



DEAD COWS DON'T LIE!! MOO NEWS TELLS YOU WHY!

Top ten things you need to know about lethal recessive genetic defects in cattle

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Over the past five or more years, we who breed cattle have been bombarded with a bunch of lethal recessive genetic defects. No breed or cross seems immune. We have TH, PHA, AM, NH, IE, OS, LND, FCS and MA*. With all these initials, how can we keep them straight? How can we breed defect free cattle? How do we make breeding decisions so we do not have defective calves? From talking to people and answering these questions, it seemed like it was time for a bit of a review ...

Genes are the biological blueprints that make us what we are. Genes are stacked on chromosomes in an orderly, linear fashion. Chromosomes are located in the nucleus of the cell. In cattle, the genes for horn development and milk yield are on chromosome 1, and they are always in the same place on chromosome 1. Cattle have 30 pair of chromosomes (60 chromosomes total) including 29 pairs and the sex chromo-

somes - either XX or XY. Half of each pair is inherited from the dam, half from the sire. The dam can only contribute an X, the sire either an X or a Y. XX is a female; XY is a male. So, basically the bull determines the sex of the calf!

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 the sire and the dam provide input to coat color and pattern.

Most genetic disorders in cattle are recessive. Recessive disorders mean that for the calf to be born with the disorder it must have inherited one defective gene from the sire and one defective gene from the dam. BOTH the sire and dam contributed a defective gene to the calf.

Understanding the inheritance of lethal recessive genetic disorders is no

different from understanding the inheritance of red in black Angus cattle. For example, the sire is black, the dam red. That is "phenotypic" information - phenotype is the trait the animal exhibits, or what you see. Genotype is the genetic information in the chromosomes. The genotype determines the phenotype. There are 2 variations in the genes for coat color - the different variations are called alleles. Black is the dominant allele and 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 that the red dam is homozygous red (both alleles code for red). If the calf from this mating is red, we know the bull carries a red allele (i.e. he is heterozygous black). If the calf is black, we don't know if his sire is homozygous or heterozygous black. **Since red is recessive, for a calf to be born red, BOTH parents must carry the red gene and BOTH parents must give that gene to the calf.** The same is true for lethal recessive defects. For the calf to be born affected, both parents must carry the mutation and BOTH parents must give the mutation to the calf.

Examples using coat color and the Punnett square illustrate the situation with recessive defects the best. For these examples, **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. A homozygous black bull (**BB**) does not have the red allele. A heterozygous black bull (**Bb**) is a carrier of the red allele. In the Punnett square, the bull's genotype is shown across the top, and the cow's genotype down the left. The 4 boxes show all potential genotype

Recessive Defects - What You Should Know

1. All recessive defects are inherited the same way
2. Breeding a non carrier to a non carrier (free to free) results in ZERO chance of having an affected calf.
3. Breeding a non carrier to a non carrier (free to free) results in ZERO chance of having a carrier calf.
4. You can only get an affected calf if you breed a carrier to a carrier.
5. If the calf is affected, both parents MUST be carriers and both passed the mutation on to the calf.
6. When you breed a carrier to a carrier, the risk of having a carrier calf is 50%.
7. When you breed a carrier to a free, the risk of having a carrier calf is 50%.
8. When you breed a carrier to a free, the risk of having an affected calf is ZERO, however the risk of having a free calf is the same as that of having a carrier - 50%
9. The risk (chance) of a certain outcome is the same for each mating - it is independent of previous outcomes.
10. The risk of having a dead calf is 1 in 4 every time you breed carrier to carrier

combinations 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.

	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**). 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 the recessive gene from both parents. The same is true for calves born with recessive diseases; the calf must inherit the gene from both parents.**

Table 2: Mating a heterozygous black bull to a red cow.

	B	b
b	Bb	bb
b	Bb	bb

If a calf gets a recessive gene from both parents, the calf will show (express) the characteristic in their phenotype (i.e. they will be red). Now, let's say that 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 - 3 of 4 calves will be born black, but only 1 is homozygous black (**BB**); 2 of 4 are heterozygous black (**Bb**), and 1 of 4 is homozygous red (**bb** - 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.

