



## Pedigree Analysis, and How Breeding Decisions Affect Genes

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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. First comes understanding dogs and cats as species, then as genetic individuals.

There is little similarity between a Chihuahua and a Saint Bernard, or between a Himalayan and a Sphynx. However, we must understand that while established breeds are separate entities among themselves, they all are genetically the same species. While a mating within a breed may be considered outbred, it still must be viewed as part of the whole genetic picture: a mating within an isolated, closely related, interbred population. Each breed was developed by close breeding and inbreeding among a small group of founding ancestors, either through a long period of genetic selection or by intensely inbreeding a smaller number of generations. This process established the breed's characteristics and made the individuals in

it breed true.

Pure-breeds have closed stud books. This means that the diversity of genes in the breed is fixed. Genes cannot be gained through breeding, only lost. In some cat breeds, cats who meet the phenotypic standard of the breed may be introduced into the gene pool. This, of course is an added source of genetic diversity for the breed.

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 mismating 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 phenotypically 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.

Geneticists' and breeders' definitions of inbreeding vary. A geneticist views inbreeding as a measurable number that goes up whenever there is a common ancestor between the sire's and dam's sides of the pedigree; a breeder considers inbreeding to be close inbreeding, such as father-to-daughter or brother-to-sister matings. A common ancestor, even in the eighth generation, will increase the measurable amount of inbreeding in the pedigree.

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.

To visualize some of these concepts, the pedigree of a Gordon Setter, Laurel Hill Braxfield Bilye will be used. The paternal grandsire, CH Loch Adair Foxfire, and the maternal grandam, CH Loch Adair Firefly WD, are full siblings, making this a first-cousin mating. The inbreeding coefficient for a first cousin mating is 6.25%, which is considered a mild level of inbreeding.

In Bilye's pedigree, an inbreeding coefficient based on four generations computes to 7.81%. This is not significantly different from the estimate based on the first-cousin mating alone. Inbreeding coefficients based on increasing numbers of generations are as follows: five generations, 13.34%; six generations, 18.19%; seven generations, 22.78%; eight generations, 24.01%; ten generations, 28.63%; and twelve generations, 30.81%. The inbreeding coefficient of 30.81 percent is more than what you would find in a parent-to-offspring mating (25%).

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. The background inbreeding has far more influence on the total inbreeding coefficient than the first-cousin mating, which only appears to be its strongest influence.

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. This can occur in breeds, due either prolific ancestors (usually males), or a small population of animals originating the breed. Based on a twenty-five generation pedigree of Bilye,

there are only 852 unique ancestors who appear a total of over twenty-million times.

In Bilye's pedigree, CH Afternod Drambuie has the highest genetic contribution of all of the linebred ancestors. He appears 33 times between the sixth and eighth generations. One appearance in the sixth generation contributes 1.56% of the genes to the pedigree. His total contribution is 33.2% of Bilye's genes, second only to the parents. Therefore, in this pedigree, the most influential ancestor doesn't even appear in a five-generation pedigree. His dam, CH Afternod Sue, appears 61 times between the seventh and tenth generations, and contributes more genes to the pedigree than a grandparent.

Foundation dogs that formed the Gordon Setter breed also play a great role in the genetic makeup of today's dogs. Heather Grouse appears over one million times between the sixteenth and twenty-fifth generations, and almost doubles those appearances beyond the twenty-fifth generation. He contributes over ten percent of the genes to Bilye's pedigree. This example shows that the depth of the pedigree is very important in estimating the genetic makeup of an individual. Any detrimental recessive genes carried by Heather Grouse or other founding dogs, would be expected to be widespread in the breed.

The average inbreeding coefficient of a breed is a measurement of the breed's 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).

Looking at the historical pedigrees of Bull Terrier breeding dogs (males and females that have five or more registered offspring), we find that for dogs born in the decade 1970-1979, the average ten generation inbreeding coefficient was 23.11% +/-6.04%. For Bull Terriers born 1980-1989, this number is 21.54% +/-5.69%. For 1990-1999, the average inbreeding coefficient is 19.01% +/-6.23. It is obvious that the 10 generation inbreeding coefficient of the Bull Terrier breed is going down with each decade. This shows that the breeders are utilizing the diversity of the gene

pool, and not breeding themselves into a corner with popular sires.

Of course, the actual diversity of genes and inbreeding in the breed is not going down. It is just that the earlier ancestors producing background inbreeding are falling beyond the 10th generation, and are no longer included in the computation. As long as the health and vitality of the breed is being maintained, and there is no epidemic of breed-related disease from detrimental recessives, this pure-bred population should be able to be maintained.

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, a natural landrace 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 fallacy of using outbreeding to maintain genetic diversity is the belief that the diversity of a breed must be maintained in every single animal.

Breeders must concentrate on the specific goals of breeding (selecting for the health and quality of the breed), versus the tools used to get there (outbreeding, linebreeding, etc.) Selecting breeding stock simply to produce the lowest possible inbreeding coefficient is not a goal that will guarantee a quality animal. Animals who are poor examples of the breed should not be used simply to maintain diversity. Related individuals with desirable qualities will maintain diversity, and improve the breed.

The process of maintaining healthy "lines" or families of animals, with many breeders crossing between lines (outbreeding) and breeding back (linebreeding) as they see fit maintains diversity in the gene pool. It is the varied opinion of breeders as to what constitutes the ideal representative of the breed, and their selection of breeding stock that maintains breed diversity.

A basic tenet of population genetics is that gene frequencies do not change from the parental generation to the offspring. 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.