

### Australian Shepherd Health & Genetics Institute, Inc.

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# The Backyard Geneticist

by C.A. Sharp

A collection of articles on the applied genetics of dog breeding

Breeders are backyard geneticists and their legacy is the future health and quality of the *breed* rather than individual dogs or a single line. It is the knowledge and integrity of breeders that determines the health and future of a breed.

- C.A. Sharp



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### **Table of Contents**

Breeding & Genetic Diversity The Price of Popularity Why Incest Isn't Best	4
Is Outcrossing Dangerous? Heresy Revisited: Is crossbreeding justified?	10 13
Breeding Strategies  Playing COI The Impossible Dream What Price Glory? Genetic Juggling The Decision Tree Not WHO You Know But WHAT You Know Side Effects	19 22 24 26 29 33 37
Setting Priorities  Bad Genes, Babies, and Bathwater Flavor of the Month	41 43
Making Genetics Work for You In the Mode Of Babies, Bathwater, and DNA Tests Pedigrees: A Breeder's Roadmap	46 50 53
Science & Your Dogs  Alphabet Soup: Why genes have weird names Genetic Counseling When Worlds Collide DNA Fatigue What Are They Looking For? Not Always Clean and Simple	58 60 63 69 71 74
Controlling Genetic Disease From the Top Down The Importance of Community Trust but Verify Testing Ground	78 80 84 87

Many more articles on specific canine health issues and how to manage them in a breeding program can be found in the articles and FAQs on the ASHGI website: <a href="www.ashgi.org">www.ashgi.org</a>. Click on "Genetics Info." For reprint permission on any of these articles, contact the author.

### **Breeding and Genetic Diversity**

# The Price of Popularity Popular Sires and Population Genetics

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Consider the hypothetical case of Old Blue, Malthound extraordinaire. Blue was perfect: Sound, healthy and smart. On week days he retrieved malt balls from dawn to dusk. On weekends he sparkled in malt field and obedience trials as well as conformation shows, where he baited to--you guessed it--malt balls.

Everybody had a good reason to breed to Blue, so everybody did. His descendants trotted in his paw-prints on down through their generations. Blue died full of years and full of honor. But what people didn't know was that Old Blue, good as he was, carried a few bad genes. They didn't affect him, nor the vast majority of his immediate descendants. To complicate the matter further, some of those bad genes were linked to genes for important Malthound traits.

A few Malthounds with problems started showing up. They seemed isolated, so everyone assumed it was "just one of those things." A few declared them "no big deal." Those individuals usually had affected dogs. All in all, folks carried on as usual.

Time passed. More problem dogs turned up. People made a point not to mention the problems to others because everyone knows the stud owner always blames the bitch for the bad tings and takes credit for the good. Stud owners knew it best to keep quiet so as not to borrow trouble. Overall, nobody did anything to get to the bottom of the problems, because if they were really significant, everybody would be talking about it, right?

Years passed. Old Blue had long since moldered in his grave. By now, everyone was having problems, from big ones like cataracts, epilepsy or thyroid disease to less specific things like poor-keepers, lack of mothering ability and

short life-span. "Where can I go to get away from this?" breeders wondered. The answer was nowhere.

People became angry. "The responsible parties should be punished!" Breeders who felt their programs might be implicated stonewalled. Some quietly decided to shoot, shovel and shut-up. A few brave souls stood up and admitted their dogs had a problem and were hounded out of the breed.

The war raged on, with owners, breeders and rescue workers flinging accusations at each other. Meanwhile everybody carried on as always. After another decade or two the entire Malthound breed collapsed under the weight of its accumulated genetic debris and went extinct.

drastic little fable This is an exaggeration--but not much of one. Here's similar, though less drastic, example from real life: There once was a Quarter Horse stallion named Impressive. The name fit. He sired many foals who also exhibited his desired traits. But when they and their descendants were bred to each other, those offspring sometimes died. Impressive had been the carrier of a lethal single-gene trait. No one knew it was there until they started in-breeding on him. The situation of a single sire having this kind of drastic genetic effect on a breed became known as the "Impressive Syndrome."

Many species and breeds of domestic animals, including dogs, have suffered "Impressive Syndromes" of their own. But cases like that of Impressive are only the tip of the iceberg. A single-gene trait becomes obvious in just a few generations. But what about more complex traits?

This is not to say that those popular sires we so admire are bad breeding prospects. Their many excellent traits should be utilized, but even the best of them has genes for negative traits.

The problem is not the popular sires, but how we use them. For a century or more, in-breeding has been the name of the game. (For the purposes of this article, "in-breeding" refers to the breeding of dogs related to each other and therefore includes line-breeding.) By breeding related individuals, а breeder increased his odds of producing dogs for the traits he wanted. homozygous Homozygous individuals are much more likely to produce those traits in the next generation.

When a male exhibits a number of positive traits and then proves his ability to produce those traits he may become a popular sire, one that is used by almost everyone breeding during his lifetime, and maybe beyond, thanks to frozen semen.

Since the offspring and grand-offspring and so on are good, breeders start breeding them to each other. If the results continue to be good, additional back-crosses may be made for generations. Sometimes a sire will be so heavily used that, decades hence, breeders may not even be aware of how closely bred their animals are because the dog no longer appears on their pedigrees.

This is the case in Australian Shepherds. Most show-line Aussies trace back, repeatedly, to one or both of two full brothers: Wildhagen's Dutchman of Flintridge and Fieldmaster of Flintridge. These, products of a program of inbreeding, were quality individuals and top-producing sires. They are largely responsible for the over-all quality and uniformity we see in the breed ring today--a uniformity that did not exist before their birth nearly three decades ago.

Working lines have also seen prominent sires, but performance traits are far more complex, genetically and because of the significant impact of environment. They are therefore harder to fix. Performance breeders will in-breed, but are more likely to stress behavioral traits and general soundness than pedigree and conformational minutiae. The best working sires rarely become as ubiquitous as the best show-line sires.

Not every popular sire becomes so because of his ability to produce quality offspring. Some have won major events or are owned by individuals with a knack for promotion.

Such dogs may prove to be wash-outs once their get is old enough to evaluate. But if a lot of breeders have been using the animal for the few years it takes to figure that out, the damage may already have been done.

Use of even the best popular sires, by its very nature, limits the frequency of some genes in the breed gene pool while simultaneously increasing the frequency of others. Since sons and grandsons of popular sires tend to become popular sires the trend continues, resulting in further decrease and even extinction of some genes while others become homozygous throughout the breed. Some of these traits will be positive, but not all of them.

The owners of Old Blue, the Malthound in the opening fable, and those who owned his most immediate descendants had no idea what was happening under their noses. They were delighted to have superior studs and even more delighted to breed them to as many good bitches as possible.

Dog breeding and promoting is an expensive proposition. One usually winds up in the hole. But owning a popular sire can change that. The situation looks like a winner for everyone--the stud owner finds his financial burden reduced while breeders far and wide get to partake of his dog's golden genes.

No one breeding dogs wants to produce sick dogs. A small minority are callous and short-sighted enough to shrug genetic problems off as the price you pay to get winners, but even they do their best to avoid letting it come to general attention.

We need a total re-thinking of how we utilize stud animals. No single dog, no matter how superior, should dominate the gene pool of its breed. Owners of such sires should give serious consideration to limiting how often that dog is used, annually, through its lifetime and on into the future, if frozen semen is stored. The stud owner should also look not only at the quality of the bitches being presented, but their pedigrees. How much will the level of inbreeding be increased by a particular mating?

The bitch owner also needs to think twice about popular sires. If you breed to the stud of the moment and everyone else is doing the same, where will you go when it comes time to make an outcross?

Finally, the attitude toward genetic disease itself has to change. It must cease being everyone's dirty little secret. It must cease being a brick with which we bludgeon those with

the honesty to admit it happened to them. It must become a topic of open, reasoned discussion so owner of stud and bitch alike can make informed breeding decisions. Unless breeders and owners re-think their long-term goals and how they react to hereditary problems, the situation will only get worse.

## Why Incest Isn't Best

First published in Australian Shepherd Journal, Nov/Dec 2002

...the breeding of purebred dogs is akin to [breeding laboratory mice]...[most breeds] are becoming progressively more inbred. My observation is that most are on the road to extinction, but most breeders do not even realize they are part of an experiment.

John B. Armstrong, PhD

Incest, the marriage of close relatives, is forbidden on moral and legal grounds in most human societies. But the prohibitions also make biological sense. Inbred individuals tend to suffer from inbreeding depression. Among humans, the ancient Egyptian pharaohs and the more recent Hawaiian and European royal families provide the better known human examples. Inbreeding depression has not been studied much in dogs but it is well researched in other species.

Many dog breeders know very little about inbreeding depression. Purebred dogs, including the Australian Shepherd, are commonly bred to related individuals and occasionally bred to relatives close enough that among humans such a match would be considered incest. These are dogs, not people, so the moral issues are not the same. However, we ignore the biological impacts of the practice at our peril—or more accurately the peril for our dogs.

Inbreeding depression is the loss of viability or function resulting from excess inbreeding. The signs of inbreeding depression most frequently cited are reproductive failures. Anything that impacts reproduction is an up-front concern for dog breeders. But inbreeding depression may

also manifest itself as poor health. Its effects are often so subtle that breeders or even veterinarians may not recognize the root cause and not every inbred animal will show signs of it.

Inbreeding-related reproductive failures include lack of libido and low sperm count in males while females may fail to get pregnant, have unusually small litters, or exhibit poor mothering ability. Non-reproductive indications of inbreeding depression can be manifested as a high frequency of immune-mediated diseases, significantly higher incidence within a line or breed of one or more diseases than is seen in the species as a whole, or even things as subtle as dogs that is are "poor-keepers" or that seem to catch every little bug that comes along. In really severe cases, a very inbreed strain may go extinct.

The level of inbreeding is usually measured using a formula called Wright's Coefficient of Inbreeding. It calculates the probability that genes may have been inherited from both sides of an individual's pedigree. It is far too complex to do by hand over more than two or three generations, but some of the better pedigree software will calculate coefficient of inbreeding (COI) for you. The usual 3-5 generation pedigree won't give sufficient

information for a useful calculation. For Australian Shepherds the author has found 10 generations to be the best indicator. Few Aussies have a complete 10-generation pedigree, so this will give you pretty much all there is to know. While some individual lines of descent may go back 20-30 generations, running the calculation for more than ten does not result in any significant change in the result. Running it for fewer than 10 frequently causes the number to drop, giving an incorrectly optimistic result.

Modern breeders should know the COI of each of their dogs and determine what the COI will be on planned litters. The average COI is for the breed, or at least the portion of it with which you work, is also important. Show line Aussies average around 12-14%. This is roughly equivalent to all of them being half-brothers and sisters. Working line Aussies are variable. Many have very low COIs, under 6%, but some are as high or higher than the show line average. Inbred dogs, especially those with a COI of 25% or higher, that are experiencing problems of the kind described above may be exhibiting inbreeding depression.

Not surprisingly, most the research on the effects of inbreeding in mammals has been done on farm animals, laboratory animals like mice, and endangered wild species. One study on cattle done by Dr. John Pollock at Cornell University indicated increasing levels of health and productivity problems as the COI rose beyond 9%.

Laboratory mice are often pointed to as proof that extreme inbreeding works. Lab mice are arguably the most inbred of domestic mammals; so much so that members of a strain are near clones of one another. This extreme inbreeding is necessary so researchers will know exactly what to expect from that particular strain.

Strains are developed by breeding mice brother to sister for many generations, producing levels of inbreeding unheard of in dog circles. But there is a price to be paid for this. In 20 generations, 80% of the lines descendant from the original pair will have

gone extinct due to lethal health problems or an inability to reproduce. Even those that make it through the bottleneck are hardly the mice their ancestors were. Take any pair of field mice put them in a laboratory cage and they survive very well, living longer than they would in the wild. Take any pair of lab mice and put them in a field and their "wild" life expectancy is zip. They are suitable only to the very controlled environment of a laboratory, where the "weather" never changes, food, water and housing are provided and there are no predators.

Wild species of all kinds employ a number of behavioral strategies to avoid inbreeding. If Nature does something so consistently, you can bet she has good reason. Among social animals, the young of one or both genders may disperse to form or join other groups. Dominant breeding males may hold their position only a short time. Solitary animals tend to be territorial, at least in breeding season, with a male's territory overlapping that of several females. Their offspring must disperse and seek territory elsewhere, sometimes traveling long distances to do so. But even in nature, conditions are ocasionally such that an animal has no choice but to mate with a relative.

The cheetah is a very inbred species. In the Pleistocene, cheetahs roamed North America and Asia as well as Africa. Genetic studies have revealed that all present-day cheetahs are near identical genetically. Ten thousand years ago, some kind of catastrophe reduced the entire world cheetah population to a very few individuals in Africa. It is possible only a single pregnant female survived. Fortunately for the cheetah, it passed through this terrible genetic bottleneck. Under the harsh selection of a natural system that tolerates no weakness and with a fortunate collection of genes in the bottleneck survivors, the cheetah has managed to hang on. Even so it is very susceptible to some diseases and has reproductive difficulties.

The cheetah's inbreeding problem is "background." Due to that long-ago

bottleneck, they are all closely related even though there are now several thousand of them. A number of breeds of dog have experienced similar but more recent bottlenecks due to things like war. Other breeds have very few founders, which is akin to a wild species living on an island.

