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Gene Pools

MAINTAINING AND IMPROVING BREEDS

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In order to understand how to maintain breeds, we have to understand the genetic forces that shape them. Natural species evolve through natural selection. Any genetic changes within a population that improve the chance of survival and ability to reproduce in the populated environment will be at an advantage and thrive. This results in a loss of genetic diversity through the disadvantaged. This loss is not detrimental to the population as it is directly related to increasing its superiority.

Dog breeds develop through artificial selection for desired phenotypes – what you can see in the dogs. These can include conformation, behavior, working ability and health. Most breeds originally started from either a small population of related founders, or as a population of unrelated dogs that conformed to a working or conformational phenotype. Some breed lines will be discarded over time due to genetic defects, or an inability to adhere to a standard. Regardless of the breed origin, generations of reproduction within a small population produce homozygosity (the fixation of gene pairs) through close breeding. This is what causes breeds to reproduce themselves with each generation.

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Genetic studies of dog breeds show that they lose on average 35% of their genetic diversity through breed formation. Genetic studies also document the increased homozygosity found in dog breeds. **Low effective population size (low number of founders) and high deep-pedigree inbreeding coefficients (homozygosity) are a natural and expected consequence of breed development.**

Breeds differ from natural populations in that only a small percentage of dogs reproduce to create the next generation. In a population sense, this represents a genetic bottleneck with each generation. Individuals chosen for breeding should represent the quality traits of the breed. Quality traits should not be lost through the absence of selection or the abandonment of quality lines.

> *"The popular sire syndrome is the single most influential factor in restricting breed gene pool diversity"*

Population expansion is an important aspect of breed maintenance. If the offspring of small population breeds are generally healthy their population can grow and expand. They are at stages of breed development where more populous breeds were earlier in their development. Breeders of small population breeds need to mentor their puppy buyers to expand their breeder base as well as the number of dogs.

Population expansion allows the creation of new "family lines." A larger population allows average relatedness of breeding pairs (based on recent generations) to be less than the prior generation. Population contraction is detrimental to breed maintenance due to the loss of quality breeding lines and genetic diversity. **Healthy breed gene pools require expanding, or large, stable populations.**

There are times when a lot of breeding is going on and registrations are increasing, and times (such as the recent past) when less breeding is going on. However, it is the offspring that reproduce (regardless if from prolific or limited-breeding parents) that contribute their genes to the next generation. Breeding quality dogs from different "lines" and areas of the gene pool prevents the loss of genetic diversity.

The popular sire syndrome is the single most influential factor in restricting breed gene pool diversity. When a breed is concentrating on a specific sire or multi-generational sire line, other quality male lines are abandoned. This causes a loss of genetic diversity to the breed gene pool in exchange for a rapidly increasing influence of the popular sire. Now is an important time to use frozen semen of quality dogs from the past to expand gene pools. Stored DNA (such as from the OFA CHIC repository) or semen can be used for breed-specific genetic testing that might not have been previously available.

All individuals carry some deleterious genes, which can increase in frequency with natural as well as artificial selection. More "lines" of naturally occurring species have died off due to genetic disorders or diminished fitness than those that have survived. As individuals propagate, deleterious mutations can become breed-related disease if they are disseminated and increase in frequency.

Studies show that some breeds have more issues of specific genetic diseases with linebreeding and others do not. This depends on the genetic load of deleterious recessive genes in the gene pool. **The genetic health of dog breeds is not a direct function of homozygosity, genetic diversity, or population size; but of the accumulation and propagation of specific disease liability genes.**

Artificial selection to maintain breeds requires active selection against deleterious genes. This is easier with dominant or additive genes, as the genotype is observed in the dog's phenotype. For recessive deleterious genes, selection involves the development and use of genetic tests that reveal the carrier state, or the identification of lines with carrier risk.

