Report on the Canadian Angus Association Annual General Meeting Roundtable Discussion on Genetic Defects in Beef Cattle

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Dr Jon Beever, molecular geneticist and associate professor at the University of Illinois, and I co-hosted the genetic defect roundtable discussion at the Canadian Angus Association's annual general meeting in June. Dr Beever has developed DNA testing for AM, NH and OS in Angus, Idiopathic Epilepsy in Hereford cattle, Spider Lamb Syndrome in sheep, Tibial Hemimelia in Shorthorn and Simmental cattle, and Pulmonary Hypoplasia in Dexter cattle.

The session began with a brief overview on the basics of inheritance, a summary of some of the genetic defects that occur in beef cattle and are monitored by Angus associations globally, and the Canadian Angus Association Genetic Defect Policy and strategy to eliminate these recessive genes from our herdbook. I will briefly recap the basics of inheritance for those of you who were unable to attend the AGM. All genes typically have two versions of it called alleles. Usually one of these alleles is dominant and dictates the phenotype (observable characteristics such as coat colour and weight). If an animal is heterozygous (or a carrier animal), then the dominant allele would mask the presence of the recessive allele.

	A	**A**
A	**AA**	**AA**
a	**Aa**	**Aa**

In this top example a homozygous or non-carrier animal is bred to another animal that is phenotypically the same but genotypically it carries the recessive allele. All the calves from this breeding will look the same, but half will inherit the recessive allele and be carriers.

Still, if they all look the same what is the problem? Well, let's breed two of these carriers:

	A	**a**
A	**AA**	**Aa**
a	**Aa**	**aa**

Now three-quarters of the calves will look the same, but half are carriers of the recessive allele, and one-quarter of the calves inherited two copies of the recessive allele. Without the dominant allele to mask the recessive allele these calves will display the recessive trait. Is this a bad thing? It depends what sort of a phenotype that recessive allele codes for.

Here is a list of phenotypes in beef cattle that are coded for by recessive alleles:

- Arthrogryposis Multiplex (AM)—formerly known as Curly Calf Syndrome; affected Angus calves are stillborn with twisted spines
- Double Muscling (DM)—affected calves are heavily muscled but weak boned
- Dwarfism (DW)—occurs in different variations and is caused by several different genes
- Fawn Calf Syndrome (FC)—now called Contractural Arachnodactyly; affected calves have deer-like postures and do not thrive and grow comparatively

- Heterochromia Irides (HI)—also known as white eye; affected calves have white or silver rings around their iris
- Hypotrichosis (HY)—hairlessness which occurs in varying degrees
- Alpha Mannosidosis (MA)—a lysosomal condition wherein affected calves develop aggression and loss of muscle control around yearling age
- Neuropathic Hydrocephalus (NH)—balloon head; affected calves are stillborn with fluid in their heads
- Osteopetrosis (OS)—formerly known as marble bone disease; affected calves are stillborn with brittle underdeveloped bones
- Protoporphyria (PR)—causes a sensitivity to light; sores and scabs develop with exposure to light
- Pulmonary Hypoplasia (PH)—affected calves are stillborn with fluid in their abdominal cavities
- Syndactyly (SN)—also known as mule foot; affected animals have fused feet
- Tibial Hemimelia (TH)—a fatal condition that causes large abdominal hernias

All of these phenotypes impact breeders' and commercial producers' bottom lines. DNA testing developed by Dr. Beever and other researchers enables Angus breeders to identify carrier animals before they throw affected calves. We can use this information in two ways: to avoid the potential of having affected calves, and also breeding carrier animals and propagating the gene further into the herdbook. One non-AI male carrier animal can potentially throw 246 carrier calves within two generations. This probability should caution all breeders from using carrier animals for breeding.

