

Pedigree Analysis and How Breeding Decisions Affect Genes

Jerold S Bell DVM, Clinical Associate Professor of Genetics, Tufts Cummings School of Veterinary Medicine

To some breeders, determining which traits will appear in the offspring of a mating is like rolling the dice - a combination of luck and chance. For others, producing certain traits involves more skill than luck - the result of careful study and planning. As breeders, you must understand how matings manipulate genes within your breeding stock to produce the kinds of offspring you desire.

When evaluating your breeding program, remember that most traits you're seeking cannot be changed, fixed or created in a single generation. The more information you can obtain on how certain traits have been transmitted by your animal's ancestors, the better you can prioritize your breeding goals. Tens of thousands of genes interact to produce a single individual. All individuals inherit pairs of chromosomes; one from the mother, and one from the father. On the chromosomes are genes; so all genes come in pairs. If both genes in a gene pair are the same gene (for instance, "aa" or "AA") the gene pair is called homozygous. If the two genes in a gene pair are unlike (for instance, "Aa") the gene pair is called heterozygous. Fortunately, the gene pairs that make a cat a cat and not a dog are always homozygous. Similarly, the gene pairs that make a certain breed always breed true are also homozygous. Therefore, a large proportion of homozygous non-variable pairs - those that give a breed its specific standard - exist within each breed. It is the variable gene pairs, like those that control color, size and angulation that produce variations within a breed.

There are ways to measure the genetic diversity of a population. One method is to measure the average inbreeding coefficient (or Wright's coefficient) for a breed. The inbreeding coefficient is a measurement of the genetic relatedness of the sire and dam. If an ancestor appears on both the sire and dam's side of the pedigree, it increases the inbreeding coefficient. The inbreeding coefficient gives a measurement of the total percentage of variable gene pairs that are expected to be homozygous due to inheritance from ancestors common to the sire and dam. It also gives the chance that any single gene pair can be homozygous.

The types of matings that you choose for your breeding animals will manipulate their genes in the offspring, affecting their expression. Linebreeding is breeding individuals more closely related (a higher inbreeding coefficient) than the average of the breed. Outbreeding involves breeding individuals less related than the average of the breed. Linebreeding tends to increase homozygosity. Outbreeding tends to increase heterozygosity. Linebreeding and inbreeding can expose deleterious recessive genes through pairing-up, while outbreeding can hide these recessives, while propagating them in the carrier state.

Most outbreeding tends to produce more variation within a litter. An exception would be if the parents are so dissimilar that they create a uniformity of heterozygosity. This is what usually occurs in a mismatching between two breeds, or a hybrid, like a Cockapoo. The resultant litter tends to be uniform, but demonstrates "half-way points" between the dissimilar traits of the parents. Such litters

may be phenotypically uniform, but will rarely breed true due to the mix of dissimilar genes.

One reason to outbreed would be to bring in new traits that your breeding stock does not possess. While the parents may be genetically dissimilar, you should choose a mate that corrects your breeding animal's faults but complements its good traits. It is not unusual to produce an excellent quality individual from an outbred litter. The abundance of genetic variability can place all the right pieces in one individual. Many top-winning show animals are outbred. Consequently, however, they may have low inbreeding coefficients and may lack the ability to uniformly pass on their good traits to their offspring. After an outbreeding, breeders may want to breed back to individuals related to their original stock, to attempt to solidify newly acquired traits.

Linebreeding attempts to concentrate the genes of specific ancestors through their appearance multiple times in a pedigree. It is better for linebred ancestors to appear on both the sire's and the dam's sides of the pedigree. That way their genes have a better chance of pairing back up in the resultant offspring. Genes from common ancestors have a greater chance of expression when paired with each other than when paired with genes from other individuals, which may mask or alter their effects.

Linebreeding on an individual may not reproduce an outbred ancestor. If an ancestor is outbred and generally heterozygous (Aa), increasing homozygosity will produce more AA and aa. The way to reproduce an outbred ancestor is to mate two individuals that mimic the appearance and pedigree of the ancestor's parents.

Inbreeding significantly increases homozygosity, and increases the expression of both desirable and deleterious recessive genes through pairing up. If a recessive gene (a) is rare in the population, it will almost always be masked by a dominant gene (A). Through inbreeding, a rare recessive gene (a) can be passed from a heterozygous (Aa) common ancestor through both the sire and dam, creating a homozygous recessive (aa) offspring.

The total inbreeding coefficient is the sum of the inbreeding from the close relatives (first cousin mating), and the background inbreeding from common ancestors deep in the pedigree. Such founding ancestors established the pedigree base for the breed.

Knowledge of the degree of inbreeding in a pedigree does not necessarily help you unless you know whose genes are being concentrated. The relationship coefficient, which can also be approximated by what is called the *percent blood* coefficient, represents the probable genetic likeness between the individual whose pedigree is being studied, and a particular ancestor. It is a measurement of the average percentage of genes the individual and the ancestor should have in common.

