



It's All In The Genes!

by Lana Kaiser, DVM

Genetics 101

Genes are the things that make us what we are. We get half of our genes from our mother and half from our father. The same is true for cattle (and all other mammals for that matter). Genes are found on chromosomes. Chromosomes are threadlike structures found in the nucleus of cells. Chromosomes carry genetic information. Humans have 23 pairs of chromosomes and cattle have 30 pairs. Chromosomes are always in pairs — one of each pair is inherited from the mother (dam) and the other from the father (sire). If the offspring is female, both parents contributed an X chromosome. If the offspring is male, the mother contributed an X and the father a Y chromosome.

The genetic information on chromosomes is arranged in a linear sequence. Basically, chromosomes have genes lined up in the same order, for example (hypothetically speaking), the gene for coat color sits on top of the gene for coat pattern, which sits on top of the polled gene, etc.

In essence, half of a calf's genetic material comes from the sire and half from the dam. Both sire and dam contribute information for all traits. For example, the sire doesn't contribute the coat color and the dam the pattern of color, both provide input to coat color and pattern.

Let's look at coat color. The sire is black, the dam red. That is "phenotypic" information. Phenotype is what you see. Genotype is the genetic information in

the chromosomes. The genotype determines the phenotype. There are two variations in the genes for coat color. The different variations are called alleles. Black is the dominant allele, red is the recessive allele. A dominant allele "trumps" a recessive allele. A black bull can be homozygous black (both alleles code for black) or heterozygous black (one black and one red allele). Because red is the recessive allele, we know the dam is homozygous red. If the calf from this mating is red, we know the bull carries a red allele (i.e. is heterozygous). If the calf is black, we don't know if his sire is homozygous or heterozygous black.

A few examples might help. B = black and b = red. The capital letter is dominant and the lower case is recessive. If the bull is black and the cow red, the bull can be homozygous black (BB), as seen in Table 1 or heterozygous black (Bb), as seen in Table 2. The bull's genotype is shown across the top and the cow's genotype down the left. The four boxes show the potential combinations of genotypes from this mating, in the first case (Table 1) all calves will be born Bb (genotype) and will appear black (phenotype).

Table 1: Mating a homozygous black bull to a red cow. All calves will be black, but carry the recessive red gene.

Table 1	B	B
b	Bb	Bb
b	Bb	Bb

Table 2 shows how both the genotype and phenotype of the calf can be affected if the bull is heterozygous for black (carries the red allele, Bb) and the cow is red (bb).

When I say "genes" – do you think Wrangler's? We are going to talk about genes, but not the kind you wear, so sit back on your pockets and off we go.



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Since black is dominant, the calves from this mating have a 50% chance of being heterozygous black (Bb - a black calf with one black and one red allele) and a 50% chance of being born red (bb). For the recessive gene to be displayed in the phenotype (i.e. for you to see it), the calf must inherit this gene from both parents. The same is true for calves born with recessive diseases; the calf must inherit this gene from both parents.

Table 2: Mating a heterozygous black bull to a red cow. Half of the calves will be black but carry the recessive red gene and half will be red.

Table 2	B	b
b	Bb	bb
b	Bb	bb

If a calf gets a recessive gene from both parents, they will show (express) the characteristic in their phenotype. Now let's say both the bull and cow are black, but they are heterozygous black (Bb - they both carry the recessive red gene). Table 3 shows us what will happen. Three of four calves will be born black, but only one is homozygous black (BB); two of four are heterozygous black (carry the recessive red gene), and one of four is red (carries a recessive red gene from both sire and dam). If you understand Table 3, you can understand recessive diseases of cattle (and other species for that matter!).

Table 3: Mating a heterozygous black bull to a heterozygous black cow. Three calves will be born black, but only one will carry two alleles for black, the other two will be heterozygous black. The fourth calf will be red.

Table 3	B	b
B	BB	Bb
b	Bb	bb

Breeding For Phenotype

When humans control the breeding of any species, they often pick the mating based on "look" or in the case of performance animals, a particular skill. This often results in inbreeding and usually because the

male is able to create more offspring than the female (although this has changed some with ET), the defect will trace back to a popular sire. This phenomenon has been seen in many breeds of cattle, horses, dogs and likely other species as well. It is the unwanted consequence of human intervention! So it is up to us to use the information to make rational breeding decisions that will promote our breed to its full potential.

