

**CONVERSATION WITH DR. JEROLD  
S. BELL ON HOW BREEDERS CAN REDUCE  
GENETIC DISEASE IN OUR BREED**

**January, 2001.**

Dear Dr. Bell,

My questions concern that group of dogs in my breed that belong to the hobby/show breeders. At this point in time, I would guess, that the pedigrees of the dogs owned and shown by this group of breeders may be a gene pool of only a thousand dogs even though the breed has a much larger number of annual registrations.

Mine is a breed that was originated almost 150 years ago by inbreeding to develop type the first 50 years. The next hundred years linebreeding /inbreeding was strongly advocated to refine type. Now, looking at the future of the breed, we see a number of genetic diseases that are rapidly increasing in prevalence although none of these probably exceeds 10% of the population in this breed.

Over the past twenty-five years we have had several popular sires. Today, there is hardly a six generation pedigree that does not contain the names of at least a couple of these dogs. And many times dogs are there multiple times. The popular sires have all sired 30 - 50 champions, and their sons and grandsons continue to do the same. The time when breeders maintained individual kennels and bred to their own and their neighbor's dogs is long past. Today we fly frozen semen from the top dogs across the United States. Everyone is looking for that outstanding puppy that will become next year's popular sire.

Simultaneously with the increased use of popular sires has come an increase in genetic disease. This is reflected in the breed's health survey percentages of each disease.

1. Should we be treating this show population as an "endangered species" population and follow the guidelines that scientists have set forth for these populations? (We know that when a population becomes too small and genetically homogeneous it may go extinct because of genetic problems.)
2. Would you propose a breeding plan for a breed that would decrease the amount of genetic disease and increase the health of the breed (without doing any genetic testing)? Can we do this through screening tests and selection alone?
3. Have any breeds really been successful in reducing genetic disease?

Sincerely,

Trudy Sample

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**DR. BELL'S ANSWERS:**

This e-mail raises many important points, and also applies to the Ins & Outs article. As this is the last piece of mail left over from the question- fest, I am going to respond to it in depth. Many breeds are seeing an increase in frequency of breed specific genetic disease. While some of this is due to better diagnostics, I

believe the frequency of such disorders in many breeds has gone up. Ms. Sample states that none of the disorders probably exceeds 10% of her breed.. I am assuming that she is referring to the affected dogs in the population. Most of the problematic genetic disorders are either recessive.

or polygenic with a major recessive component. (There are dominants with incomplete penetrance, but they are in the minority.) With recessive disease, the number of carriers in the population far outnumber the affected dogs. Time out for one of my pet peeves.

Some breeders and even geneticists use the Hardy-Weinberg law to extrapolate the frequency of carriers in the population based on the number (frequency) of affected dogs observed. This is a proper use of the HW law in populations at equilibrium in the absence of selection, migration, or genetic drift. This means that all offspring are allowed to reproduce, and mates are chosen at random. Domestic animal breeding does not allow equilibrium or the other random factors to satisfy the use of the Hardy-Weinberg equation. If some breeders are linebreeding on carriers, they can see high numbers of affected, even if the carrier state is not at the HW high level to support that number of affected. If people are using breeding practices to prevent affected dogs from being produced, the HW law can compute a carrier frequency far below what actually exists. The results of genetic screening programs show that Hardy-Weinberg relations of carrier to affected do not uniformly exist in domestic animal breeding populations. Therefore, while we can state that the carrier population far exceeds the affected population, we cannot predict a carrier frequency without genetic screening or pedigree analysis. OK, back to the e-mail response.

The problem with increasing genetic disease, and the perception of decreased genetic diversity in dog breeds is from the popular sire syndrome. If popular sires carry deleterious recessive genes, then these will be spread into the gene pool through their descendants. This is called the founders effect. I have seen genetic disorders become fixed and increase in frequency in breeds through the founders effect. This is not a problem only for breeds with small gene pools. I have seen it in some of the larger AKC registered breeds.

So, what is really causing the problem? Is it popular sires, or the variable use of inbreeding /linebreeding? I contend that the major problem is popular sires. No one says the problem is INBRED popular sires. Outbred popular sires are just as bad. When single individuals contribute their genes to the gene pool far beyond what is necessary (and that is a judgment call), it causes genetic drift (toward his genes, and away from less prolific dogs). When his offspring then become popular sires, the gene pool gets even smaller. It doesn't matter whether these popular sires are only used in outbreedings. As a matter of fact, doing so would only act to spread his genes faster, and eliminate "lines" that are free of his genes.

As I stated in the article, it is the variation in what each breeder selects for that fosters genetic diversity in a breed. Everyone going in only one direction will cause a loss of diversity. Now, let's digress again:

Up to now, molecular genetic studies on dog breeds have shown that there is a lot more genetic diversity present than we expected to find. I have yet to see a dog breed that is actually suffering from the kind of inbreeding depression seen in captive endangered animals. There are many breeders in many large gene pool and small gene pool breeds that do intensive inbreeding generation after generation. Many of these breeders do not see smaller litter size, decreased immunity, or other signs of inbreeding depression. They may see increased expression of genetic disease. I personally feel many of these breeders believe that inbreeding is a goal, and not a tool. In other instances, we see that even mild linebreeding increases genetic disease or smaller litter size. It is my contention that inbreeding depression is not due to overall homozygosity, but the presence of deleterious recessives. When creating inbred lines of research mice, many do not survive, but those without deleterious genes are very hardy at greater than 99%

homozygosity.

Still, it is not too soon to think about the long term results of our breeding decisions, and what we can do to maintain more diversity. I do not believe that an extremist approach of only linebreeding being the right way to breed or only outbreeding being the right way to breed is correct. We want diversity in breeds. However, it is not important for every member of the breed to be the most diverse (outbred, heterozygous) for a breed to be healthy. In many animal conservation schemes, you want to have linebred lines (that have been bred best to best), that you can outbreed into to infuse new genes and vigor. If everyone outbred every generation, you would rapidly lose any unique lines to be able to outbreed to. Selection and utilizing the different breeding methods available, depending on your needs for the next generation is the most reasonable method to improve your breeding stock.

Sincerely,

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Dr. Bell's article "The Ins and Outs of Pedigree Analysis" can be found online at :  
<http://www.compuped.com/bell.htm>

**[Go to Part Two of Conversation with Dr Bell](#)**