The Effects of Genetic Testing: Constructive or Destructive?

A genetic test that should be

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By Jerold S. Bell, DVM, Tufts Cummings School of Veterinary Medicine (This article originally appeared in the June, 2001 issue of the AKC Gazette)

limiting it.

Every breed has genetic disorders. Finding tests that identify carriers of the genes which cause these disorders is a goal in all breeds. Once a genetic test is found, however, it is a double-edged sword: Its use can enable breeders to improve a breed or devastate it.

Without genetic tests, the number of

that can doas be identified as carriers is low, even though many dogs may be suspected of being carriers because they have relatives that are known to be

Without tests. affected. though. control involves genetic-disease breeding higher-risk dogs to lower-risk dogs. Dog breeds have closed gene pools; in other words, the diversity of genes in a given breed is fixed. The dogs removed from number of consideration for breeding based on concerns regarding a specific genetic disease is usually low, and therefore does not greatly alter the breed's gene pool, or diversity.

However, once a genetic test is developed that allows

breeders to positively determine if a dog is a carrier of a defective gene, many owners are likely to remove carrier

dogs from their breeding stock. Although doing so is human nature, this temptation must be overcome. Any guality dog that you would have bred if it had tested normal should still be bred if it tests as a carrier.

In such circumstances, carriers should be bred to normal-testing dogs. This ensures that affected offspring will not be produced. Carrier breeding stock should be subsequently replaced with normal-testing offspring that exceeds it in quality. If the only quality offspring is also a carrier, then use that offspring to replace your original carrier. You have

> improved the quality of your breeding stock, even though the defective gene remains in this It is generation.

certainly true, though, that the health of the breed does depend on diminishing the carrier frequency and not increasing You should therefore limit the it. number of carrier-testing offspring that you place in breeding homes. This does not mean, however, that you should prevent all of them from being bred. It is important to carry on lines. A genetic test that should be used to help maintain breed diversity should not result in limiting it.

Consider All Aspects

We know that most dogs carry some unfavorable recessive Any quality dog that you genes. The more

would have bred if it had genetic tests that are developed, the greater tested normal should still be chance there is of bred if it tests as a carrier. identifying an undesirable gene in your dog. Remember, however, that your dog is not a single gene, an eye, a hip, or a heart. Your dog carries tens of thousands of genes, and each dog is a part of the breed's gene pool. When considering a breeding, you must consider all aspects of the dog - such conformation, as health issues, temperament and performance - and weigh the pros and cons. When a good-quality dog is found to carry a testable defective gene, there is a better option than removing that dog from your breeding program. That option is to breed it, so that you can keep its good qualities in the gene pool, and then replace it in your program with a normal-testing dog.

There are breeders who contend that no more than 10 percent of carrier dogs should be removed from breeding in each generation. Otherwise, they say, the net loss to the gene pool would be too great. In fact, less than 10 percent of all dogs in a breed are ever used for breeds do breeding. Dog not propagate according to what is known as the Hardy-Weinberg equilibrium, where all members of a group reproduce and pass on their genes to the next generation. Breeders already place tremendous pressure on their gene pools through selective breeding decisions. Indeed, breeders who focus their selective pressure on the more elusive traits in their dogs, rather than on testable and predictable single-gene conditions, are right to do so.

The Dangers

It is important that breed clubs educate their owners on how genetic tests should be properly interpreted and used. History has shown that breeders can be successful in reducing breedwide genetic disease through testing and making informed breeding choices. You should remember, however, that there are also examples of breeds that have actually experienced more problems as a result of unwarranted culling and restriction of their gene pools.

These problems include: reducing the incidence of one disease and increasing the incidence of another by repeated use of stud dogs known to be clear of the gene that causes the first condition; creating bottlenecks and diminishing diversity by eliminating all carriers of a gene from the pool, instead of breeding and replacing them; and concentrating on the presence or absence of a single gene and not the quality of the whole dog.

Breeders are the custodians of their breed's past and future. "Above all, do no harm" is a primary oath of all medical professionals. Genetic tests are powerful tools, and their use can cause significant positive or negative changes. Breeders should be counseled on how to utilize test results for the best interests of the breed.