In order to monitor issues caused by recessive alleles, all breeders should report abnormal calves to the Association. We can run a Geneprob herd analysis to identify any animals in your herd that should be tested. It is recommended that testing be done in stages, testing the dams and sires first. If the parents are not carriers you don't need to test any of their calves because the cycle has been broken and the gene cannot be passed on. To request a test kit, simply submit a genetic defect test request form. The testing costs $26 per test and can be performed on hair samples. The results are sent to the breeders and maintained in the Association database. We post the results on our online herdbook, on the animal registration certificates, and our sales catalogue pedigree extracts. The results are displayed as AMF for AM free, or AMC for AM Carrier. In a similar fashion we also use NHF, NHC, OSF, OSC, MAF, MAC.

Why share your test results? So that other breeders using the same genetics doesn't have to repeat the same tests. For example, if you have found a sire not to be a carrier then other breeders using the same sire might not have to test their calves. Or, if you know that there is a carrier in the pedigree then sharing the test results allows other breeders to make informed decisions. And finally, because this is information commercial producers need and want. You would be surprised how many bull buyers call me for pedigree analysis, and how positive they are about having this information available to them. **We are the only breed that is giving bull buyers complete pedigree information.**

This is the era of genetic technology. We're going to start using markers for traits to give us better EPDs soon. We use DNA technology for parentage testing to make sure that our pedigrees are accurate; these DNA tests are simply a selection tool to help us realize our goal of eliminating these genes from our herdbook.

Some of the questions asked of Dr. Beever were:

Q: Are these defects caused by mutations?
A: Yes, every time DNA is replicated mistakes occur. Whether this mistake gets translated into a phenotype depends on where this mutation occurs. Mutations do provide us with variation, some of which we select for and some against.

Q: Have there even been any affected calves?
A: Even though the probability of breeding two carriers to each other is extremely low, there have been affected calves. In Canada these have been minimal and limited to commercial producers. In the U.S. there have been a larger number recorded. It is when a breeder experiences economic loss in affected calves that researchers like me are approached to identify what is causing the issue and provide some sort of screening tool against the issue.

Q: Now that the bovine genome has been sequenced will you be coming out with more of these things that we need to test for?
A: Yes, the bovine genome has been sequenced which makes it somewhat easier to sequence the causative gene for a phenotype. However, scientific researchers do not go around trying to find new genetic defects. These genes have always occurred in all animals. Researchers work on identifying these genes when someone in the industry, a breeder or an association, approaches them to help find out why they are consistently losing high numbers of their calves to similar and fatal phenotypes. Then researchers step in to try and identify why this is happening, and provide a tool for screening against this occurrence.

Q: Can you give us an update on Fawn Calf Syndrome (now CA)?
A: Researchers in Australia have line bred several affected animals and potential carrier animals and have identified five animals thought to be carriers of the recessive allele thought to cause this condition. My own research has been very frustrating but is now close to completion. There should be a valid and robust test for the causative gene shortly. The Association will receive an update on this and will make the information available as soon as possible.

Q: Is Fawn Calf Syndrome a lethal condition?
A: Fawn Calf is not fatal; the animals do survive and can recover to become relatively normal phenotypically. However, in my mind the phenotype is lethal because an affected animal will likely never make it into a breeding program.

Q: What probability would you test at?
A: Testing should be focused on animals that have upwards of 24% probability of inheriting one of these recessive alleles. Testing should begin at higher generations; test dams and sires first rather than all the calves. Of course, this also depends on the way in which the animal will be used; a herd sire may warrant testing at lower levels of probability.

Q: Is this genetic defect stuff really a big deal?
A: The goal is to avoid economic loss from affected calves. When the gene frequency in the population is high the potential for this to happen is increased. So minimizing the gene frequency should be the goal. The frequency of these recessive alleles is typically very low in any herdbook unless they are propagated heavily by using carrier AI sires. Now that this has happened for AM, NH, OS and MA the issue can be controlled and minimized by identifying carriers and not breeding them. If you are testing selectively, based on pedigree then keeping very accurate pedigrees and reporting all abnormal calves to the Association is a must. We need to be diligent so that this sort of recessive gene propagation does not happen again. The last few years have been a learning experience for everyone in the industry. We are now well equipped to deal with recessive traits.

Members are welcome to contact Kajal Devani at kajal1@cdnangus.ca or 1-888-571-3580 with any questions on genetic defects and management of recessive traits in Angus cattle.