TH - The Journey

Although Dr. Dave Steffens, University of Nebraska, described TH (tibial hemimelia) in Shorthorn calves in a paper published in 2000 in *Veterinary Pathology*, it was the efforts of Dr. Chuck Hannon, Indiana, who "chased down" calves with TH and obtained their pedigrees and Dr. Jonathan Beever, University of Illinois, developed a genetic test for carriers of the defective gene.

TH is a disorder of multiple congenital defects seen in calves. Translated, the name refers to absence of all or part of the tibia (tibial = the bone between the knee and ankle in humans or the stifle and hock in cattle and hemimelia = absence of all or part of a limb). But TH encompasses more defects than the name implies. TH is lethal, as the calf may be born dead, but if not dead the condition is incompatible with life. These calves have large abdominal hernias (weakening of the abdominal muscles that allows intestinal contents to bulge out), twisted legs sometimes with absent or deformed bones. They may be cryptorchid (undescended testicles), have a meningocele (defect in the skull with tissue coming out) and a long shaggy hair coat.

TH was first described in Galloway cattle in the 1960's and in Shorthorn cattle in 2000. Since that time hundreds of calves with TH have been identified. Studying the pedigrees of affected and unaffected cattle has led to the conclusion that TH is a recessive disorder. For



Figure 1 - Shorthorn calf with TH, note the large abdominal hernia and the twisted back legs. Notice the hoof of the top leg is backward from its normal position.



Figure 2 - Shorthorn calf with TH. Note the large abdominal hernia and the twisted back legs.

the calf to be affected, it must have inherited the defective gene from both parents.

Using genetic information from affected calves and their sire and dam, Dr. Beever was able to determine the genetic defect was a deletion of part of the gene (i.e. part is missing), to identify the original bull that introduced the defect and to develop a genetic test to determine if cattle carry the defective recessive gene. Knowing the TH status of our Shorthorn and Shorthorn influence cattle will allow us to make informed breeding decisions, which of course is in the best interest of our cattle and the breed.

The same genetic principles apply to the TH gene as the color gene. Let's call the normal TH gene "T" and the defective gene "t".

Table 4: Mating a TH carrier bull to a TH carrier cow. Three calves will look normal at birth, but two of the three will be carriers for TH. The fourth calf will be born with TH. Mating two TH carriers gives you a 25% chance of having a TH calf.

Table 4	T	t
T	TT	Tt
t	Tt	tt

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Figure 3 - Shorthorn calf with TH. Note the large abdominal hernia and the twisted back legs.

As seen in Tables 5 and 6, you cannot get a calf with TH (or any other recessive disease) unless both parents are carriers. Since TH is lethal, neither the sire nor the dam can have TH and pass it on, or to put it a different way, you cannot have a bull or cow with the genotype “tt”.

Table 5: Mating a TH carrier bull (Tt) to a normal cow (TT). All calves will look normal at birth, but two of the four will be carriers for TH (Tt). With this mating, 50% of the calves will be TH carriers.

Table 5	T	t
T	TT	Tt
t	Tt	tt

Table 6: Mating a normal (TT) bull to a TH carrier cow (Tt). All calves will look normal at birth, but two of the four will be carriers for TH. With this mating, 50% of the calves will be TH carriers.

Table 6	T	t
T	TT	Tt
t	Tt	tt

Currently, TH has been seen in Shorthorn cattle and Shorthorn influence cattle, including Maine-Anjou, Chianina and crossbreds. Any animal that traces back to the original Shorthorn bull imported to the US in the 1970's has the potential to be a carrier of the defective gene.

PHA - The Story Continues

When Dr. Hannon was finding TH calves, he also discovered some Shorthorn calves thought to have

TH actually had something different — PHA (pulmonary hypoplasia with anasarca). Translated, that means little, poorly formed lungs (pulmonary hypoplasia) and lots of excess, retained fluid (anasarca). This is also a lethal defect and the calves are usually born dead. Calves may also be aborted early. The anasarca is a major problem. Calves can retain so much fluid that an 80 pound calf can weigh 200 pounds! PHA often causes dystocia and may require a C-section. One cow with a PHA calf had to be shot. Even with a C-section they were unable to remove the calf. This is a bad deal.