Islands that are well away from the nearest mainland gain species only rarely, when a very few individuals arrive through some accidental circumstance. If they survive the initial inbreeding depression they adapt to their new environment, sometimes to the point of forming entirely new species as can be seen with the finches and tortoises of the Galapagos Islands. But because island species have such a narrow genetic foundation they are highly susceptible to anything that changes their environment. Native Hawaiian species have been severely impacted and many driven extinct by their inability to adapt to the presence of species that accompanied early Polynesian migrants as well as more recent introductions by American, European and Asian settlers.

Wolves are more relevant to dog breeders than tortoises or finches and they provide an excellent example of this process. For the most part, wolf populations are scattered across great landmasses of the northern hemisphere. Normally, only the alpha pair of a pack will breed and most offspring disperse during their first or second year to seek new territory. Those that remain are strongly discouraged from breeding by higherranking pack members. But there is a population of wolves on Isle Royale in Lake Superior that descends from a few animals that crossed 15-20 miles from Ontario when the lake was frozen over sometime around 1950. Except for one released captive in the early 50s, no other wolves could have contributed to the current population.

There are moose on the island, so for many years the fortunes of the increasingly inbred wolves followed the boom and bust of the moose population cycle. The moose and wolves of Isle Royale have been the subject of the

longest-running predator/prey study ever conducted (now 44 years and counting.) The wolf population sometimes exceeded 50 animals, but in the late 70s things began to go wrong. The wolves suffered waves of parvo, distemper and mange. Ultimately the population sank to a handful of animals and there was fear they might go extinct. Scientists debated whether new wolves should be introduced or nature left to take its course so they could observe how the moose fared without a predator to cull the weak and unfit.

The ultimate decision was to let things be and the wolves managed to pull through, with 19 counted in 2001. The population appears healthy, but the next canine plague may again leave their fate hanging in the balance.

Our dogs don't have to hunt for their dinners or survive in the elements. They are provided with comfortable environments, sometimes to the extent of air-conditioning and a space on the couch. Obtaining a meal requires no more effort than a trot to the food bowl. It is neither moral nor ethical for a dog breeder to subject his animals to the kind of harsh culling process exacted by Nature. However, we should give health, fitness and reproductive issues much greater importance in our selection process.

Australian Shepherds are blessed with around 300 founders, so our background inbreeding is very low.
However, our breed like most others has experienced historic inbreeding stemming from the choices made by recent generations of breeders who have used one sire more than others or frequently sought the output of a particular kennel. Early preference for the Flintridge-type dog in the show ring is a prime example in Aussies. Even in working lines there are early sires and kennels whose names occur with considerable frequency.

Any time a single dog or kennel is responsible for producing a significant portion of the breeding animals in a breed, the breed gene pool is skewed toward whatever genes those dogs had. Breeders

do this because they want to skew things toward genes that produce the desirable traits they found in those animals. But along with those genes come others that are less desirable.

Inbreeding depression isn't a matter of specific genes that cause the sort of health problems we are always on the lookout for, like bad hips or eve disease. It lies in genes that have a subtler effect. Such genes work only a little less well than their "good" versions. A breeder isn't likely to notice a 5-10% reduction in bodily function. But as levels of inbreeding rise these genes may accumulate. Over time reproductive and health problems increase. The changes are so gradual they are often blamed on diet, pollution and other environmental causes. All of these may contribute but it is genes that make dogs susceptible.

Every individual has 3-5 "lethal equivalents." These are an accumulation of mutated genes that, if matched with like genes, would either kill the organism early in life or prevent it from reproducing. The portion of lethality carried by any one gene can range from 1-100%. A single lethal equivalent may be one gene, 10 that reduce fitness by 10%, or a hundred that reduce it 1%. The mutt that lives down at the corner probably has in excess of 100 such genes. Your purebred Aussie likely has more because they have been inadvertently concentrated by inbreeding.

Each puppy will inherit half of its lethal load from each parent. If the parents are unrelated, there may be little or no impact on that puppy. But if they are, the risk of pairing up genes that are part of the load increases. The more ancestors there are common to both sides of the pedigree, the greater the risk. This is why monitoring COI is important.

Historic inbreeding and recent or "close" inbreeding (matings that would be considered incestuous among humans) are the primary factors contributing to inbreeding depression in purebred dogs, with the historic inbreeding being the

greater factor since many don't recognize it as inbreeding at all.

There is a strong possibility that inbreeding depression may be becoming a factor in Australian Shepherds.
Reproductive problems do occur. Almost 25% of the breeders who responded to the 1999 Australian Shepherd Club of America breed health survey reported having dogs with reproductive failures (low sperm count, lack of libido, failure to conceive, small litters or poor mothering ability). This has sobering implications for the breed. There is no way from this data to know the COIs of the affected dogs, but such a large response indicates that breeders need to pay more attention to reproductive issues.

Autoimmune disease data was gathered in the same survey. (Allergy data is being excluded here because the survey did not distinguish between mild or isolated allergy attacks and severe chronic conditions.) 17% of the respondents reported having had at least one dog with autoimmune disease, another sobering statistic.

Understanding the problem is one thing, but what can a breeder do about it? Very few practice close inbreeding generation after generation. And breeders aren't in the business of breeding COIs, they want to produce quality dogs. This can be accomplished by giving preference to assortative mating—the breeding of individuals of the desired phenotype with preference given to those least related.

Say your bitch as a COI of 12%. You have looked at possible studs and narrowed your list to three that you think are equally good. Dog A is from the same line as your bitch; the puppies would have a COI of 21%. Dog B, an older stud who hasn't been bred often but has produced consistently good offspring, would produce puppies that are 10%. Dog C, an up-and-coming star already booked for a number of bitches, would produce puppies that are 8%. Dog C might seem like the way to go at first glance, but he has high potential for becoming a popular sire. Your puppies might wind up half-siblings to everybody

else's. Dog A causes a significant increase in inbreeding. Dog B may not give the lowest COI, but he does produce puppies that will be lower than their dam and you are more likely to find less-related mates for them because he has not been over-used.

We also need to do what we can to reduce the lethal load carried by our dogs. We should not use animals that exhibit signs of inbreeding depression. Consistent reproductive failures are an excellent argument against further breeding. There is nothing more natural than reproduction. The dog that is unable to produce, deliver or rear puppies without veterinary intervention or considerable help from the breeder should not be bred. Chronically unhealthy or unthrifty dogs are not good breeding candidates, even if they have no identifiable hereditary disease. Nor are dogs with severe allergies or any chronic autoimmune

disease. Some of these diseases can affect reproduction, thyroid disease being a prime example. In the author's opinion, the administration of thyroid hormone to bitches that will not settle so they can produce puppies is foolishly short-sighted at best and highly unethical if done by someone knowledgeable.

Even with the rapid rise of inbreeding levels in the Australian Shepherd to the point that so many of our dogs are virtual half-siblings, our short history as a registered pure breed and our broad founder base has given us the potential to keep our breed healthy and viable. The Aussie is in much better shape than so many other breeds and need not risk the extinction warnings of the opening quotation. It is up to breeders to see that it stays that way.

## Is Outcrossing Dangerous?

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Breeders' lore has long maintained that outcrossing ought to be done only occasionally if at all because doing so dilutes the qualities of a line courts disaster. There are numerous tales about breedings that went wrong because of an outcross. But is this really the case?

#### What is an outcross?

Before deciding whether outcrossing is or is not a good idea, we need to have a good understanding of what it is. Generally speaking, an outcross is the mating of two largely unrelated animals within the same breed. Virtually every dog in a breed will be at least slightly related to all others if you search far enough back in the pedigrees.

When people review printed pedigrees of prospective mates and see few if any common names behind both dog and bitch, the resulting litter is assumed to be an outcross. However, if one does not have a thorough knowledge of the breed's pedigree history, an apparent outcross may be nothing of the sort.

In populous breeds like the Australian Shepherd it is very possible that two dogs will share no common ancestry on a three-to five-generation pedigree but will actually be closely related. The breed average 10-generation coefficient of inbreeding (COI) for Australian Shepherds is somewhere around 14%, or slightly higher than that of half-siblings who are otherwise unrelated. The COI is calculated over ten generations because fewer will not represent an accurate measure of background inbreeding. Most Aussies are already closely related, though it is possible to find some with substantially different pedigrees.

The best way to gauge whether a cross is or is not an outcross is by calculating the COIs of the parents and the proposed litter. If the result for the cross is low (under 6.25%) or at least substantially lower than that of the lowest parental COI, it can be termed an outcross. Here are some examples:

#### **Do the Numbers**

For those who enjoy delving into the complexities of mathematics, here is some information on formulae that pertain to the discussion of linebreeding vs. outcrossing and their impact on the likelihood of producing undesirable traits.

An excellent explanation of Wright's Coefficient of Inbreeding (COI) can be found in Willis, Genetics of the Dog (Howell 1989) p. 320-5. Discussion of the Hardy-Weinberg equilibrium formula can be found in Ackerman, The Genetic Connection (AAHA 1999) p. 18.

Under the Hardy-Weinberg law, the possible genotypes for a gene with alleles A and a, where p is the frequency of A and q is the frequency of a in the population:

AA pxp Aa 2xpxq aa qxq

If you factor in COI, represented here by F, you get:

AA pxpx(1 - F) + pxF = pxp + pxqxFAa 2xpxqx(1 - F) = 2xpxq - 2xpxqxFaa qxqx(1 - F) + qxF = qxq + pxqxF

As the COI increases, the proportion of heterozygotes (Aa) decrease while the homozygotes (AA and aa) increased. If aa is the genotype for an unwanted trait, the number of dogs exhibiting that trait will have increased because of inbreeding. The higher the COI the more frequently you will see the unwanted trait.

The author would like to thank James Seltzer, PhD, mathematician and Dalmatian breeder, for his assistance with this sidebar.

Litter A: The dog has a COI of 16.4%; the bitch's COI is 11.3%. Their pedigrees have nothing in common for

at least five generations. The COI for their litter is 13.0%, therefore they actually have a great deal in common beyond the printed pedigree. Litter A is not an outcross.

- Litter B: Both dog and bitch share a common great grandsire. The dog's COI is 9% and the bitch's is 6.3%. Except for that one great grandsire, their extended pedigrees share almost
- nothing. The COI of the cross is 1.8%, of which half comes from that common great grandsire. Litter B is an outcross.
- ➤ Litter C: Both dog and bitch are tightly linebred though from different bloodlines, with COIs of 25% and 37% respectively. The cross between them would have a COI of only 2.3%. Though they are both rather inbred they have almost no common pedigree. Litter C is an outcross.
- Litter D: The dog's COI is 1.3% and bitch's is 6.9%. Their litter is 13.2%. The dog's sire and the bitch's dam were from the same line. Litter D is not an outcross.

Now that we have established what an outcross is (and is not) we need to examine the question of how useful it is as a breeding tool.

#### But does it work?

There are volumes of breeder lore supporting the assumption that outcrossing is not a viable breeding strategy if done over multiple generations, though it might be done occasionally within a linebreeding program but not without some risk. That assumption is not supported by genetic science or mathematical analysis, [see sidebar] so why is this belief so pervasive?

In large part it stems from the assumption that linebreeding is superior because it allows the breeder to concentrate desirable genes while at the same time eliminating those that are undesirable. Linebreeding is very effective for fixing trait that are readily observed or measured. A trait is genetically "fixed" when it is consistent throughout a population, like the color black in

Schipperkes. If linebreeding were not effective in this respect, we wouldn't have so many distinct breeds of dog.

Genetically complex traits can be difficult to fix, though diligent selection for them over many generations can significantly increase their frequency in a breed population. Herding ability is a genetically complex trait involving multiple genes plus environment. Even in bloodlines where selection for a high level of herding ability has been strong for generation after generation, the breeder still will produce some pups that don't measure up.

The success of linebreeding as a technique for consistently producing desirable traits has fed the myth that undesirable traits can be totally purged from a line or breed. While an undesirable trait—whether it is something cosmetic like color or a serious health concern like epilepsy—can be reduced in frequency through diligent selection against it, in most cases it will not be possible to eliminate the trait entirely unless it is the result of a single gene dominant. Most such traits were weeded out of modern breeds a long time ago.

If the trait is a single-gene recessive, careful and consistent selection against it will eventually reduce its frequency to low levels. The trait will not be produced often, but it will still crop up from time to time. Rare breeds may actually lose the gene through a process called genetic drift simply because their populations are so small that there is a fair chance the unwanted gene won't get passed along. In populous breeds, however, this is unlikely to happen. The key to reducing the frequency of unwanted genes as much as possible is consistent negative selection no matter whether linebreeding or outcrossing is employed.

As we saw with herding ability, desirable traits with complex inheritance are impossible to fix. Total elimination of such traits is equally impossible outside of very small breeding populations. Hip dysplasia (HD) is but one example. The key to reducing the frequency of something like HD is diligent and consistent selection.

Though traits like HD can't easily be eliminated, it may be possible to "clear" a line by elimination of some portion of genetic puzzle that produces them. A single line is often a small breeding population within the breed as a whole. Here is how it works: If BINGO represents the combination of genes necessary to produce HD, a combination of linebreeding and diligent negative selection may produce a

line lacking one or more elements. This line would never produce that trait so long as it is kept closed.

But he's such a nice dog...