We know that a parent passes on an average of 50% of its genes, while a grandparent passes on 25%, a great-grandparent 12.5%, and so on. For every time the ancestor appears in the pedigree, its percentage of passed-on genes can be added up

and its "percentage of blood" estimated. In many breeds, an influential individual may not appear until later generations, but then will appear so many times that it necessarily contributes a large proportion of genes to the pedigree.

The average inbreeding coefficient of a breed is a measurement of its genetic diversity. When computing inbreeding coefficients, you have to look at a deep pedigree to get accurate numbers. An inbreeding coefficient based on 10-generation pedigrees is standardly used, but requires a computerized pedigree database to compute.

The average inbreeding coefficient for a breed will be based on the age and genetic background of the breed. A mating with an inbreeding coefficient of 14 percent based on a ten generation pedigree, would be considered moderate inbreeding for a Labrador Retriever (a popular breed with a low average inbreeding coefficient), but would be considered outbred for an Irish Water Spaniel (a rare breed with a higher average inbreeding coefficient).

Most breeds start from a small founding population, and consequently have a high average inbreeding coefficient. If the breed is healthy and prolific, the breadth of the gene pool increases, and the average inbreeding coefficient can go down over time. Some dog breeds were established on a working phenotype, and not on appearance. These breeds usually start with low inbreeding coefficients due to the dissimilar backgrounds of the founders. As certain individuals are linebred on to create a uniform physical phenotype, the average inbreeding coefficient can increase.

There is no specific level or percentage of inbreeding that causes impaired health or vigor. If there is no diversity (non-variable gene pairs for a breed) but the homozygote is not detrimental, there is no effect on breed health. The characteristics that make a breed reproduce true to its standard are based on non-variable gene pairs. There are pure-bred populations where smaller litter sizes, shorter life expectancies, increased immune-mediated disease, and breed-related genetic disease are plaguing the population. In these instances, prolific ancestors have passed on detrimental recessive genes that have increased in frequency and homozygosity. With this type of documented inbreeding depression, it is possible that an outbreeding scheme could stabilize the population. However, it is also probable that the breed will not thrive without an influx of new genes; either from a distantly related (imported) population, or crossbreeding.

Fortunately, most breeds do not find themselves in the position of this amount of limited diversity and inbreeding depression. However, the perceived problem of a limited gene pool has caused some breeders to advocate outbreeding of all individuals. Studies in genetic conservation and rare breeds have shown that this practice actually contributes to the loss of genetic diversity. By uniformly crossing all "lines" in a breed, you eliminate the differences between them, and therefore the diversity between individuals. Eventually, there will not be any "unrelated line" to be found. Everyone will have a mixture of everyone else's genes. This practice in livestock breeding has significantly reduced diversity, and caused the loss of unique rare breeds.

A basic tenet of population genetics is that gene frequencies do not change from generation to generation. This will occur regardless of the homozygosity or heterozygosity of the parents, or whether the mating is an outbreeding, linebreeding, or inbreeding. This is the nature of genetic recombination. Selection, and not the types of matings used affect gene frequencies and breed genetic diversity.

If two parents are both heterozygous (both Aa) for a gene pair, on the average, they would produce 25% AA, 50% Aa, and 25% aa. (These are averages when many litters are combined. In reality, any variety of pairing up can occur in a single litter.) If a prolific male comes out of this litter, and he is homozygous aa, then the frequency of the "a" gene will increase in the population, and the frequency of the "A" gene will decrease. This is known as the popular sire syndrome. Of course, each individual has thousands of genes that vary in the breed, and everyone carries some deleterious recessive genes. The overuse of individual breeding animals contributes the most to decreased diversity (population bottlenecks), and the increased spread of deleterious recessive genes (the founders effect). Again, it is selection (use of this stud to the exception of others), and not the types of matings he is involved in that alters gene frequencies. Breeders should select the best individuals from all lines, so as to not create new genetic bottlenecks.

Decisions to linebreed, inbreed or outbreed should be made based on the knowledge of an individual's traits and those of its ancestors. Inbreeding will quickly identify the good and bad recessive genes the parents share, based on their expression in the offspring. Unless you have prior knowledge of what the offspring of milder linebreedings on the common ancestors were like, you may be exposing your litters (and buyers) to extraordinary risk of genetic defects. In your matings, the inbreeding coefficient should only increase because you are specifically linebreeding (increasing the percentage of blood) to selected ancestors.