In PHA, like TH, the defective gene is recessive, meaning both parents appear normal (phenotype) but both parents carry the defective recessive gene (genotype). If you have a cow that has a PHA calf, you know both the sire and dam are carriers of the defective gene (see Table 7). Back in the old days, before genetic tests, genetic defects were determined by “test breeding.” This means taking an animal that had a defective offspring and breeding it to animals that did or did not have affected offspring. Basically, that is where we are now with PHA in “field tests.” Our pastures and our cattle are the research laboratory! We know it is recessive, but to determine which gene is defective and what the defect is we need samples from PHA calves, their sire and dam and their pedigrees. This information is required to develop a test for PHA carriers.

Table 7: Mating a PHA carrier bull to a PHA carrier cow. Three calves will look normal at birth, but two of the three will be carriers for PHA. The fourth calf will be born with PHA. Each time you mate two PHA carriers there is a 25% chance of having a PHA calf.

Table 7	P	p
P	PP	Pp
p	Pp	pp

To date, PHA has been seen in Shorthorn, Maine-Anjou, Chianina and crossbred cattle. In all cases,



Figure 4 - Maine calf with PHA. Note the huge size of the calf and the swollen appearance.



Figure 5 - Head of Maine calf with PHA. Note the complete loss of detail secondary to the fluid accumulation.



Figure 6 - Lungs from a calf with PHA. Note the small size and the texture that looks more like liver than lung.

it seems to trace to Maine-Anjou bloodlines. Interestingly, the bull considered responsible for passing the defective gene to Shorthorn cattle is not believed to be the animal(s) primarily responsible for the defect in Maine cattle.

The Plot Thickens

We now know the gene responsible for TH is different from the gene responsible for PHA. This means a Shorthorn bull with Maine influence can be a carrier for both TH and PHA, TH but not PHA, PHA but not TH or not carry either defective gene! It also means a Maine bull with Shorthorn influence (or a Chianina or crossbred bull with Maine and Shorthorn influence) can be a carrier for TH and PHA, TH but not PHA, PHA but not TH or not carry either defective gene! The same is true for cows!

The Bottom Line

Knowledge is power, and knowing the genotype of our cattle will help us make breeding decisions that are good for our operation, our calves, our bottom line and our breed. How you approach this depends on your goals, plans, market and the amount of risk you are willing to take with your cow herd and your calves. TH

results in a dead calf, but usually the cow is OK. PHA can kill a cow. Since we have a test for TH, we should use it! Thousands of Shorthorn cattle have been tested and the results are listed on the American Shorthorn Association Web site (www.shorthorn.org). TH negative bulls are also listed on www.showsteers.com (this list is at the owners request, has been verified

by Dr. Beever, but not necessarily parentally verified).

Right now, PHA is a little more complicated. We do not have a genetic test. Help us develop a test by supplying samples from the sire and dam of any PHA calf born on your farm (samples from the calf too). If you have a PHA calf, you know the sire and dam are both carriers, repeating that mating means a 25% chance of getting another PHA calf. Is it worth the risk?

TH and PHA - The Facts

- TH and PHA are two distinct genetic conditions
- TH and PHA are the result of two different genes that are abnormal
- TH and PHA are both considered autosomal recessive
- Cattle with TH trace to Shorthorn breeding
- Cattle with PHA trace to Maine-Anjou breeding
- An animal with both Shorthorn and Maine influence can be a carrier of both, neither or one of the abnormalities
- A calf born with TH or PHA received one abnormal gene from each of its parents
- The sire and dam of a calf born with TH are both carriers for TH
- The sire and dam of a calf born with PHA are both carriers for PHA
- There is a genetic test for TH - it can tell you if your animal is normal (not a carrier for TH; TH free) or a carrier for TH (TH carrier)
- There is not yet a genetic test for PHA, but we know the parents of a PHA calf are carriers of the defective gene

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