A serious impediment to significantly reducing the frequency of unwanted traits is the set of priorities every breeder must establish for her breeding program: Which traits are vital, which can be tolerated, and which are unacceptable. It is impossible to achieve every breeding goal in a single litter, so the breeder will rearrange that priority list somewhat from one mating to the next. This can interfere with the effort to eliminate unwanted traits because unless most or all breeders are consistent in viewing the trait as intolerable, it will persist. Even if the gene frequency is significantly reduced over time, all it takes is one popular sire who is a carrier to make the trait common once again as people linebreed on him and his near kin. Too often one hears the refrain, "but he's such a nice dog!" even though that dog has or produces a serious defect or disease.

The complexities of breeder priorities are not the only reason unwanted genes persist. Breeders may not know those genes are there until the trait crops up in a litter. The days of huge kennels with meticulous records on generation after generation of dogs are long gone. Most breeders have only a very few dogs and regularly need to make use of outside studs or purchase new stock from another breeder. Unless everyone keeps all their cards on the table, linebreeding cannot be fully effective in clearing genes for unwanted traits.

If the chain of information is broken anywhere in the breeder network, someone will make a disastrous mating choice no matter whether they linebreed or outcross. When it happens in an outcross, people tend to ssume the outcross was the cause. This attitude prevails because provides an easy answer hat allows the owners of both stud and brood bitch to assume it must have come from the other side. In truth, both need to acknowledge the fact that the genes are there and it could happen again.

There is actually a greater risk of producing unwanted traits through linebreeding than outcrossing. If a trait has occurred, the genes are present in the line. Continued linebreeding will inevitably bring them together again. COIs can be useful here. The COI is a measure of how likely it is that both copies of a gene will be identical by decent from each side

of the pedigree. The higher the COI, the more likely you are to double up on genes both good and bad.

The COI is a mathematical demonstration of why people linebreed. The tighter the linebreeding, the more likely desirabale traits will be produced. Unfortunately, the same is true for unwanted traits. There is not a dog in the world that does not have a few undesirable genes

#### Assortative mating

Too often people equate outcrossing with mating two dogs that are as different as can be. Certainly if there is a huge disparity between them, the breeder is unlikely to produce animals of acceptable quality. In Aussies, this might mean taking something from the show lines, all of which are heavily based on the Flintridge line of the 1960s and early 70s, to a working dog whose background is derives largely from the old Woods line. Such dogs would be different in structure, coat, color, and behavior. Their offspring would be unlikely to please the owners of either parent because of their significant differences will produce a littler with little uniformity. However, this is not because the litter is an outcross, but because the particular individuals used were so dissimilar and therefore unsuited to each other.

For outcrossing to work effectively, assortative mating should be employed. It

allows the breeder to maintain the desirable qualities and the same time reduce the risk of producing unwanted outcomes.

Assortative mating is the selection of pairs based on phenotype—what you see or know about the dog and its family—rather than the pedigree. To be successful in any breeding endeavor, the breeder must have a clear idea of what traits he wants and which he does not, as well as how much potential his bitch has for carrying those genes. Studs should be evaluated based on what they have produced and the phenotypes of the members of their extended families, including full- and halfsiblings plus parents and grandparents and their full and half siblings. If that family of dogs is consistent for the desired traits and lacks those the breeder wishes to avoid, the mating has a strong probability of success no matter what the pedigrees involved.

Risk for unwanted traits, like health issues, are maximally reduced by giving preference to suitable studs are the least related among the group you are considering.

Is outcrossing dangerous? Not if done with suitable animals in conjunction with knowledge of both desired and unwanted traits in the family background of both sides of the equation.

# Heresy Revisited Is crossbreeding ever justified?

First Published in Double Helix Network News, Fall 2011

A dozen years ago I wrote an article called "Speaking Heresy." It was about crossbreeding, the intentional mixing of different breeds of dog. Since then the designer dog fad and it's plethora of whatever-doodles has, if anything, made purebred dog enthusiasts even more critical of the practice. In spite of this, the Dalmatian Club of America recently voted to allow the "Backcross Dals," descendants of a long-ago crossbreeding with a Pointer, into their AKC studbook. In view of that and other developments I felt it was time for another look at the practice of crossbreeding, our almost

universal knee-jerk condemnation of the practice, and whether it might have a place in purebred breeding.

#### Anathema

For over a century cross-breeding has been looked on with scorn as an adulteration of pure canine blood that took generations of dogs and humans alike to develop. "Designer" dogs are disdained for their mixed ancestry, as are those who seek to create new breeds by combining the genes of established breeds. Within recognized pure breeds it is, with

extremely rare exceptions, forbidden. Whatever the reasons for crossbreeding today – and the motivations can be highly varied as to purpose and ethical practice – practitioners are generally sanctioned, socially and sometimes formally, by purebred dog enthusiasts and organizations.

Despite the overwhelming pejorative view of the practice in dogs, crossbreeding is a legitimate and useful breeding technique. Early in the history of pure breeds it was commonly practiced for a variety of reasons. Understanding this history, why those crosses were done, and how the practice can be effectively employed today is something every breeder should be aware of even though most will never employ it.

#### Early history

The modern concept of pure breeds is just that: Modern. Prior to the 19th century there were no breeds as we know them today. There were particular types of dog found in certain geographical regions, developed over generations to meet needs varying from hunting to farm work, or vermin control. Members of the aristocracy might selectively breed a unique strain but many types arose because dogs who would do a certain job were important to people's livelihood. None of these strains were purebred in the sense of studbooks and formal closed registries. Breeding records might be kept by the literate, but in those days literacy was the exception. Even so, though some nonliterate cultures, like the nomadic Bedouin, had a strong oral tradition and might commit detailed pedigrees of valued dogs to memory.

Unhindered by closed studbooks and registry restrictions, if a nobleman of times past thought a different sort of dog had something to offer his bloodline or a farmer felt it might throw better work dogs with his bitch, that other sort of dog would be used. If the offspring proved worthy, they would be kept and bred from with no one thinking any the less of them for their mixed ancestry.

Crossbreeding was a feature in the development of many of our modern breeds, with perhaps the most well known being the Doberman, created by tax Collector and pound-master Louis Dobermann, who selectively bred a variety of dogs to get the personal protection animal his tax duties required. However, the Doberman is far from the only breed that sprang from purposeful crossbreeding. The Bullterrier, originally called the Bull and Terrier was, as that early name suggests, derived from a cross

between bulldogs, terriers, and possibly other breeds. In 1859 the Victorian dog authority "Stonehenge," John Henry Walsh, condemned the early Bullterrier for its mixed heritage, but little more than a decade later Walsh included it as one of eight recognized terrier breeds.

In the 19<sup>th</sup> century a whippet was crossed with a Black and Tan Terrier (today called the Manchester) to produce a more elegant animal that appealed to city gentlemen who wanted a vermin dog but also wanted something stylish, forever changing the look of that breed. For similar reasons Borzoi blood was introduced to the Collie, making the one-time farm dog more appealing not only to the aristocracy but to the up-and-coming middle class.

The Irish Wolfhound is the product of a purposeful attempt to recreate an historic breed, few if any of which had survived to modern times. Dogs which had the look of the ancient breed, as depicted in artwork, were selected. These dogs had little or no known pedigree and no clear connection to the original breed. The related Scottish Deerhound was used to help fix the type. Borzoi and Great Dane were introduced to increase size.

Not all crossbreeding took place early in purebred history. The Red Setter, a field version of the Irish, was banned from the Irish studbook after breeders introduced English Setter to improve working traits. More recently breeders who desired a coated sighthound of moderate size crossed a Shetland Sheepdog to a whippet then back-crossed to whippets selecting for the long coat. The result was the Long-Haired Whippet, which has a modest and dedicated following but remains an object of scorn to Whippet enthusiasts and lacks AKC recognition.

#### The Rise of Registries

Dog registries were originally developed for record-keeping purposes but ultimately morphed into arbiters of canine purity. That aspect of their function is the child of the class-conscious European social thinking of the 19<sup>th</sup> century, when "pure blood" was tantamount to good breeding for humans, not only among the nobility but also for the rising merchant class and gentleman farmers. Marrying "beneath one's station" was thought to introduce "inferior blood" to a family and would necessarily result in offspring of poor character and limited ability. This thinking was transferred to the breeding of animals, including dogs.

Other species have pure breeds with formal studbook registries, too, but only in dogs is the use of crossbreeding almost uniformly condemned. Pure breeds of livestock are maintained, but for commercial production growers don't hesitate to crossbreed if the result is a more marketable carcass or improved production of food or fiber. Various horse registries will admit crossbreds under certain circumstances: Racing lines of Quarter Horse can have some Thoroughbred background and part-Arabians are allowed registration.

### Why crossbreed?

We tend to focus on negative applications of this very old breeding practice: The blatant commercialization of the "designer dogs" or the *sub rosa* use of one breed to improve ones chances of competing successfully, as with the use of Pointers with some field dog breeds, Border Collies with working line Australian Shepherds, or the alleged use of an Afghan Hound a couple decades ago to improve show coat in Irish Setters. Turning dogs into consumer fad items or cheating in competitive events are rightly condemned, but was the fault here crossbreeding or some combination of hubris and greed?

Crossbreeding has some well-established benefits, which explains its continued use in livestock. The first generation offspring of a cross between two pure strains exhibits hybrid vigor, an improvement of health and other biological qualities derived from increased heterozygosity of the genes in the offspring. Designer dog merchants often tout improved health conferred by hybrid vigor when marketing their product. However, hybrid vigor only occurs in the first generation. If you backcross those dogs to each other or either parent breed, you reduce the level of heterozygosity and no longer have the benefit of hybrid vigor.

Crossbreeding can be used to develop an improved strain suited to a specific purpose, to reintroduce a lost trait, or improve on existing ones in an established breed. We saw examples of this in the formation of our pure breeds as well as in livestock and horse breeding. There are legitimate examples of crossbreeding of dogs in recent history. Some guide dogs associations have crossbred Labrador and Golden Retrievers because they found the crossbred offspring were less prone to hip dysplasia and successfully completed their

training more frequently than purebreds of either breed. Before Labradoodles became fashion statements, their early breeders' goal was a well-tempered, readily trainable, low-maintenance pet for the average family.

Crossbreeding can be effective in introducing a trait that is lacking, correcting a problem, or expanding a tight gene pool. Its use for purposes like these should be considered legitimate, provided the crossbreeding program is open as to its practices and goals and, if focused on a particular breed, it works within the established breed organization and registry.

#### **Documented Crossbreeding Efforts**

The Backcross Project - Prior to this project, all Dalmatians had two copies of a mutation that causes high uric acid, which frequently leads to bladder stones. In the mid-1980s Robert Schible PhD, a medical geneticist, crossed a Dalmatian bitch to a Pointer in order to introduce the normal version of the gene causing high uric acid. For the subsequent four generations the progeny were bred to Dalmatians, resulting in dogs that looked like purebred Dals, though they did have a tendency to have patches in addition to spots, a significant color fault in the breed. This effort was done with the knowledge and support of the board of the Dalmatian Club of America (DCA). At the request of that board, the American Kennel Club agreed to register two of the 5<sup>th</sup> generation pups. However, club membership had been unaware of the project and when they found out they were outraged and overturned the sitting board. The new board requested that AKC no longer accept the backcross dogs and the registry complied. In spite of the set-back, a dedicated group of Dalmatian breeders maintained the Backcross bloodline which today is indistinguishable from other Dalmatians.

The genetic mutation that causes high uric acid was identified a couple years ago and there is now a test available. A proposal was made to the DCA membership that the backcross dogs, whose only non-Dalmatian heritage was that one Pointer a quarter century ago, be allowed back into the AKC registry. Initially, the membership voted the proposal down. Proponents launched an education campaign and earlier this year the DCA members voted, by a narrow majority, to allow the dogs in. These dogs and the DNA test will allow Dal breeders to gradually reduce the

frequency of the high uric acid mutation in the breed.

The Bob-tail Boxer Project – With the probability of a docking ban in the United Kingdom, Bruce Cattanach PhD, a medical geneticist and Boxer breeder, developed a plan to introduce the bob-tail gene into his long-tailed but traditionally docked breed. He bred one of his bitches to a bob-tail Pembroke Welsh Corgi then backcrossed to Boxers for four generations, selecting the most Boxer-like offspring to create each subsequent generation. Despite the significant morphological differences between the breeds, the 4<sup>th</sup> generation dogs looked like purebred Boxers.

Docking is now banned in the United Kingdom. The Kennel Club agreed to accept the dogs into its registry, but with the bob-tail gene present in the breed, Boxer breeders can produce short-tail dogs if they wish.

The Swedish Clumber Spaniel Project -The Clumber Spaniel has a restricted gene pool. Members of the Swedish Clumber Spaniel Club have been concerned about this for years and discussed crossbreeding as a way of widening their gene pool. Drs. Lennart Svennson and Per Erik Sundgren agreed to assist. In 2003 a Clumber bitch was bred to an English Cocker Spaniel of working type. The ten resulting puppies were examined by show judges and the two bitches of the most consistent Clumber type were admitted into the Swedish Kennel Club's registry, which includes an "X" in the registration numbers of project dogs so they can readily be identified. At this time those bitches have had three litters between them. Some of those dogs are competing successfully in conformation across Europe. Since most of the dogs are still relatively young the health benefits and working quality can't yet be assessed but it is hoped the new genetic material will result in improved health and longevity.