Don't set too many goals in each generation, or your selective pressure for each goal will necessarily become weaker. Genetically complex or dominant traits should be addressed early in a long-range breeding plan, as they may take several generations to fix. Traits with major dominant genes become fixed more slowly, as the heterozygous (Aa) individuals in a breed will not be readily differentiated from the homozygous-dominant (AA) individuals. Desirable recessive traits can be fixed in one generation because individuals that show such characteristics are homozygous for the recessive genes. Individuals that pass on desirable traits for numerous matings and generations should be preferentially selected for breeding stock. This prepotency is due to homozygosity of dominant (AA) and recessive (aa) genes. However, these individuals should not be overused, to avoid the popular sire syndrome.

Breeders should plan their matings based on selecting toward a breed standard, based on the ideal temperament, performance, and conformation, and should select against the significant breed related health issues. Using progeny and sib-based information to select for desirable traits, and against detrimental traits will allow greater control.

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Popular-Sire Syndrome: Keeping watch over health and quality issues in purebreds

By Jerold S Bell, DVM, Tufts Cummings School of Veterinary Medicine

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An important issue in dog breeding is the popular-sire syndrome. This occurs when a stud dog is used extensively for breeding, spreading his genes quickly throughout the gene pool. There are two problems caused by the popular-sire syndrome. One is that any detrimental genes which the sire carries will significantly increase in frequency – possibly establishing new breed-related genetic disorders. Second, as there are only a certain number of bitches bred each year, overuse of a popular sire excludes the use of other quality males, thus narrowing the diversity of the gene pool.

The popular-sire syndrome is not limited to breeds with small populations. Some of the most populous breeds have had problems with this syndrome. Compounding this, there are several instances where a popular sire is replaced with a son, and even later a grandson. This creates a genetic bottleneck in the breeding population, narrowing the variety of genes available.

Every breed has its prominent dogs in the genetic background of the breed. But most of these dogs become influential based on several significant offspring that spread different combinations of the dog's genes over several generations. The desirable and undesirable characteristics of the dog were passed on, expressed, evaluated by breeders, and

determined if they were worthy of continuing in future generations.

The Challenges

The problem with the popular-sire syndrome is that the dog's genes are spread widely and quickly - without evaluation of the long-term effects of his genetic contribution. By the time the dog's genetic attributes can be evaluated through offspring and grand-offspring, his genes have already been distributed widely, and his effect on the gene pool may not be easily changed.

In almost all instances, popular sires are show dogs. They obviously have phenotypic qualities that are desirable, and as everyone sees these winning dogs, they are considered desirable mates for breeding. What breeders and especially stud-dog owners must consider is the effect of their mating selection on the gene pool. At what point does the cumulative genetic contribution of a stud dog outweigh its positive attributes? A popular sire may only produce a small proportion of the total number of litters registered. However, if the litters are all out of top-quality, winning bitches, then his influence and the loss of influence of other quality males may have a significant narrowing effect on the gene pool.

In some European countries, dog-breeding legislation is being considered that limits the lifetime number of litters a dog can sire or produce. If, however, certain matings

produce only pet-quality dogs, but no quality breeding prospects, should the dog be restricted from siring a litter from a different line? The popular sire's effect on the gene pool is on the number of offspring that are used for breeding in the next generation, and how extensively they are being used. This cannot be legislated.

At what point does a stud-dog owner determine that their dog has been bred enough? It can be difficult to deny stud service when asked, but the genetic effect of a dog on the whole breed must be considered. If everyone is breeding to a certain stud dog, the intelligent decision may be to wait and see what is produced from these matings. If you still desire what the stud dog produces, it is possible that you can find an offspring who has those positive attributes, and also a genetic contribution from its dam that you may find desirable. If a popular stud dog deserves to make a significant genetic contribution to the breed, doing so through multiple offspring, and therefore getting a mixed compliment of his genes, is better than focusing on a single offspring.

Wait-and-See Approach

All breeding dogs should be health tested for the conditions seen in the breed. If your breed has enrolled in the AKC-Canine Health Foundation/Orthopedic Foundation for Animals CHIC program (www.caninehealthinfo.org), prospective breeding dogs and bitches should complete the recommended breed-specific health testing prior to breeding. These may include hip radiographs, CERF eye examinations, or specific genetic tests.

It is important to monitor the positive and negative characteristics being produced by popular sires. While it is satisfying to own a popular stud dog, a true measure of a breeder's dedication is how negative health information in the offspring is made available. All dogs carry some undesirable traits. Based on the variety of pedigree background of bitches who are usually brought to popular sires, there is a greater chance that some undesirable traits could be expressed in the offspring. It is up to the stud-dog owner to keep in touch with bitch owners, and check on the characteristics that are being produced.

Some breeders will argue that the strength of a breed is in its bitches, but the fact remains that the stud dogs potentially have the greatest cumulative influence on the gene pool. There will always be popular sires, and that is not necessarily bad for a breed. But a dog's influence on a breed should be gradual, and based on proven production and health testing. Maintaining surveillance of health and quality issues in breeding dogs and their offspring, and preserving the genetic diversity of the gene pool, should allow a sound future for purebred dogs.

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