Some hereditary disorders and disease-predisposing phenotypes have been actively selected for by breeders. The most evident and widespread is the brachycephalic obstructive airway disorder, seen in extremely short-muzzled breeds. Other extreme phenotypes include excessive skin, excessive skin folds, excessive hind limb angulation, excessive size, excessive coat, dome-shaped skulls, and eyelid abnormalities. **It is important that breed standards and selection practices specifically avoid selection for extreme phenotypes that cause**

disease liability. For the show ring, judges' education should be directed towards rewarding moderation of disease-related extreme phenotypes.

Regular breed health surveys should be conducted by breed clubs to monitor for the presence and changing prevalence of genetic disorders. The OFA offers on-line health surveys for breeds. **Breed genetic health should be judged on breed health surveys that document the occurrence of genetic disease.**

Parent breed clubs should determine realistic pre-breeding genetic screening requirements based on the prevalence and severity of testable disorders in the breed. **Health testing requirements should be listed in the OFA CHIC and AKC Bred with H.E.A.R.T. program websites.**

Without direct selection against genetic disorders, the genetic health of breeds will decline. Breeders who refuse to do pre-breeding health screening should be directed to find a different hobby or profession that they can actually be good at. It is not ethical to breed dogs without selection for genetic health. **Selection of healthy breeding stock is the most important aspect of maintaining breeds.**

Each breeder must prioritize their selection for positive traits and against disease traits with each mating. Some breeders feel that genetic screening will reduce the genetic diversity of breeds. **The proper use of genetic screening actually increases breeding choices by allowing quality dogs at higher-risk of carrying disease liability genes to be bred:**

- Quality carriers of testable disease-causing recessive genes should be bred to normal testing mates and replaced for breeding with quality, normal testing offspring.
- Quality dogs with a less desirable phenotype (such as fair or even mild hip dysplasia in breeds with high frequencies of dysplasia) should be bred to dogs with desirable phenotypes (good or excellent hips) and replaced for breeding with offspring whose phenotype is better than the parent.
- Quality non-affected dogs from lines expressing disorders that do not have genetic tests (such as epilepsy) should be bred to mates from families or litters not expressing the disorder and replaced for breeding with a quality, healthy offspring.

In small population breeds with high frequencies of genetic disorders, breeders are often "frozen" from breeding for fear of producing disease. This causes continued breed decline due to population contraction. Breed improvement requires selection of the best breeding choices in matings that can reduce the frequency of genetic disease. As the population and breeding choices expand, the ability to reduce the frequency of disease expands with it. **Breed improvement involves; 1) selection of breeding dogs, 2) appropriate pairing of mates, 3) breeding, and 4) replacement of less desirable breeding dogs with more desirable offspring.**

An unfortunate development in dog breeding is recommendations designed for the preservation of rare and endangered species. These involve outbreeding (reducing homozygosity and average inbreeding coefficients) and increasing minor gene or chromosome segment frequencies. Dog breeding requires diverse lines, and not a homogenized and randomized outbred population. Outbreeding will not reduce the frequency of breed-related genetic disease, as the causative genes are already dispersed in the breed gene pool. Genetic selection for quality and against undesirable traits is what causes homozygosity and reduces the frequency of minor genes and chromosomal segments. Blindly selecting for them without knowing their effect could significantly reverse selection-based breed improvement. **Homozygosity is synonymous with pure breeds. It is not inherently correlated to impaired genetic health, and does not have to be artificially controlled.**

Expanding populations with different breeders undertaking different types of matings and selecting on different lines, while monitoring and selecting against genetic disease provides for a healthy, diverse breed gene pool.

Official genetic screening results should be made available to prospective breeders, and to the pet and breeding-stock purchasing public. This is facilitated through open genetic health databases like the OFA. It doesn't matter whether a breeder is a large commercial breeder, or only breeds once. It is no

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longer acceptable to say that genetic disease "just happens." In today's environment, not testing for documented breed-related hereditary diseases is irresponsible and unethical breeding. **Breed-specific pre-breeding health screening should become as universal as equine pre-purchase examinations.**

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BREED MAINTENANCE AND IMPROVEMENT REQUIRES:

- A large or expanding breed population
- Avoidance of the popular sire syndrome
- Avoidance of extreme phenotypes that can produce disease liability
- Monitoring of health issues in the breed
- Constant selection for quality and health