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Removing the stigma of genetic disease

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Reducing

genetic

raising

Dealing

effort.

An inevitable consequence of breeding is the occurrence of genetic problems. No one wants to produce affected dogs, yet some breeders and owners are quick to assign blame. There are no perfect dogs, and all dogs carry some detrimental genes.

The emotional reaction to producing a dog with a genetic disorder often follows what is called the grief cycle:

* Denial: This isn't genetic. It was caused by something else.

* Anger: This isn't right! Why is this happening to my dogs?

* Bargaining: My dog sired more than 100 other dogs that are healthy. So this one doesn't really count, right?

* Depression: My kennel name is ruined. No one will breed to my dogs.

* And, finally, acceptance: My dog was dealt a bad genetic hand.

There are ways to manage genetic disorders, breed away from this, and work toward a healthier breed.

Getting beyond denial

Unfortunately, many breeders can't get beyond the denial stage. Some will hold to increasingly improbable excuses, rather than accept that a condition is genetic. They will falsely blame relatively rare disorders on common viruses, bacteria, or medications. The fact that these organisms or drugs are common to millions of dogs annually who do *not* have these disorders is not considered. Some owners state that their veterinarian recommended not sending in a hip radiograph to the Orthopedic Foundation for Animals (OFA) because the dog would probably be diagnosed with hip dysplasia.

> Then these owners lull themselves into believing that since the dog wasn't evaluated, it does not have hip dysplasia. The fact that a dog does not have an official diagnosis does not mean the dog has normal hips, "not affected" with hip

dysplasia.

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It is important to confirm diagnoses of with genetic disorders blood tests. radiographs, pathology or specimens. However, the primary concern should always be for the individual dog. If an affected dog is not suffering, it should not be euthanized simply to obtain a pathological diagnosis. The increased availability of noninvasive techniques has made diagnoses easier to obtain.

Once confirmation of a genetic disorder is made, denial sometimes becomes deception, which is not acceptable. There are breeders who actively seek to prevent diagnoses and later necropsies, but who eventually realize those actions are detrimental to the breed, and in the long run to themselves.

Working together to improve our breeds

Reducing the stigma of genetic disease involves raising the level of conversation from gossip to constructive communication. Dealing with genetic disorders is a community effort. Each breeder and owner will have a different level of risk or involvement for a disorder. We do not get to choose the problems with which we have to deal. Breeders should be supportive of others who are making a conscientious effort to continue breeding their dogs while decreasing the risk of passing on defective genes.

Breeders should follow up on the puppies have placed. Breeders they should periodically contact their puppy buyers and ask about the health of the dogs. Some breeders fear they will be castigated if a dog they placed develops a problem. However, the vast majority of owners of affected dogs are *pleased* that their breeder is interested in their dog, and in improving the health of the breed so that other affected dogs are not produced.

A breeder cannot predict or prevent every health problem. If an owner's dog is discovered to have a problem, show your concern.

Breeders and breed clubs should be cooperative and supportive of researchers studying genetic disorders in their breed. Through research funded by breed clubs and by the AKC Canine Health Foundation (CHF), new genetic tests for carriers of defective genes are continually being developed.

The Canine Health Information Center (CHIC; www.caninehealthinfo.org) was established by the CHF and the Orthopedic Foundation for Animals (www.offa.org). CHIC is an online registry that works with the breed parent clubs to establish a panel of testable genetic disorders that should be screened for in each breed. The beauty of the CHIC concept is that dogs achieve CHIC certification by completing the healthchecks. Passing each health test is not a requirement for certification. CHIC is about being health conscious, not about being faultless.

My hope for each breed is that there will eventually be so many tests for defective genes that it will not be possible for any dog to be considered "perfect." Then we can put emotions aside and all work together on improving our breeds.

Breeders must lead the way to remove the stigma of genetic disorders. The applications for both the OFA and CHIC health registries include options that allow for open disclosure of all health-test results or semi-open disclosure listing only normal results. It is up to breeders to show that they are ready to move genetic disorders out of the shadows and check off the boxes for full disclosure.

More and more national clubs are having health seminars and health screening clinics at their specialties. This shows those breed clubs and breeders care about the genetic health of their breeds, and are working toward a healthier future.

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