#### How-To Manual

If crossbreeding is to be employed it must be for a well-defined purpose and with a detailed plan of action. It must also be open, with everyone involved aware of the heritage of all dogs produced in the effort. If the project is intended to introduce something lacking to an established breed, the national club and the registry must approve the effort beforehand, be fully aware of the details, and club membership needs to be informed and educated about the

purpose and progress of the effort. Failure to do so can doom the project, as exemplified by the long, rocky history of the Dalmatian Backcross Project.

The DCA board of the time failed to inform membership about what was being done. It was presented as a *fait acompli* after AKC registered the two Backcross dogs. Not surprisingly, the membership voted the responsible board members out of office and the new board stopped the registration of any further Backcross dogs. It took a quarter century to undo the damage.

The Bob-Tail Boxer and Clumber projects have had much better initial success. Both were done with the knowledge and approval of all parties. Dr. Cattanach at the time was a member of the KC's health committee. The greater majority of the Swedish Clumber club's membership were the guiding force behind that effort and allowed the SKC close oversight of everything they did. However, the Boxer project received a setback when the German breed club (Germany is the breed's country of origin) petitioned the Federation Cynologique Internationale (FCI), which governs shows and kennel clubs throughout most of the world, to ban the showing of any Boxer with a "naturally stumpy tail." The bob-tail Boxers can still compete in the UK, Australia and the United States, whose kennel clubs are not FCI members.

#### Where to go?

The choice of the other breed for a breed improvement crossbreeding project requires careful consideration. The more similar in appearance and behavior the other breed is, the easier it will be to get consistent quality in the backcross generations. A Pointer was chosen for the Dalmatian project because the breeds are similar in body structure. In addition, Pointers are typically mostly white with patches and small ticking spots where properly colored Dals are entirely white with large round ticking of relatively uniform size and distribution. The field-bred English Cocker was used for the Clumber project because the breeds are related and have similar hunting styles and coat patterns.

The use of a Corgi for the Bobtail Boxer project may seem a stretch, but there is no bob-tail breed that is morphologically similar to a Boxer. Dr. Cattanach demonstrated that even with such a drastic difference one can produce quality within a very few generations. It

helped that two of the most un-Boxerlike Corgi traits, short legs and a longer coat, proved to be recessive and easy to breed out.

The choice of the other breed is also dependant on the purpose for the effort. For the Dalmatian and Boxer projects, the goal was to introduce the desirable version of one gene. An individual from any breed in which each dog has two copies of the desired gene version would pass a copy on to each of its crossbred offspring. However, using a breed physically and/or behaviorally similar – if available – would make the job easier.

If the problem to be addressed involves many genes – as with augmenting a tight gene pool – the problem becomes far more complex. The cross could influence many important traits. not all of them easily observed or measured. Insofar as this improves the health, viability, and longevity of the breed, this is all to the good. But behavioral and physical traits that don't conform to the breed standard will need to be selected against and more generations may be needed to fix breed type. At this point there is no easy way to track all of the newly introduced gene versions from one generation to the next, so we can't know how many are retained as we backcross to the original breed. We will lose those that are linked (close on the same chromosome) to genes that we don't want those that contribute to incorrect appearance or behavior - because of strong selection against that neighboring gene.

The Clumber project is still in the early stages so traits more typical of the English Cocker still occur. In time that will be sorted out. but in the process some Cocker genes not causative for the Cocker traits will go with them in the effort to get proper Clumber type back. Thus far the Clumber project has used only a single outcross and the long-term benefits remain to be seen. With projects of this sort it may prove necessary to repeat the process more than once to maximize the inflow of beneficial gene variants. The different outcrossed lines would need to be kept distinct from each other not only until acceptable breed type was established but for a few generations thereafter to make sure the type remained consistent. During this time, the strains could be crossed with any suitable pure line. The point would be to avoid having more than one dog of the other breed in the recent pedigree.

MHC profiling

An approach toward improving health in breeds with extremely tight gene pools that no one, to my knowledge, has tried yet would be to focus on introducing increased diversity at the Major Histocompatibility Complex (MHC) a group of genes that govern the functioning of the immune system. Those genes are all grouped on canine chromosome 12 and tend to be inherited together in sets called a haplotypes. MHC genes are highly variable and many different haplotypes exist in the dog as a species. However, individual breeds each have only a subset of the total.

Popular sire breeding can reduce the number of haplotypes in a breed by unintentionally selecting against those which the popular sires do not possess. A dog can have, at most, two haplotypes – assuming the one inherited from his sire was different that the one he got from his dam. Breeds with small founder bases or small populations also tend to have fewer haplotypes. This can impact immunity to infections and the frequency of immune mediated diseases.

At this point researchers have identified MHC haplotypes for around 80 breeds, though the findings thus far may not be complete. Also, there is no commercial diagnostic test available to tell you what MHC haplotypes a particular dog has, so launching a MHC diversity crossbreeding project at this point isn't practical. In time it may be.

If MHC genotyping becomes available and the haplotypes present in a breed are known, a breed with different haplotypes could be identified and individual dogs tested to verify that they carry two "new" haplotypes before they are used for a crossbreeding. Such dogs could be bred to bitches of the first breed and the progeny backcrossed to the original breed then selected for breed traits and the novel haplotypes. In time one could introduce greater MHC diversity and better health to the original breed. More than one outcross should be done so multiple new haplotypes could be introduced.

#### Measuring Success

Crossbreeding is not something to be approached lightly. For it to be successful it must be well planned and executed with the support of all stakeholders. Provisions must be made to reintroduce descendants of the outcross to full registration as members of the breed. England's KC will do so after four backcross generations and the AKC requires seven. Perhaps the most important way to

ensure success is to engage the breed club's membership. Without them the project will fail or, like the Dalmatian project, may take decades to gain acceptance.

Beyond the political considerations, success must be measured in how well the descendants are integrated into the breed as a whole. Even with full registration, if they remain shunned by the majority of breeders, the benefit derived from the crossbreeding will be limited.

Crossbreeding is not a panacea for pure breeds with substantial health issues or which have lost once-valued traits. However, with due consideration and planning, it is a legitimate and effective practice which could benefit some breeds. To dismiss it out-of-hand is short-sighted and of no benefit to our dogs.

### **Breeding Strategies**

# Playing COI Using inbreeding Coefficients

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$$F_X = \sum \left[ \frac{1}{2} \right]_{1}^{n_1 + n_2 + 1} (1 + F_A)$$

Breeding dogs is a numbers game. Even though math problems are the last thing on your mind, what you are doing when you breed is calculating the best odds for getting a desired result. But a little applied mathematics, in the form of a coefficient of inbreeding (COI) can be helpful and even enlightening. Now that technology allows even the mathematically challenged to put them to use, COIs are a tool that should be applied by every breeder.

Research in the fields of genetics, immunology, and veterinary medicine, is turning up more and more information indicating that high levels of inbreeding can have deleterious effects on health. Inbreeding depression, a complex of behavioral and physical reproductive problems, has long been recognized. Inbreeding can increase the frequency of a disease in a population, sometimes quite rapidly. Inbreeding leads to increased incidence of immune-mediated disease and cancer.

All pure breeds of domestic animals are inbred. (Keep in mind that to a scientist "inbreeding" means the breeding of related animals, which would include what we call "linebreeding.") But how much is too much? Without it, the breeds could never have been developed and would not breed true to type. However, almost all breeds of purebred dog already have well-established type. There is no mistaking a Chihuahua for a St. Bernard. Or even a Greyhound from a Whippet. Given this, breeders would be well advised to retain as

much genetic diversity as possible within the existing breed population in order to avoid or reduce such unwanted health problems as those mentioned above. Along with screening and maintaining detailed health records, another tool available to you is the COI. Track COIs on your breeding stock. Calculate them on proposed matings, with an eye to keeping the numbers low if they already are or lowering them where possible.

So, how is this done? Via a formula called Wright's Coefficient of Inbreeding. It appears directly beneath the title of this article. Before you drop this publication in a mathphobic panic attack, be advised that the only practical way to use it is with a computer. For those who enjoy math or want more detailed background, there is an excellent discussion of applying the formula hands-on in Malcolm Willis' *Genetics of the Dog*, pages 320-326. For the rest of you, there are other options.

The easiest way to incorporate COIs into your strategy is to purchase a pedigree database program that will calculate them. Select pedigree software than offers COI calculation as a feature. You will also need a comprehensive pedigree database, including as many of the ancestors of present-day dogs as possible. Some vendors can provide starter databases for various breeds.

Now that we've soothed the math anxiety, what exactly is a COI? It is the probability that a homozygous gene pair will be

identical by descent from both sides of a pedigree. In the formula, Fx is your dog's COI, F<sub>A</sub> is that of the ancestor common to both sides of the pedigree. 1 and 2 are the numbers of generations on each side between your dog and that ancestor. In other words, if your dog Flux is a double-grandson of Fabulous the calculation tells you how likely it is you can get exactly the same gene passed down to Flux through each of his parents. (In case you are wondering, the probability is 12.5%.) If F<sub>A</sub>bulous happened to be a Collie Eye Anomaly carrier, Flux would have a 12.5%, or a one in eight chance, of having CEA thanks to Fabulous. Total actual risk would be dependent on whether there were any other carrier or affected individuals in the remainder of the pedigree, but whatever that might be, it is evident that F<sub>A</sub>bulous provided a significant part of it.

Multiply this times a dog's 20 thousand genes and it is apparent how quickly you can concentrate some genes—both good and bad—while others drift out of your kennel's gene puddle. Multiply that by all the people breeding a particular kind of dog and it can have remarkable effects on the breed's gene pool, especially if large numbers of them are making similar mating selections via the use of popular sires or heavy linebreeding on the product of successful kennels.

COIs can be calculated on any number of generations, the simple two-generation example of Flux and Fabulous was useful to make a point (and keep the math simple) but few purebred dogs have only one common ancestor on both sides of the pedigree and the more generations that can be included in the calculation the more common ancestors will be found and the more accurate the COI will be. The typical three to five generation pedigrees in common use are almost always insufficient. In my breed, the Australian Shepherd, five generations may appear to be loosely linebred or even outcrossed, but pedigrees extended to 10 generations will prove this is often not the case.

But how far should you go? How many generations to use will vary from breed to breed, depending on how many founders a breed had, how populous the breed is, whether there have been genetic bottlenecks, whether "new blood" has been introduced, and how long the breed has been in existence.

Some breeds descend from a very few individuals who are its genetic founders.

Samoyeds, for example, go back to about 20 dogs. Tracing everyone back to the founders in such a situation will result in COIs that may vary only by tiny fractions of a percent. Therefore selecting some intermediate number of generations for the calculation is the best option, unless the breed is very recent in origin and only a handful of generations away from those few founders.

For breeds with large populations, sufficient generations should be calculated that the results will have leveled out, so only tiny numerical differences will be achieved by pushing the calculation back a generation further. For example, if a one-generation COI is calculated on good old Flux, the COI would be 0%. You are considering only his sire and dam who obviously aren't going to be the same dog. Extending it far enough to include Fabulous (two generations) produced the 12.5% we saw earlier. What if the sire's dam was one of F<sub>A</sub>bulous's daughters? Going into the third generation would tell us the COI was 18.75%. If the dam's dam was out of FAbulous's full brother (linebreeding on the cross that produced F<sub>A</sub>bulous.), we go back to the 4<sup>th</sup> generation to include F<sub>A</sub>bulous's parents behind his brother the COI becomes 23.4%.

With each additional generation the COI will tend to climb but at some point the increase from one generation to the next will be negligible. COIs should be calculated over sufficient generations so that most current dogs will be at this point.

If breed population is small, preserving its remaining genetic diversity is vital. Calculate COI's back far enough to reach founders and then, working together, breeders can use them to equalizing representation of those founders in the over-all breed population. For example, if a breed had ten founders but most present individuals descended only from three of them, much of the genetic potential of the breed's gene pool is at risk of being lost as genes from the less-represented founders fail to get passed down by their fewer descendents. You can preserve under-represented genes by equalizing founder representation by giving breeding preference to individuals that do not descend from the most-represented founders and in avoiding crossing their descendents to each other. Since low-population breeds are at greater risk from genetic disease, due to "no place to go," maximizing genetic potential in this manner may be the line between extinction and

survival. In fact, it is the very technique used by zoos and others who are trying to preserve endangered species in captivity.

Some breeds have suffered genetic bottlenecks. By the end of World War II, many European breeds, including the English Mastiff and French Poodle, were reduced to a handful of survivors. Today these breeds trace their pedigrees back to those dogs, who are therefore effective founders. Even though known pedigree exists behind them, there is little point in extending a COI calculation beyond them. The only reason would be to determine how inbred those effective founders were themselves and what their inter-relationships might have been.

Sometimes a breed's gene pool may receive infusions of new genetic material. Some European registries allow registration of descendants of unpapered dogs brought in from the country of breed origin, such as a desertbred Saluki from Arabia. Occasionally, even such conservative registries as the AKC will, at a parent club's request, open a registry to new undocumented individuals. This has happened on several occasions since the 1980s when a few Basenjis were imported from Africa. Obviously, such "new blood" could have significant effects on the average COIs in a kennel or even throughout the breed, depending on how many dogs are added and how often. How much and with whom they are used in breeding will determine their contribution of new genes..

In calculating Australian Shepherd COIs, I use 10 generations because Aussie pedigrees are rarely complete to ten generations due to the breed's recent origin. By running 10 generations, I get pretty much all there is to get for my breed.

