Monitoring Abnormalities Online

Mutations/Genetic Defects

How to locate the site: www.simmental.org/
Select ***"Genetics"*** on left side menu Select ***"Genetic Defects"***

Charles Darwin observed that changes continuously occur in populations and that these changes are critical to the success of the species. The changes Darwin noted were caused by genetic mutations, i.e., a change in the genetic code from what previously existed. Though Darwin was focused on the advantageous changes brought about by mutations, we now know that the majority of mutations in nature are not advantageous. In fact, they tend to hinder (be deleterious to) a population's success — and a few of the mutations are quite harmful (e.g., dwarfism) or even lethal (e.g., hydrocephalus). Mutations of this nature are often referred to as genetic defects.

Although most mutations are deleterious, the fact that mutations continuously occur is critical to the success and even the survival of a population; though rare, the occurrence of a favorable mutation provides the population with a building block that may allow it to adapt to a changing environment or become more competitive in a stable environment.

For example, though likely not favored in the wild, mutations resulting in increased docility or earlier puberty are sought after in domesticated cattle. The polled gene is an example of a single mutation that became very desirable when cattle were domesticated.

Desirable mutations tend to increase in frequency, while deleterious mutations are eliminated or exist at low frequency in a population — but their frequency and the speed at which they are eliminated or integrated has much to do with whether they are dominant or recessive in their expression.

Dominant mutations that are highly advantageous (polled for example) spread very quickly through populations, while unfavorable dominant mutations are eliminated in short order. This is because dominant genes always influence an animal's phenotype and therefore the mutation can easily be selected for or against. Recessive mutations, however, tend to exist in a population even when deleterious to the point of being lethal. This is because animals can carry the deleterious recessive without showing any signs of it.

Fortunately, technology has evolved to the point where animals that appear normal yet are carriers of recessive genetic defects can be identified via DNA testing. Tests of this nature are currently available for many genetic defects and many more are certain to follow.

These tests are able to determine the presence of a single recessive gene that causes the abnormality. As mentioned earlier, carrier animals appear normal; however, when mated to other carriers the resulting offspring have a chance of showing symptoms.

A 2 x 2 Punnett Square can be used to illustrate the outcome of various matings. In the two examples below we have mated a male carrier to a female carrier and a non carrier in scenarios A and B, respectively.

A) Dam (top row); Sire (left column)

	N	n
N	NN	Nn
n	nN	nn

B) Dam (top row); Sire (left column)

	N	N
N	NN	NN
n	nN	nN

In these examples, N is the normal gene while n is the abnormal recessive. The cells with single letters contain one copy of each of the sire's (left column) and dam's (top row) genes. Since we have used a male that carries the abnormal gene (n) in both examples each Punnett Square has an N and n on the sire side. As explained earlier, in example A we have mated the sire to a carrier female (Nn) while in B he is mated to a non carrier female (NN).

Through the use of Punnett Squares we can readily visualize what the resulting offspring will look like from our example matings. In scenario A we can see that the 4 potential genotypes from the mating are NN, Nn, nN and nn — each with an equal probability (1/4) of occurring. Since the presence of N has complete dominance over the expression of n (i.e., N completely covers up the symptoms of n) we know that only the calf receiving nn will show the symptoms of the abnormal gene; the other 3 will appear normal. Because they received the n gene (Nn and nN); however, 2 of the 3 normal calves in appearance will be carriers of the abnormal gene. In example B we can see that all of the resulting offspring will appear normal, while half of them (2 of 4) will be carriers. The above examples also work to illustrate other situations where a single recessive is involved, such as polled/horned or red/black. ◆