

It is a pleasure to welcome Dr. Jerold Bell back to our pages. This article originally appeared in the proceedings of the 2017 AKC Canine Health Foundation National Parent Club Canine Health Conference.

A large number of individual dogs in a breed population allows greater choices when making breeding decisions. Multiple breed “family lines” support greater breed diversity: the genetic difference between individuals in the breed. When selecting on several different traits or disorders, a large population should allow for several choices of mates that fulfill

different selection preferences. A goal of all breeds is to grow and maintain a large, diverse, and healthy population.

All breeds originate from a small population of either related dogs or dogs who share a common conformational, behavioral, or working phenotype. Through selection, a breed standard is developed.

Individual dogs that do not adhere

Breeding by the Numbers



Bearded Collie Bailie of Bothkennar (r.) with Jeannie of Bothkennar and their pups in a composite photo created by Thomas Fall Studio. Dr. Bell reminds us that Bailie “contributes 32.6 percent of his genes to every modern Beardie.”



This Bichon Frises share, on average, 17.5 percent of its genes with Pitou, a dog born in 1924.

The aspect of population size on healthy breeding in dog breeds

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FAR LEFT: AKC GAZETTE COLLECTION; RIGHT: DAVID WOO ©AKC

to the standard or who demonstrate deleterious traits or disorders are purged from breeding. Those individuals who demonstrate and propagate desirable characteristics will have an increasing influence on the gene pool through multiple generations of descendants. Once breed characteristics are fixed in the population, it can go through an expansion stage where the population grows.

All breeds will have several influential ancestors that appear far back in pedigrees, but pass on a high percentage of their genes to every individual in the breed. For example, all Bichons Frises share on average 17.5 percent of their genes with Pitou (born in 1924), which is between the contribution of a grandparent and great-grandparent. He does not appear on average until the 16th generation, but appears over four million times in every Bichon pedigree and 38 percent of his alleles have been retained in the breed population. Bearded Collie Bailie of Bothkennar was born in the 1940s, and contributes 32.6 percent of his genes to every modern Beardie.

This process of breed evolution



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causes a loss of genetic diversity through the purging of undesirable individuals and the concentration of genes of influential ancestors. All breeds are partial clones of their influential ancestors. This is an expected consequence of breed evolution and is not detrimental to the breed.

Genetic disorders can be due to ancient disease liability genes that preceded breed formation and are shared by many breeds, or by recent mutations that cause breed-specific disease. These can originate from a random mutation and be propagated through breed ancestors. Conversely, genes causing genetic disorders can be linked on a shared chromosome to a selected trait (for

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example, hyperuricosuria and Dalmatian spotting), or genetic disorders can be caused by direct selection for disease-causing phenotypic traits (such as brachycephalic obstructive airway disease).

IS POPULATION SIZE DIRECTLY CORRELATED TO BREED HEALTH?

Evidence from registration figures and valid breed-health surveys show that the size of a population does not determine whether the breed will suffer from higher frequencies of genetic disease. There are many large-population breeds with high frequency genetic disorders, and many small-population breeds that show excellent health. In a small-population breed, individual mating choices and individual litters have a greater effect on the breed frequency of disease liability genes because they represent a larger percentage of the total gene pool. *It is the lack of selection for genetic health in either large- or small-population breeds that allows the propagation of genetic disorders.* Breed genetic health depends on selection against disease liability genes regardless of the size of the population.

DOES A LARGE POPULATION AUTOMATICALLY CONFER GENETIC DIVERSITY?

When analyzing entire breed-population databases back to founders, every dog breed—regardless of its population size—has the same findings; high homozygosity and low effective population size (minimum number of ancestors explaining the complete genetic diversity of a population). These are necessary and expected consequences of breed formation and evolution. As a breed gene pool expands, the average recent generational relationship (inbreeding and kinship) between mates can decrease. However, the average total generational relationship between dogs back to founders does not decrease. Breeds with small populations look the same as breeds with large populations did much earlier in their evolution and development.

In both large- and small-population breeds, genetic diversity can be lost if breeders do not utilize dogs from the breadth of the gene pool. This is most evident in the popular-sire syndrome. This can be compounded when a popular sire is



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FIELD SPANIEL/COURTESY LYNDIA BEAM



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replaced by a popular son, who is replaced by a popular grandson, and the entire breed truncates on a single popular-sire line. This causes a loss of genetic diversity from the breadth of the gene pool that would be propagated from other quality male lines.

Another issue with popular sires is

that their genetic contributions can only be evaluated after their prolific breeding period is over, and their genes have already been disseminated throughout the gene pool. Many recently identified genetic disorders that rise in frequency in a breed are caused by genes carried by

popular sires. This is different from an influential ancestor, whose qualities and influence are constantly evaluated every generation. If an influential ancestor's descendants are not producing quality, then they are not bred and the ancestor's influence diminishes. With the popular sire

syndrome a breed population may expand in numbers, but if breeding is concentrated in only a portion of the gene pool genetic diversity will diminish.