	B	b
B	BB	Bb
b	Bb	bb

As Table 3 indicates, in mating a heterozygous black bull to a heterozygous black cow, there is a 25% chance that the calf will be homozygous black, a 50% chance that the calf will be heterozygous black, and a 25% chance the calf will be red. It is important to note that the Punnett square tells us the risk or average chance of having a certain genotype for each mating. It does not tell us that if we have 3 black calves the 4th will automatically be red. Like tossing a coin, the chance of heads is 50% every time.

Now let's take the same principles and apply them to recessive genetic defects. All these recessive defects are inherited just like the red coat color. Since all the defects are inherited in a similar fashion, we will use **F** = the normal gene and **f** = the defective recessive gene. The capital **F** is dominant, and the lower case **f** is recessive. A normal looking animal (cow or bull) can be homozygous for the normal gene (**FF**) or can be a carrier of the mutation (**Ff** - heterozygous for the normal gene). Since these are lethal recessive defects, neither the sire nor the dam can be homozygous for the defect (i.e. neither can be **ff**). If they were **ff** they would be dead and therefore unable to mate. So, we are dealing with phenotypically normal looking animals that are either free (**FF**) or carriers (**Ff**) of the lethal recessive genetic defect. Therefore, there are three possible scenarios...mating free to free, free to carrier, or carrier to carrier.

Free to free is easy - all calves will be normal in phenotype and genotype (**FF X FF = FF** only). It gets more complicated when we mate a free (**FF**) to a carrier (**Ff**), or carrier (**Ff**) to carrier (**Ff**). In the first example (Table 4), the sire is free (**FF**) and the dam is a carrier (**Ff**). The results would be exactly the same if the sire was the carrier and the dam was free.

Table 4

	F	F
F	FF	FF
f	Ff	Ff

Table 4 shows that when you breed a free to a carrier, there is a 50% chance of having a carrier calf with each mating. There is also a 50% chance of having a free calf. In other words, with every

free to carrier mating there is a 1 in 2 chance (50%) that the calf will be a carrier. There is zero chance of having an affected calf. **For the recessive gene to be displayed in the phenotype (i.e. for you to see it) the calf must inherit the recessive gene from both parents.**

What happens if you mate a carrier (**Ff**) to a carrier (**Ff**)? The Punnett square in Table 5 will tell you. Both the sire and dam in this example are carriers for a lethal recessive defect.

Table 5

	F	f
F	FF	Ff
f	Ff	ff

In Table 5, there is again 50% chance of having a carrier calf with each mating (**Ff**) - however there is only a 25% chance of having a free calf (**FF**) and there is also a 25% chance of having an affected dead calf (**ff**). **For the recessive gene to be displayed in the phenotype (i.e. for you to see it) the calf must inherit the recessive gene from both parents. So if both parents are carriers, there is a one in 4 chance of having a dead calf.**

So what is the bottom line? Anytime you use a carrier animal for breeding, the risk of having a calf that carries the recessive mutation is 50%. It doesn't matter if you breed carrier to carrier or carrier to free - 50% of your calf crop, on average, will be carriers. The danger comes when you breed carriers to carriers - 50% of your calf crop, on average, will be carriers and 25% of your calf crop, on average, will be dead. You need to decide if the risk is worth it.

Note: for simplicity we are calling these lethal recessive defects, however, calves with FCS, IE, LND, FCS, and MA can be born alive and live for some time. Calves with TH, PHA, AM, and NH are usually born dead, and if alive, do not survive; calves with OS are aborted early. TH = tibial hemimelia; PHA = pulmonary hypoplasia with anasarca; AM = arthrogyrosis multiplex; NH = neurohydrocephalus; IE = idiopathic epilepsy; OS = osteopetrosis AKA marble bone; LND = long-nosed dwarf AKA long head dwarf; FCS = fawn calf syndrome; and MA = alpha mannosidosis