Once you have a PC, software that does COIs, a good breed database, and sufficient knowledge of your breed's history to decide how many generations to use in your calculations, it's time to put it all to work.

First, run COIs on all your own dogs. Since few dogs will have such diverse pedigrees that only one common ancestor will be found, the COI will be a reflection of all the ancestors common to both sides of the pedigree. In order to have a handle on what the numbers you will get mean, in terms of level of inbreeding, it is helpful to keep in mind what various percentages would be equivalent to, if there

were only one common ancestor or pair of ancestors in the pedigree:

25.00% - parent/offspring or full brother/sister cross

12.50% - half sib, grandparent/grandpup, or double first cousins crosses

9.75% - great uncle or aunt/great niece or nephew cross

6.25% - first cousins

Think back to our pal Flux and his 23.4% COI. He is very nearly the equivalent of a parent/offspring mating. If you bred him to his sister, given their already high level of inbreeding, the pups' COI would be 44.4%. Anybody glancing at Flux's pedigree would probably consider him inbred, and certainly heavily linebred. But it is possible to achieve high COIs without this kind of close breeding. Linebreeding on dongs several generations back can result in elevated numbers if the dogs appear frequently enough on both sides of the pedigree. While this inbred cross of FAbulous's grandpups might serve the breeder's short-term goals, it significantly increases the risk of turning up something unpleasant. And so would a linebreeding with a similar COI.

What's a breeder to do? We are breeding dogs not numbers and many factors other than COIs need to be considered when planning a mating. Even so, whenever possible you should try to achieve litter COIs that are at or below the average COI of the two parents. Thus, if the sire had a COI of 20% and the dam was 10%, you would want the pups' COI to be 15% or lower. If a kennel or line's average COIs have crept dangerously high, serious consideration should be given to avoiding further crosses to dogs descending from the most frequently seen names in the pedigrees and, as much as possible, to finding mates which are significant outcrosses.

The nice thing about COIs is that they can't be a secret. If you have a dog's pedigree, you can calculate the COI. In the privacy of your own computer station, you can figure out the COIs of all the prominent dogs in your breed. You can play with hypothetical matings between any two dogs you choose and see what the pups' COI would be.

For a real-life example, my dog Tank was the result of a father/daughter mating (their idea, not mine!) and had a COI of 40.9%. No doubt about it, he's inbred. Using my pedigree

software I can set up all kinds of hypothetical social activity for the old boy and see where the COI goes. He is heavily linebred on a particular stud dog of a number of years back. However, that dog is not common in most show-line Australian Shepherd pedigrees, so I can easily find mates—even those with fairly high COIs themselves—who will give him puppies with much lower COIs. If I'm really determined I can hypothetically mate him to working-line Aussie bitches and in many cases I will drop the COI to less than 5%. All of this without having to risk finding out what any of the owners of those bitches (especially the working breeders!) think of the idea of poor old show line Tank having a fling with their girls.

Getting reliable hereditary disease history on your dog's ancestors and on his

potential mates can be difficult to impossible. If you know your dog has family background for a disease and there is no available testing to let you know whether he might be carrying the genes for it, breeding for low COIs while at the same time avoiding doubling-up on any ancestors you know are problematic may reduce your risk of producing the problem. With a lower COI you are lowering the probability of pairing on those unwanted genes you know are back there somewhere.

Every breeder should play COI: Coefficients of inbreeding are an important tool to apply to your breeding program. Whatever the needs of your kennel or your breed, COIs provide you with a vital bit of information that should be part of your decision-making process.

# The Impossible Dream Breeding for Perfection

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When breeders discuss goals the catchphrases "improving the breed" and "striving for perfection" are all but routine. But what exactly are perfection and improvement? Are they even achievable? For that matter, are the terms even meaningful?

This isn't to imply that breeders are spewing hype with no goal greater than attracting business. What most mean by these phrases is, "I'm trying to produce the best quality dogs of this breed possible." But in the process the concept of quality gets entangled with ideas of improvement and perfection in ways that may not ultimately benefit the breed.

### A contradiction in terms

Consider improvement. On the face of it, that might mean moving closer to perfection. Even if one assumes, for the sake of argument, that perfection is a constant, most purebred dogs are bred for show. Show breeding tends toward the exaggeration of physical appearance.

When a breed has a signature trait, such as size, coat type, or a distinctive shape of head, that trait frequently becomes more exaggerated over time. The Pekinese once had a muzzle, the American Cocker Spaniel's coat did not always flow to the ground, and the

Dachshund's back was not nearly so long nor low to the ground. Big dogs are bigger, small ones smaller and in coated breeds the competence of the groomer can be just as important as the qualities of the dog for success in the ring.

Is this type of change truly improvement? In some cases it renders the dogs unsuitable for their original purpose or predisposes them to health, soundness or maintenance difficulties. There is a term coined by enthusiasts of vintage architecture to describe improvements gone wrong: Remuddling. To find a remuddled breed one needs look no farther than the modern English Bulldog, which cannot even reproduce without significant human intervention

#### If it ain't broke...

In some cases, maintaining the status quo may be the best tactic. Author and Border Collie trainer Donald McCaig has said "Refining a breed is a mistake. The best we can do is leave it no worse than we found it."

This is obviously true when the improvement has rendered the dog more difficult to work or live with. In the case of a function breed still regularly utilized, like McCaig's Border

Collies, if it is already doing the job supremely well meddling may lead to disaster.

The tides of breed fashion heavily influence the concept of improvement. Look at historical photographs of most breeds and you will see marked differences between the greats of today and those of several decades or a century back. As the needs and preferences of people change, so will the traits considered ideal in a dog. Success by one breeder can spur others to "improve" their stock so it more closely resembles that of the winner. Much of this revolves around esoteric aspects of appearance rather than essential breed traits. Chasing fashion in this manner can lead to a narrowing of the breed's gene pool as one or a few lines are favored above all others.

#### Setting the standard

Written breed standards were developed to serve as descriptions of ideal dogs. Their "original purpose," if you will, was to serve as a blueprint for generations of breeders, without the risk of change or loss inherent in the transmission of oral history and knowledge. But standards may contain errors—as with the persistent myth of the 45 degree shoulder. Standards also can vary from country to country or even within a breed. There are two US standards for the Australian Shepherd, the original one developed by the Australian Shepherd Club of America, an independent club/registry, and the AKC standard. They are very similar but there are differences. Which of the two is "more perfect" depends upon whom vou ask.

As discussed earlier, a successful show and promotion campaign or the whims of fashion may redefine perfection. Those whims may or may not be supported by the breed standard, but that doesn't prevent breeders from breeding for the new style. The AKC standard for the German Shepherd Dog describes the back as "level" and says it is "straight, very strongly developed without sag or roach..." The illustrated version of the standard shows this clearly, yet the outline of many show dogs today does not conform to the outline in the illustrations. The roached back has become so common that detractors have coined the phrase "banana back" to describe it.

Standards cannot adequately address health or behavior. Keeping up appearances is important in breeding show dogs, but excessive focus on show points can obscure the importance of other aspects of the breed.

Failing to consider these as part of the perfection package can destroy a breeding program. Can the most beautiful, smoothmoving dog in the world truly be perfect if it has or produces health or behavior faults?

And standards can be changed, sometimes to adjust the wording to reflect what the breed has become: What was once perfect, ceases to be so.

#### Different strokes for different folks...

Any breeder worthy of the name wants to produce quality animals, though the definition of "quality" will depend on that breeder's larger goals. Are these to be show dogs? Pets? Performance event dogs? Working or service dogs? Or perhaps some combination of these? Each breeder needs a clear vision of her goals. If the breed is multi-purpose, she must have a good understanding of how dogs bred for a different sets of goals will (or will not!) meld with her own stock. Those other breeders' goals are not necessarily less worthy, but differing goals may be mutually exclusive.

Times do change and so do dog breeds in response the evolving needs and preferences of people. Cultural and technological changes can have a huge impact on a breed. The breed's original purpose may cease to exist or becomes so insignificant that few dogs are required to perform it. In such cases change is inevitable as the breed is adapted to suit a new role.

When some breeders pursue traditional, function-based goals while others breed to produce show dogs, pets, or dogs used for a different kind of function a significant divergence in type and behavior can arise between the different strains within the breed. More than a few breeds have split along show and function lines, sometimes to the point that what was once a single breed is essentially two, with little or no genetic exchange between them.

The idea that a breed's original, functional, purpose can be maintained by maintaining a particular appearance without at the same time carefully selecting for the necessary behaviors and instincts is a common misconception. Some part of those behaviors may be preserved, but unless they are an integral part of the definition of breed perfection, they will lessen, fragment or be lost entirely. Show ring perfection cannot be achieved consistently unless breeders keep that goal firmly in mind, so how could one logically

assume that other complex traits would be any different?

When it comes to breeding for purposes other than the show ring, written standards may not even apply. The International Sheep Dog Society maintains no standard for working sheepdogs, operating on the premise that the proof is in the pudding. ISDS Border Collie breeders look for their vision of perfection in the field, not on a piece of paper. There is no published standard for what makes a good pet, either, and most dogs today are companion animals.

No dog can be all things to all people. Very few dogs exhibit high quality in more than one or two areas of canine endeavor. The experienced breeder knows what he's aiming for and will select only those dogs that meet his criteria. However, those rejected dogs might be valuable to another breeder with a different set of goals.

#### Reality check

If the definition is narrow enough, perfection may be attainable. But what is that sort of perfection worth? A breeder must consider the whole package—appearance, behavior and health. Health issues are often the neglected stepchild of perfection, a consideration that is secondary to the prime goals of producing the best show or function dogs.

Poor health is anything but perfect, occasionally inspiring novice breeders to declare they will not breed any dog that has any health risks, however minor. Not every genetic defect is equally "imperfect." One can't put a missing tooth on the same level as cardiomyopathy or epilepsy any more than a minor color fault is on the same level as a significant structural defect. Genetic perfection does not exist. No breed or line, no matter how carefully selected, is without at least some risk for something unwanted. Novice breeders will either learn to set realistic goals or get out of the game.

The trick is to have a comprehensive vision of the ideal dog that includes health and behavior as well as appearance, with a view toward the purpose these dogs are bred for, whether it is work, show or companion status. The vision should be far-sighted, encompassing not just at the litter this year but those that will be born decades hence. It must be substantial enough to withstand the buffets of passing fancy.

True perfection is a myth. The best of breeders know that, but they develop a clear vision of the breed based on knowledge of its history and a thorough understanding of the health and behavior of dogs. When a breed is blessed with enough breeders who share such a vision, quality of the breed will be maintained into the future.

# What Price Glory? What Happens When Winning is Everything

First published in Double Helix Network News, Fall 1999

Nearly twenty years later, I remember baiting Shadow with an empty hand in a Best in Match ring lit only by streetlights. I ignored the crisp November air as, with one eye on the judge, I concentrated presenting my dog at his best. Australian Shepherds were not AKC recognized. Only at matches like this could they go head-to-head with the other breeds. Shadow had a shot at the top prize in one of the largest all-breed matches in our area, and I'd run out of liver half way through Working Group.

Soon we were at the head of the line. The judge examined Shadow and moved us. As we circled the ring to the end of the line, the watchers applauded. These were no semi-organized group of friends and admirers. Most the Aussie people had long since gone home. Shadow's progress was cheered by strangers who appreciated a good dog.

The judge continued with the individual exams. I kept Shadow "on," in case she glanced our way. My handsome black tri kept his gaze intent on my liverless hand. I thanked the gods of dog shows that he was an incurable foodhound.

The judge finished with the Toy Group winner then motioned us all to gait around the ring once more. The Borzoi leads off, followed by the Pointer. I moved Shadow out, easily keeping pace with the leggier beasts while the smaller dogs scribed a narrower circle behind us. The judge held her hand overhead, watching us intently. Shadow didn't' miss a beat. I hoped I wouldn't.

Her hand dropped. She was pointing to me! Shadow had won Best in Match.

Winning that show was one of the highlights of my years in conformation competition. I can relive those heady emotions a score of years later. To win, and win something big, is a wonderful experience. But it was, after all, only a dog show.

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Exhibition of purebred dogs started about a century and a half ago as an outgrowth of stock shows, where breeders would exhibit their finest cattle, sheep and hogs. As with the stock shows, the intent was to showcase fine breeding stock. But over the years the emphasis has changed from providing a showcase to a major criterion for determination of "breeding quality." The old adage "breed the best to the best" has subtly changed in meaning toward breeding the best-looking to the best-looking. Success in the conformation ring has become so important that a few will subvert the process to ensure greater success.

Today, the conformation show system in the United States has spawned an entire industry. People can and do make a living producing canine competitions, handling dogs and selling a wide variety of goods and services targeted at exhibitors. Breeders advertise major wins and publish lists of their titled dogs—too often with no mention of pedigree or health clearances. There is nothing wrong with celebrating a win or taking an opportunity to crow a little, but titles and wins, however prestigious, are not inherited by a dog's puppies.

A good judge will recognize physical quality in a dog, but due to its nature the conformation ring offers little opportunity to evaluate a dog's mental acuity and physical stamina. Even in breeds where gait is emphasized, a few laps around the ring can only point out the extremely unfit. A good breeder should be equally competent to evaluate a dog's

physical attributes and shouldn't require outside opinions, in the form of show wins, to bias his judgement. In addition, the wise breeder will make an opportunity to observe and interact with dogs away from the shows to gage their mental and physical mettle.