Some breeds may lack enough health and vitality from the start, and these breeds collapse and do not

progress beyond the purging stage of development. Other breeds may have a robust and growing population, but due to other factors experience a population contraction and decline that could significantly eliminate the genetic diversity present in the gene pool. The recent economically induced decline and then rise in AKC registrations is not detrimental to a breed as long as it was a temporary slowing, and not a loss of breeding lines. Frozen semen is also an important hedge against the loss of diverse lines.

Population contraction is a serious detriment to breed genetic diversity if it includes the loss of diverse within-breed lines. In extreme cases, a breed may require opening up its studbook to bring new genes into its gene pool. However, most current dog breeds show acceptable genetic diversity and only require health-conscious breeding and population expansion to maintain their gene pools.

DO OUTBREEDING PROGRAMS IMPROVE GENETIC DIVERSITY AND GENETIC HEALTH?

Conservation geneticists versed in rare and endangered species have

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designed species survival plans (SSPs) that call for outbreeding: mating together animals that are least related to each other. The purpose of SSPs is to prevent the homozygous expression of deleterious recessive genes. However, natural species and artificially selected breeds have completely different, and in many instances completely opposite selection pressures and desired outcomes. SSPs call for using all available individuals in breeding and only outbreeding. Dog breeding calls for selection, which requires differences between prospective mates and therefore genetic diversity between individuals.

Outbreeding homogenizes the population by removing the genetic difference between individuals in the breed and making everyone “alike.” If two unrelated parents are bred together, the offspring make the two lines related. If an offspring is then outbred to a further unrelated line, their offspring make all of the lines related. Outbreeding is a self-limiting process as there will eventually be no unrelated dogs. In order to have selective pressure for positive traits and against negative

traits or disorders, there must be variation and genetic differences between individuals in the gene pool. This requires distinct family lines that are eliminated by outbreeding programs.

Thus, the basic conceptual point is, “What constitutes genetic diversity?” Is it the diversity within each dog (heterozygosity through outbreeding)? Or is it the diversity between each dog (maintaining diverse family lines)? These two concepts are diametrically opposed to each other and breeders and breed organizations must decide which is in the best interest of their breeds.

The genes causing common breed-specific genetic disorders have already been dispersed in breed gene pools. Therefore the chance of breeding two carriers together is based on the frequency of the deleterious gene(s) in the population, and not necessarily the type (outbreeding or linebreeding) of mating. Outbreeding propagates deleterious genes in the carrier state and randomizes the occurrence of genetic disease; the same as is seen with common genetic disorders in mixed-breed dogs. *The only way to select against specific genetic disorders is to*

specifically select against the causative or liability genes through direct genetic testing or phenotypic genetic screening.

ADDITIONAL FACTORS IN SMALL-POPULATION BREEDS

Small-population breeds have added issues because each mating has a much greater influence on the entire gene pool. If a breed has particular hereditary disorders at a higher frequency, mates should be selected that can minimize or lower the risk of producing these disorders. A quality higher-risk dog (closely related to affected) can be bred to a lower-risk dog and replaced with a lower-risk offspring. As this process is repeated, the carrier risk and deleterious gene frequency will diminish in the population. As most disorders are complexly inherited and have no tests for carriers, carrier risk must be based on knowledge of phenotypic pedigree depth (parents and grandparents) and breadth (littermates and littermates of parents).

Some breeders in small-population breeds are afraid to breed and possibly cause more disease.

However, if no breeding is going on, the breed will certainly become extinct. Mates must be selected that reduce the risk of producing genetic disorders. Breeders need to do their best to select for health and quality and then see what they produce.

In small-population breeds a greater number of offspring should be placed in breeding homes to expand the population. However, breeders of some small-population breeds try to constrain breeding and limit it only to themselves. This is a shortsighted attitude. Breeders should recruit and mentor puppy buyers to become thoughtful breeders. As a population expands, the choices of mates increase and the average recent relatedness of mates will decrease. Decreasing average recent generational inbreeding coefficients is a natural consequence of expanding populations utilizing the breadth of their gene pools. It does not need to be artificially manipulated. Breeders all doing something a little different with their mating choices—i.e., which individuals they are selecting, the types of matings utilized, and so

on—is what maintains breed genetic diversity. With health-conscious breeding, there are greater choices available to produce healthier offspring.

CONCLUSIONS

All breeds require expanding or large, stable breeding populations. Mates should be selected that represent the breadth of genetic diversity in the gene pool. It is mate selection and not the types of matings that they are involved in (linebreeding or outbreeding) that maintains genetic diversity.

Large- and small-population breeds show the same population indices of high homozygosity, low effective population size, and high relationship to influential ancestors. The difference between large and small populations is in the available choice of breeding individuals.

Health-conscious selection through breed-appropriate genetic screening of prospective breeding individuals is the most important aspect of improving and maintaining the genetic health of any breed, regardless of its population size.—**J.S.B.**