain an animal's TH status.



Jerry Lipsey

Dr. Jon Beever from the University of Illinois has validated a DNA marker for Tibial Hemimelia. He has filed his discovery with the U of IL Office of Technology Management and that office is currently negotiating potential licensing agreements with the American Shorthorn Association. Members of the Shorthorn Association have used Dr. Beever's marker test to identify carrier animals. ASA members can also receive services from Dr. Beever.

ASA Policy for monitoring TH genetic defect

1. ASA will test the top 50 sires based on the number of progeny reported in the previous calendar year and thereafter any new sire entering the top 50 sires list.
2. Progeny out of a registered suspect animal can not be registered until the suspect animal (parent) is tested.
3. A suspect animal is an animal who is a) out of a TH carrier and has not been test free or b) who has not been tested and has 1/8 or more Shorthorn, Maine Anjou or Chianina blood.
4. ASA's procedure for TH testing includes parentage verification.

Controlling Genetic Abnormalities

Managing genetic abnormalities has always been part of wise and prudent livestock production. As DNA technologies increase our abilities to identify carrier animals, we must use them wisely. For example, you may have heard that another genetic abnormality has been diagnosed in Shorthorns, Pulmonary hypoplasia with anasarca (PHA).

Pulmonary hypoplasia with anasarca has been documented in Shorthorns and rumored to exist in some Maine Anjou and Chianina cattle. The following is paraphrased from the Shorthorn website www.shorthorn.org (then go to Programs, then to Genetic Defect). PHA in Shorthorns (from the Shorthorn Website) Pulmonary hypoplasia with anasarca (PHA) is a new syndrome reported in over a dozen Shorthorn calves over the last few years. To date only four cases have been well documented. The ASA and researchers at the University of Nebraska request your assistance in identifying cases and delivering calves or select tissues for laboratory examination. The cause of the syndrome has not yet been determined; but, it is uniformly lethal. The cows also suffer from dystocia related to the large size of the calf. This syndrome is distinct from tibial hemimelia (TH).

Anasarca refers to the collection of fluid in the skin and body cavities of the calf. In some cases the calves had been reported as "bulldogs" due to the facial appearance caused by this fluid collection. The term should not be applied as this syndrome is distinct from bulldog dwarfism and using the term could be misleading. The fluid markedly increases the size and weight of the fetus causing dystocia at time of delivery. In the four cases examined at the University of Nebraska, small, under-developed lungs were found. The full complement of abnormal features in these calves has not been fully explored. The Shorthorn Association is encouraging breeders to report affected calves.

Summary

Spring calving season is upon us. If you have unusual or mis-shaped calves, please call either Wade Shafer or Jerry Lipsey as soon as you can. Either keep the calf alive or chill/freeze the dead calf until you have had time to report the occurrence. All communications are kept in complete confidence, and ASA staff will try to verify the cause of each problem. ♦