Breed-oriented Internet discussion lists go on at length about the importance of breeding to titled dogs, especially Champions . Special emphasis is put on pedigrees which feature the titled "greats" of the breed. Titles are the tail that wags far too many dog breeders..

Conformation shows have become an end rather than a means. People are breeding dogs to win dog shows instead of using the shows to present their breeding efforts. If a dog does not win, is it without question unworthy of breeding? If it wins a lot, should it necessarily be bred a lot? While on the surface this might seem to make sense, the reasoning is flawed.

A dog's worth as a breeding animal is dependent upon its genetic makeup. Its appearance and behavior in the ring can give an indication of its potential, but all the ribbons and trophies in the world won't help if it doesn't pass those qualities to its offspring.

In the US, considerable emphasis is placed on presentation and, in coated breeds, grooming. An inept handler or groomer can make the best dog look bad. Conversely, a talented handler or groomer can present a mediocre specimen in a manner that distracts from its weaknesses. Good handlers can school a dog with the proper disposition to exude presence. The kind of dog which makes bystanders say, "Look at him! He's asking for the win.' Ring presence is at least as much nurture as nature. Is a dog that "asks for it" a better breeding candidate than the one with better structure but a less showy attitude? A dog's grooming or show presence may be a credit to the person who put the work into creating them, but they won't go with the puppies unless the breeder includes the groomer's or handler's business card along with the papers.

Winning has become so important to some people that they employ a number of practices to improve their odds of winning. Some are as simple as carefully studying judges and choosing which to exhibit a dog under.

Others will alter their dogs to better conform to the standards or the current winning fashion. taping or gluing of ears to correct the set, dving of coats to cover color faults, tattooing pink skin that should have been pigmented, straightening crooked teeth, surgically altering tail sets or other physical features, and a variety of other practices. All of these alterations disguise the dog's phenotype—the look that his genes gave him. In spite of this, the genes remain what they are. The prick ears, gay tail or bad bite will come out in the progeny. Unfortunately, the person who bred to the dog or bought its pup may be ignorant of the alterations and assume she is buying genes to produce what she sees in the ring.

Even when a top winner is equally outstanding as a sire, the emphasis on his wins can lead to the popular sire syndrome. No matter how good the dog, he will have a few bad genes. The wide use of a popular sire and subsequent inbreeding and linebreeding on him will increase the frequency not only of the genes everyone wants, but those they don't want as well. If a dog and his near kin are used too extensively, particularly in breeds with small populations, there may be few places to go when the unwanted genes make their presence known.

And what of the dog that doesn't win, or maybe never sees the inside of a ring? Is it, of necessity "not breeding quality?" Dogs are as individual as people. Some do not like shows and will not show well. If the animal is in other respects an excellent example of the breed, why pass it by? What of the bitch scarred by some

accident, precluding her from any chance at a win? That scar or injury won't be inherited. If she's a good quality, why not breed her? Occasionally a fine animal will belong to someone who hasn't ever shown it and doesn't want to. In most cases, such dogs will belong to people who acquired them as pets. Pet status is not synonymous with poor quality. A knowledgeable breeder who knows the dog's pedigree background and how its near relatives have produced may be able to make excellent use of that dog in his breeding program.

The relative success of any two dogs in the ring has no dependable correlation with their success as breeding animals. The big winner can be a dud stud while the dog with a more modest competitive record may throw marvelous puppies. The youngster who seems to be winning everything and has thrown puppies with promise is a greater gamble than the mature dog with a less stellar career. You will know what that older dog is and probably what he has produced, but a few years down the line that bigwinning wunderkind could mature into mediocrity or develops a late-onset hereditary disease.

Winning is great fun and it doesn't come easy. Those who are successful deserve to be congratulated for their efforts. But those wins should never be a prime consideration when making breeding decisions. What that winner is, what he does and what he produces *outside the ring* are far more important to breeding than even the highest titles or top honors from the most prestigious events.

# Genetic Juggling How to manage polygene traits in your breeding program

First published in the Fall 2009 issue of Double Helix Network News

Traits with simple, single-gene inheritance are easy for breeders to deal with. We all know how to get merle pups in our litters if we want them—or not, if we don't. But when it comes to complex traits, those that are governed by multiple genes and also sometimes have environmental components, the effort to

maintain or avoid them can cause breeders endless frustration.

Many desired traits are polygenic. Some behavioral traits like the working stockdog's ability to move livestock, or aspects of conformation like dentition or the front end assembly, are the result of polygenic inheritance.

Experts agree that multiple genes are involved in hip dysplasia and that exercise levels and nutrition are contributing environmental factors. Autoimmune diseases are genetically predisposed, with genes setting the dog up for illness and environmental factors making the final determination on who gets sick and who does not. Even coat color, which we tend to think of one gene at a time, is actually polygenic. If you are breeding Pomeranians or Chihuahuas and want to breed away from merle, if you not be able to determine whether your yellow or sable dogs have the merle gene.

#### What's a breeder to do?

The first step is to use whatever phenotype screening tests may be available for traits of concern: Hip or elbow x-rays, eye exams, etc. Screening exams will tell you what version of the trait the dog exhibits. Knowing your dog is OFA Excellent is a big check mark in his favor, but it isn't the whole picture. For that you would need to know his genotype—what genes he has—and therefore what he as potential to produce. For that you need a DNA test and, as this is written, there are no DNA tests that will give you your dog's genotype for most or all of the genes involved in any polygene trait.

But sometimes there is a test that will give you part of the picture. In the case of hip dysplasia, researchers in Germany have recently discovered that there is a gene of major effect, a gene with a significant impact on the phenotype, one version of which is highly associated with dysplasia and, interestingly, more so for one hip than the other. There isn't a test yet available, but if one is eventually offered, knowing your dog's genotype for this particular gene can help you make informed choices when breeding him.

The phenotype of any one individual isn't sufficient. You also need to know something about her family history for the trait in question. What do you know about her parents, grandparents and other ancestors? Equally important, what do you know about near relatives that are not directly on the pedigree – her siblings, aunts and uncles, and cousins? Dr. Jerold Bell, a Gordon Setter breeder and veterinarian who teaches veterinary genetics at Tufts University, has said, "The most important factor in selecting against a polygenic disorder like hip dysplasia is to seek breadth of

pedigree." By "breadth of pedigree" he means the status of these collateral relatives.

#### Do you know where your puppies are?

Offspring are an important source of information. Check in periodically with the owners of puppies you have sold or placed so you can stay up-to-date on how they are shaping up and whether any problems have arisen. If you are seeing something frequently, good or bad, in half siblings it indicates that the common parent dog has a higher than average collection of genes for that trait. If the trait is a good thing, you can list it among the dog's virtues as a breeding animal. If it isn't, the knowledge will help you make better choices in who he should or shouldn't be bred to.

Information gathering is an ongoing process. Develop a method of recording information on traits you feel are important so you can readily retrieve the information when you need it. The best way to do it is by utilizing pedigree software that allows you to append notes to a dog's record. That puts the information right where you need it as you research pedigrees. Be sure you develop a systematic way of searching your database that will allow you to retrieve the information you have on the collateral relatives, too. Over time and with cooperative information sharing among breeders, you can gather significant amounts of data to review when you are considering potential crosses.

#### Finding Fault

Both parents contribute to a polygene trait, so there is no future in he did/she did finger-pointing game if an unwanted polygenic trait comes up in a litter. Both parents passed genes to the pup that allowed it to happen. However, the contribution may be unequal. Some dogs will only occasionally produce something like HD, others will have multiple affected offspring. The dog that produces the trait more often, especially with multiple mates, carries a heavier load of genes for the trait than one that does not. Recognizing this enables you to a better job of breeding for or away from the trait.

If you want to preserve a polygene trait, you should to start with a pair of dogs that have the trait. If they also have numerous relatives that exhibit the trait, even better. Best of all would be that the dogs and their relatives are known to produce the trait consistently. With this kind of individual and family history the odds

are in favor of there being puppies from your pair that will have the trait you want.

If there is a trait you don't want, you wouldn't select breeding stock that had it. But you must also make sure there is little of that trait in your dog's family. If she produces it too often, you might do better to use someone else for breeding.

### **Setting Priorities**

All traits are not equal. If you have many good things in a dog that are consistently produced it his family, but there is one consistent trait that you don't want, you need to look for mates that are not closely related to your dog (because you know the problematic genes are there) which are clear of the trait. have not previously produced it, and have little or no family history for it. Such a mating will reduce the risk of getting the unwanted trait in the litter. Even if everything is perfect with the litter, at least some of them will carry the problematic genes, so those which are bred will also need careful mate selection to reassembling the polygene set and producing the unwanted trait.

In many cases, breeders have to do a risk/benefit analysis of proposed matings to capitalize as much as possible on desirable polygenic traits while minimizing risk of getting the undesirable. But when there is a DNA test available, they can use it to skew the odds in their favor.

#### Science to the rescue!

At this point we only have single-gene tests available, but some of those are for genes of major effect, genes that are key to how or whether a trait develops, in polygene traits. One example is the CEA/CH gene. The gene mutation is the one that causes choroidal hypoplasia (CH,) the most common Collie Eye Anomaly defect. CH is a relatively minor eye defect; most dogs with it have functional vision. However, other CEA defects, like optic nerve coloboma and detached retinas, can be blinding. The more serious defects are caused buy other. as yet unidentified genes. However, no dog gets CEA at all unless it has a double dose of the CEA/CH mutation. Because this gene's actions are so predominant in this disease, CEA can be treated like a single gene recessive even though we know there are other genes that contribute to the phenotype. And, since we have a DNA test for that key gene, there is no need to remove carriers from the breeding pool

so long as we breed them to non-carriers. Over time, we can give preference to the non-carriers and reduce the frequency of the mutation in the breed.

But most polygenic traits, even those with a test for a gene of major effect, aren't so cut and dried. Cataracts in Australian Shepherds are an example. A dominant mutation in a gene called HSF4 is associated with approximately 70% of the cataracts in the breed. HSF4 was first found to have a mutation causing a simple recessive form of cataract in Boston and Staffordshire Bull terriers. In the process, researchers found the Aussie mutation. too. Not every Aussie with cataracts has the HSF4 mutation and some dogs live out long lives with the mutation and never get cataracts. Even if you rule out those cataracts caused by environmental effects (injury, infectious or acquired diseases, nutritional imbalance) there are still some dogs without the mutation that will develop cataracts.

#### State of Confusion

The lack of black-and-white answers from the HSF4 test has lead to great confusion among some breeders, spurring calls to reject the test as inaccurate or useless. Such opinions are short-sighted. The breeders who espouse them are rejecting a tool, albeit an imperfect one, that can help them reduce the frequency of cataract in their lines. The truth is that most of the simple and easy one-gene traits have already been identified. From here on out, Most of the gene tests developed will be for genes like HSF4 that indicate a risk factor instead of a sure thing. Even so, the results of these tests put us in a better decision-making position than where we are with most polygenic traits, like the autoimmune diseases, for which we have no DNA tests at all.

The HSF4 mutation in Aussies is highly (70%) associated with cataracts. It is dominant, so a dog with only one copy is just as likely to develop them as one that has two. Cataracts are very common in the breed and often, though not always, ultimately blinding. In general, the best advice is not to breed dogs that have even a single copy of this mutation. However, the test is relatively new (released in March of 2008) and some breeders have made the unhappy discovery that many of their dogs have it. So, should these breeders dump all their stock — including animals that have many very find traits — and start all over? Of course not.

If you find yourself in the unfortunate position of having a breeding program riddled with HSF4, you can take steps that will allow you to hang onto the baby while you drain the dirty bathwater. Since you can positively determine the HSF4 genotype, breed your best individuals that have one copy of the mutation to clear mates. Select the best clear offspring from those litters to carry on with. You will have eliminated the problem in one generation. If you have the misfortune to have an otherwise exceedingly valuable animal with two copies, breed it to a clear mate. All the puppies will have one copy of the mutation, but if you breed the best among them to clear mates, and select the best from that second cross to carry forth with, the mutation will be out of your breeding program in two generations.

The HSF4 Aussie cataract mutation is of a type called "incompletely penetrant," meaning not every individual with the mutation has the trait. The penetrance for this one is fairly high, but sometimes penetrance will be low. Such is the case for the Juvenile Renal Dysplasia (JRD) mutation.

JRD is well-studied in the Tibetan Terrier. A dominant genetic mutation was found in that breed occurs in all dogs that have the disease. However, many dogs have the mutation but never become ill. Since only a small number, estimated at 3-5%, of the dogs with the mutation become ill, there is no point in removing healthy dogs that have the mutation

from breeding. However, those that do become sick often die and those that aren't killed outright by the disease often have shortened life-spans. So, what to do?

#### Decisive Action

The JRD mutation should be treated as a major fault. It isn't a fault that will move you to the bottom in a conformation class or get you excused from the performance ring, but it is a fault nonetheless: One that in a few cases will be fatal to the dog. The best thing breeders can do if they discover a healthy, breeding-quality dog has this mutation is add that fact to the list of pros and cons for this particular dog. If the good stuff outweighs the negatives, including JRD, then breed it to something that has tested clear. As outlined above with HSF4, you can be free of the mutation—and the worry that you might produce a dog with clinical JRD—in a maximum of two generations.

Polygenic traits, both good and bad, can be managed in a breeding program, the key is good record-keeping, doing your homework before you put dog to bitch and, where possible, making educated use of available DNA tests. We are far from the point where we can guarantee all outcomes in our litters, but the more informed you are the better equipped you will be to work toward the outcomes you want.

# The Decision Tree A tool for better breeding

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Top breeders don't get that way by accident. Each and every breeding is preceded by careful planning drawn from an encyclopedic knowledge of the breed and of individual dogs, past and present. Top breeders have standards that guide their decision-making process and a clear view of what they want to accomplish, not only in individual breedings but with their line of dogs. It requires knowledge, determination and experience.

So how do you, an up-and-coming breeder, get where they are? While accumulating knowledge and gaining

experience, you must develop a consistent and thorough evaluation system that will help you select breeding stock or a stud for your bitch and, if you are a stud owner, determine whether or not you want to make him available to a particular bitch. What you need is a decision tree.

#### Growing a system

A decision tree doesn't have leaves or roots, but it can sprout branches. It is a graph indicating starting and finishing points, and routes through all the decision points in

between, including possible alternate paths to yourr final decision. For those who learn visually, sketching out a graph like this might prove helpful. Those who are not visual learners may want to develop a different approach, but you still must have a way to work through all the important variables.

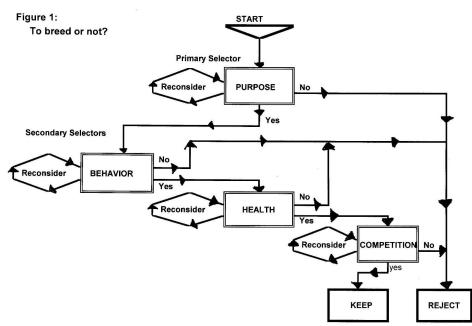
Like real
trees, your decision trees can
be dynamic and changing, as
you adjust them to suit new or
different needs. You may
"grow" a branch here, or
prune one there. Do what
works for you and be willing to
adjust should the need arise.
The charts used in this article
are basic. You may think of additional decision
paths you want included for your own purposes,
or want to re-arrange the sequence.

Before you can begin, you have to know your starting point. What are the strengths and weaknesses of each of your breeding dogs? What are your short- and long-term goals as a breeder? You need to consider and prioritize all pertinent issues of conformation, performance, behavior and health.

#### **Pruning**

You can't be a top breeder without quality dogs. You need to evaluate each dog's worth as a potential breeder either before you buy or, with a puppy, as it matures. One can never know for sure if a puppy will grow into a dog adequate to your breeding needs. Figure 1 is a decision tree chart showing the major decision points for whether or not to keep a puppy for breeding.

The primary selector, your first decision point, is based around your primary goal as a breeder. This might be conformation, some area of performance competition, or even real-world work. Whatever it is, the puppy must meet certain basic criteria for your chosen field or there is no point to keeping it in your breeding program even if it is a stellar individual in all other respects.



Sometimes a puppy may require reevaluation before a final keep/don't keep decision is made. It may have some minor conformation faults that could improve with age or not yet be showing sufficient interest in livestock. The loop away from and back to the decision point for the primary breeding goal indicates the potential need to wait a while and re-evaluate.

If the puppy passes muster for your primary selector, you need to move on to several secondary selectors. There should be at least two (behavior and health) but might be more, depending on your particular circumstances, as has been done in Fig. 1. For example, if your primary area of interest is some sort of competitive event, you want a secondary decision point for that. (Or multiple points if your dogs compete in more than one venue.)

While Fig. 1 shows these secondary selectors in a particular sequence you could rearrange the order to something that better suits your own practice. In actuality, you probably will be working on all of these things at once. The purpose of the chart is to focus your thinking so you don't let superior qualities in one area cloud your evaluation of others: Superb conformation and great temperament are wonderful, but if health screening results are disastrous, the pup ceases to be a good breeding candidate.

A similar chart can be used if you want to evaluate an adult dog you might purchase or leasing. Changes might include additional secondary selectors (evaluating its family members, breeding history.) For an adult dog, you wouldn't need so many re-evaluation loops; you won't be waiting for it to grow up and health testing may already have been done.

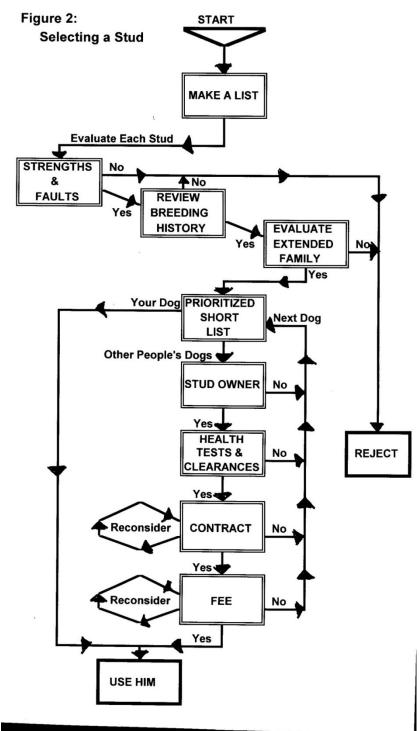
#### **Pollination**

Consider another common breeder's decision: Selecting a stud for your bitch. (Figure 2) The first thing you need to do is put together a list of possible candidates. Even if you think you want to use a particular dog, going through the process will help you determine whether your prime target actually is the best option for this particular breeding. A secondary benefit of evaluating multiple studs is that you will gather data that may prove helpful in future breeding decisions for this bitch or some other you own.

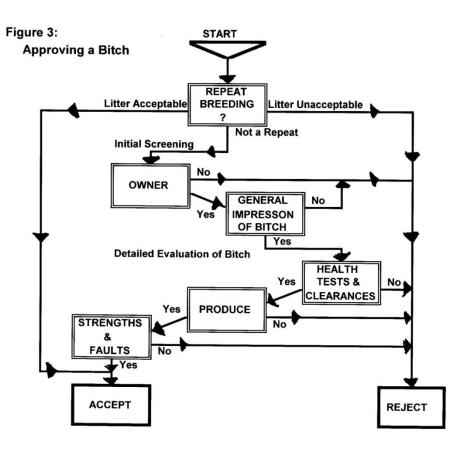
Once you have a list, you must research and evaluate each dog. As with the secondary selectors in Fig. 1, these evaluation points might be put in any order and, in actual practice, done concurrently. Study each dog's strengths and weaknesses. If he's been bred, what are the qualities and weaknesses of what he's produced with special attention to whether he throws his own faults or those of your bitch. Finally, and especially important if the dog has not been bred before or his offspring are too young for meaningful evaluation, what strengths and weaknesses are common among his family members, including direct ancestors and their siblings and offspring plus the dog's full and half siblings and their get.

Once you've evaluated all of the dogs, the next decision point on is to develop a prioritized short list of potential sires for your litter.

If you own the dog at the top of this short list, your decision is made. If not the key decision point is the dog's owner: Is this



someone you are willing to do business with? If yes, then have the necessary health clearances been done? (In this author's opinion, any dog at public stud should have had every test available for the breed and be current on those, like eye exams, that must be repeated.)



your standards, so you won't repeat; or no this hasn't been done before.

In the last case you move to the initial screening decision points. As with selecting a stud, the first consideration is the bitch's owner. A stud owner has little or no control over what happens with the litters. Is this person someone to whom you want to entrust your dog's puppies? If you expect the arrangement to be satisfactory or you have only minor concerns which can be addressed through discussion or adjustments to your contract, it is time to move on to the next decision point: Evaluating the suitability of the bitch for your stud.

What is your general impression of this bitch? If something negative immediately stands out, you'll want to give her a pass. If not, you can proceed to details.

You want to make sure she has all necessary health tests and clearances and that the results of DNA-based tests are complementary to those of your stud. Though it is not included in Fig. 3, you might want a reconsideration loop on this decision point in case tests haven't been done and you are willing to reconsider after results are obtained and presented to you.

If all health issues are acceptable, you need to review whatever you know about her prior litters (if any) and make a more detailed review of the bitch's strengths and weaknesses to determine whether the potential litter should meet your standards for quality.

If your review indicates the two would not complement each other it is best to decline the breeding. Potential problems that are either at a low risk of occurring or will not have serious repercussions should a puppy inherit them should be discussed with the bitch owner and the two of you must come to a mutual agreement on whether the potential benefits of the mating outweigh the risks. Decision points for contract and fees are not included here because you, as the stud owner, get to call the shots; you don't have to negotiate if you don't want to.

If health evaluations are in order and complement those of your bitch (you wouldn't want to breed two carriers of a recessive health issue to each other) it is time for business negotiations: The contract and the fee. The stud owner will provide you with a contract and tell you the stud fee. If either is a problem for you, you may be able to negotiate a more acceptable deal, hence the reconsideration loops.

If you run into something unacceptable at any of these points you return to your short list and start the process over with the next dog.

#### Planting seed

The final decision tree example (Fig. 3) puts the breeding shoe on the other foot: Screening bitches for your stud dog. Everything else depends on the answer to this question: Has this particular mating been done before and, if so, what happened? There are three possible answers. Yes and the litter was acceptable, in which case you would do the repeat breeding; yes and the litter did not meet

#### Planting a Grove

These are only three examples of decision trees related to dog breeding. You may be able to think of more or come up with alternative approaches to the basic breeding questions evaluated here, but the point is that if

you want to succeed as a breeder you must develop a consistent and thorough method of evaluating all pertinent information. Until you have decades of experience and knowledge under your belt, you need a system that helps you cover all the bases. A decision tree is one way to make that happen.

# Not WHO You Know But WHAT You Know The importance of good data to good breeding

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Some breeders excel at producing quality dogs over the course of decades while others have only a brief flash of success or none at all. The thing that separates those with long-term success from everyone else is the knowledge base they draw upon to make breeding decisions. Some people develop an incredible mental storehouse of information on dogs present and past. If you are fortunate enough to have a capacious and razor sharp memory you are doubly blessed. For most of us, diligent record-keeping is advised.

Even if you do have a great memory, maintaining a comprehensive set of data is a good idea. Age can play games with recall even for the sharpest elder and if nothing is ever recorded, a senior breeder's knowledge can be lost to subsequent generations. For your own sake and the sakes of those who follow you, gather as much information as you can on your breed and store it in an easily retrievable fashion.

#### Where to begin?

The first step is knowing what kind of data to collect. Details will vary depending on your breeding goals. A show breeder will have different concerns and priorities than someone managing a guide dog breeding program or breeding competitive performance dogs. But whatever the specifics, information worth storing will fall into three general areas:

- What a dog is
- What it has produced
- What its relatives are and have produced

What the dog is includes its appearance, behavior, skills and accomplishments in competitive events. When using this information keep in mind that some things are highly influenced by environment: The best dog may not win consistently if its preparation and handling aren't equally impeccable. Even for traits that are entirely a matter of genetics, the appearance or behavior of the dog itself may not reveal its genetic makeup: You can see that a dog is black, but if you could look at its genes you might also know that it carries liver, tan trim and yellow.

The best information you can gather for health is results from DNA screening tests. They aren't available for all health issues, but where they are they will tell you the genetic make-up of the dog. It doesn't matter what its ancestors or other siblings were, with these tests you know exactly what the dog can pass along to its offspring.

That brings us to the production record. The more you know about what a particular dog has produced, the better idea you have of its genetic makeup. A dog that consistently produces traits you want is likely to do so again. Hence the old breeders saying, "If you like the son, breed to the father."

Finally, knowledge of a dog's extended family will further enhance your understanding of his genetic potential, both for good and for ill. Consider hip dysplasia; breeding the highest scoring dogs to each other will not significantly reduce the amount of HD. However, if you combine hip scores (what the dog is) and information about the hip status of its offspring, predecessors, and collateral relatives (those just off the printed pedigree) you are in a much

better position to reduce the risk of producing dogs with HD.

#### The search begins

Knowing what kind of data you need is only a start. You also need to know where to find it. A lot of useful information can be gathered by personal observation. Watch dogs at events; not just in the ring or arena, but out of it as well. If possible, speak to the owners or handlers and interact with the dogs themselves. If you have the opportunity to observe dogs in their homes or in casual environments, all the better. This is a great way to learn about a dog's movement, structure, and behavior.

Where available, hard facts are best, but some things aren't apparent to the eye or a dog that interests you may live somewhere you aren't able to observe it. Sometimes breeders will share video clips, but these are marketing tools and therefore will be edited to reveal the dog in the most positive light. Networking with fellow breeders and other breed enthusiasts will help fill in the gaps in your data and provide new information. Another breeder may live close enough to a stud that interests you to give you her personal impressions of that dog.

Gathering data through others has its drawbacks; it can take a while before you know someone well enough to determine whether he is an honest and reliable source. You need to develop a filtering system that helps you decide which bits of information you glean from others are trustworthy and which are not. Part of this is learning to become a good judge of human nature, but a simple rule to follow if you have doubts is not to accept something as fact until it is independently confirmed by a second person in a position to know who has no close ties to the first one.

Most breeders maintain a website. These are also marketing tools. Even so, the pictures and other information offered can give you valuable insights into the breeder's dogs and breeding goals. Some breeders will also post significant health information but this is by no means a regular practice.

For health information, online databases like those offered by OFA and a number of European kennel clubs can tell you about health test results on numerous dogs in your breed. Some health information can be gleaned from breed magazines and newsletters, along with things like titles earned, event results, competitive rankings, and upcoming litters.

Keep in touch with all your puppy people; they hold the best information about how your breeding program is progressing. Dr. Carmen Battaglia, a German Shepherd Dog breeder who writes and lectures extensively on dog breeding, holds an annual picnic for his puppy people and their dogs. This gives him an opportunity to talk to them and see and handle the dogs. He also takes pictures and video for future reference.

Information can be gathered in various forms. Keep a notebook to jot down phone notes and significant observations when you are out and about. Keep copies of correspondence. Collect health screening certificates and other important documents on other dogs as well as your own. Mark your event catalogues; they tell you not only who placed how but provide ownership and parentage information. Don't neglect visual media. In this digital age collecting and storing stills and video is easy and relatively inexpensive.

#### What's Important

The type of information you gather will be dictated by your breeding goals. If you breed your dogs for any type of competition, you will want as much information as you can get about how dogs have performed; not just your own but any dogs whose bloodlines you might someday tap. At any given event, did the dog win, place or was it out of the running? What dogs placed over it? If the event involves a scoring system, what were the scores? Who was the judge?

Environmental issues—weather, a change of handler, a rough trip to the event, etc.—can impact performance, so make note of anything that happened which might have influenced the result.

Once you have enough data on a particular dog, or a related group, you can look at the overall record to see what is consistently good or bad and what isn't.

If your chosen area of competition is conformation, you need to evaluate and record details of structure and type on multiple generations of dogs. The best system I have come across for doing this is Dr. Carmen Battaglia's "Stick Dog" concept: Make a 3-generation pedigree of stick-figure dogs – head, ears, neck, body, four legs, and, where applicable, a tail. Make a color coded grading system for excellent, good, fair, and poor. Color each part of the dog to reflect the level of quality. If there is something you don't know, leave that section uncolored until such time as you are

able to check it out. You can make notes about details adjacent to the stick-figures.

If your area of competition is a performance event, you can adapt the stick dog concept to feature color coded symbols for important aspects of performance. For example, if you compete in stockdog trials the outrun (the way the dog leaves the handler and approaches the livestock) is one important aspect of each run. You could use the letters OR, written in or highlighted by the appropriate color of ink. In this case you would not want to base your rating on a single performance. The color coding should reflect the most consistent level of quality for that particular aspect of the overall performance. Again, notes could be added to provide detail.

If your breeding program is aimed at producing dogs for real world function, which could range from family pets to highly trained working animals, the scheme could be tweaked to reflect the key aspects of behavior and physicality pertinent to your particular area of endeavor.

No matter what your major goals for your dogs, temperament, disposition and health should not be neglected. Temperament and disposition are often used interchangeably, but they are different aspects of behavior. The late Vicki Herne told a story that illustrates the difference. Once, while riding the New York subway, she temporarily commandeered an elderly lady's ill-tempered Yorkshire Terrier to back down a couple of thugs who were harassing the passengers. The dog bit her thumb before he realized his job was to serve as a hand weapon, at which he acquitted himself admirably: The thugs exited the car at their first opportunity. As Vicki said, this dog had a lousy disposition but an excellent working temperament. Notes for this Yorkie might include words like "snappish," "active dislike of strangers," and "fearless."

Health tracking can be complex. Make a list of your breed's most common and most serious heath issues. Listings of common health concerns can often be found on breed club or breed health organization websites. Some issues with severe health impact may not occur frequently in your breed, but should be noted because you wouldn't want to double up on genes for that type of trait. Gathering family history is key here. Dogs that have a disease often don't appear in pedigrees (or their status may have been kept under wraps.) Make note

of near relatives of affected dogs as those are far more likely to appear on a pedigree.

#### **Getting Your Ducks In a Row**

The best collection of information in the world is useless if you don't have it stored and organized in a manner that allows you to retrieve it at need. Physical media (paper documents, photos, DVDs, tapes, etc.) should be kept together and organized in files, boxes or on shelves, depending on the media. The storage place should protect the items from incidental damage (weather, rodents, dogs, etc.) Digital records can be kept on your computer's hard drive, but it is always wise to have a back-up on a flash drive or other storage medium that can be kept physically separate from the computer in case of theft, fire or other disaster.

Some information you may not need to store yourself, at all. For example: All OFA open results are available on the organization's website and can be accessed at need. It may be easier for you to simply check their website than transcribe the information into your own storage system.

Storage is only half the battle. You have to keep it organized. Maintain individual files on each of your dogs. Information on other dogs might be organized in a variety of different ways: By breeder, area of competition, subject or some combination of these. I've found that the easiest way to put data where I can quickly find it is through the use of a pedigree program with a "notes" feature. I jot important things about a dog within it's record in the pedigree database. If I need to know something about that dog. either for itself or because I'm interested in a relative, I pull up the record and see what's there. You may not be able to store large volumes of information this way, but a simple note - "MDR1 certificate on file" along with the test result - tells me what I am most likely to want and directs me to a paper file if I need something more.

Finally, no database is ever complete. New things happen every day. You need to continue your data gathering as long as you are breeding dogs. Try not to let the filing or data entry pile up too high. It will make things easier to find when you want them and keep you from becoming overwhelmed by piles of disorganized paper.

#### Putting it all to work

Once you have your database set up, you can utilize it as a research tool and as an

aid to decision-making. In time, it can also help you review your past breedings to check your progress.

Pedigree analysis is the process of reviewing a pedigree to determine what traits – good or bad – you are apt to get in a given mating. The more you know about the dogs in the pedigree (hence the need for your database) the better your educated guesses will be.

For desirable traits, refer to your "stick dog" or equivalent pedigrees. Once you've completed this process you can tell at a glance what the strengths and weaknesses are in a given pedigree. Your goal, over time, is to have more and more good and excellent colors across the pedigree. Comparing "stick-dog" pedigrees of prospective mates indicates where they complement each other and what faults you might see in the litter.

"Stick dog" is best for analyzing the traits you want to see. But what about the undesired ones? Since full information on serious faults of conformation or behavior and health issues is virtually never available and because many of these traits are influenced by multiple genes, it is important to consider breadth of pedigree. Find a method of noting not only dogs that had the traits, but their parents and grandparents, as well. That way you can more readily connect the trait to a given pedigree and take it into account before you breed. Not every connection to an unwanted trait is equal. Having an affected dog in the pedigree is a greater problem than having its parent and a grandparent would be an even smaller issue. What generation the dog appears in is also an issue. The parent of a dog will be passing half of its genes to its offspring, but a much smaller number will come down from a great, great grandsire unless he is in the pedigree several times. Finally, the number of dogs in a pedigree that connect to an unwanted trait is important. A pedigree with a dozen connections is apt to be more risky than a pedigree with only one or two. Find a consistent way of scoring a pedigree that accounts for how many dogs with connections to the trait appear in the pedigree, where they appear and what

degree of relationship those dogs have to an affected dog. An example of this type of system is explained in the article "Reflections on Pedigree Analysis" (www.ashgi.org/home-page/genetics-info/pedigree-analysis/reflections-on-pedigree-analysis)

Not all traits are equal, so you need to setting priorities. What are the traits that are most important to your breeding goals? Which are less so? Health issues that can potentially impact the dog's soundness or quality of life are a greater issue than something that is readily treated. Keep in mind that your priority list for the litter you plan to have this spring may be somewhat different than the list you will make for the litter that will come from one of these puppies a few years down the line. Your data base will help you track how well you are meeting your priorities and whether something needs to be moved up or down the list.

#### Pay it forward

Once you have gathered a good amount of data, make concrete plans for what will happen to it when you retire or when you are gone. Do this even if you are relatively young. Stuff happens. What you have learned can help the upcoming generations of breeders, but all your effort and hard work can be lost forever if you don't get around to making provision for where it should go when the time comes.

It's said that most people are in dogs for about five years before they move on to something else. Those are not the people who have a lasting positive impact on their breed. A breeder needs to be in for the long haul if she wants to make any real contribution, so planning for both the short and long term is vital. You can't do this unless you have a supreme grasp of the qualities and drawbacks of a significant number of dogs. The more you know the better. A good, accessible database is key to putting it to use. While quality dogs will form the foundation of your breeding program, information forms the bricks out of which you build the structure and be part of the legacy you leave for those who come after.

# Side Effects Selection factors you may not have considered

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The success or failure of any breeder's efforts hinges on the selection criteria she uses. A good breeder pays close attention to physical and behavioral traits important to her goals. These primary selection criteria are regular topics of discussion, not only between owners of the prospective sire and dam, but at shows, trials and almost anywhere breeders get together.

But there a number of factors surrounding individual breedings—and the practice of breeding in general—which can cause side effects to your breeding plan, for good or for ill. These factors may seem peripheral. They may even be things you don't think much about at all.

There are a variety of decisions a breeder makes that aren't related to the qualities of the dogs involved. Even so, these decisions can have a very real influence on which stud you select for your bitch. Cumulatively, they can impact the genetic future of your breed. You might not think of these things have an influence because they have more to do with business, economics and marketing than with dogs.

### Secondary selection criteria

Geography matters, though perhaps not as much as it did a few decades ago. Today, we can ship semen to the bitch, eliminating the risk and expense involved with sending her great distances and into an unfamiliar environment. Even so, there are still costs involved and sometimes that will tip the scales in favor of a more local stud. You are also more likely to be familiar with a dog in your area, as well as his family and his get, leading you to choose the local boy.

Personalities can enter into the mix. All of us know people we really like and a few we really don't. It's human nature to want to do business with someone we like, or even someone for whom our feelings are neutral, rather than one who makes us uncomfortable. While this may sometimes have a direct bearing on the dogs involved – as in the case of someone known to be particularly honest (or not!) in his approach to health issues – sometimes the desire or reluctance to deal with

a particular person boils down to whether or not the two of you get along.

A dog with a winning show or performance career or one bearing a major kennel name can be a useful marketing tool for your litter. Accomplished dogs and successful breeders can have enormous positive impact on a breed. But a kennel name, no matter how revered, is only as good as the dog who carries it. Not every puppy produced, even by the most esteemed kennel, will be of breeding quality. Even a top winner might require second thoughts if he is siring large numbers of litters; his reproductive success can limit your future breeding options.

The devil, as they say, is in the details. Before any mating can take place, owners of stud and bitch need to come to an agreement on the terms and conditions relative to the breeding. A high stud fee may be too much for your budget, closing the door on a potential litter before serious discussions ever take place. The stud owner is the one who presents a contract to the owner of the bitch. If you, as the bitch owner, want to change something, the stud owner is free to agree to an amendment or not. If the two of you can't come to terms, the breeding won't take place.

#### Environmental impact

Breeding decisions don't take place in a vacuum containing only the people and dogs directly concerned. All of us operate in a wider environment which can shape our decisions as breeders. What is or is not acceptable in dogs is colored by our human cultures, whether of the country in which we live or the smaller "dog culture" in which we operate.

Geography also has its role here. The nature of the place you live cannot help but influence breeding decisions. Someone who operates in an intensely urban area may have a different idea of the suitability of one dog over another than someone who lives in the middle of 10,000 acres. What works in cultivated farmland may not work in near-wilderness. Climate and topography impact the way we—and our dogs—live.

Locale aside, we in dogs have our own varied cultures. What is the primary purpose for which you breed? It may be for the conformation ring, any of various competitive events, real world work, or for family companions. Each area of human/canine activity has different requirements. Our views of what is or isn't acceptable in a dog will be colored by the dictates of that particular discipline and the sub-culture in which it operates.

Our registries and clubs also influence a breeder's choices not only through their specific rules and regulations, but by the nature of each organization's unique "corporate culture." National kennel clubs wield enormous influence on canine activities within their respective countries, impacting not only those breeds which they register, but often the practices of groups or organizations of breeders and dog enthusiasts that operate out from under the kennel club umbrella. A national breed club will have a similar influence on its breed; if it is one of the few that also operate a registry, the influence can be even greater. Even regional clubs can have an impact, as the members of these groups often mentor newcomers, shaping their attitudes and helping develop their goals.

Another environmental factor is kennel management – not only on the gross scale of good or bad, but even in such differences as the size and nature of your facilities and how many dogs are typically resident. Effects on breeding decisions will be subtle, but you are most likely to keep an animal for breeding that suits your particular management style.

Some management decisions may, in excess, have a negative impact on the long-term viability of the breeder's line, or even the entire breed. For instance, the use of artificial insemination (AI) and c-sections have relatively common. AI has only been in general use by dog breeders for a handful of canine generations. The technology has advanced dramatically, enabling a breeder on one side of the world to import frozen semen from a dog on the other. Likewise, surgical techniques have improved so that c-section, while still a major surgery, is highly successful in the vast majority of cases. But overuse of these technologies can have a down side.

The classic example is seen in those breeds, like the bulldogs, where c-section has become routine for delivery. Breeders don't select for bitches that can free whelp because

surgical delivery has come to be considered "normal."

Using AI won't affect an individual animal's ability to breed naturally, but the convenience it offers may distract us from consideration of a dog's breeding behavior. A stud with little or no libido might make his contribution with collected semen. Likewise, a bitch that refuses to accept any stud can be impregnated at no risk to the dog.

However, nothing is more basic to biology than reproduction. No species can exist without it. Before resorting to artificial or assisted breeding practices you need to be sure both dog and bitch can get the job done Nature's way.

#### And the winner is...

Big wins and competitive titles are something to brag about, but how valid are they as guides to selection? No dog becomes a major competitor in any venue without a lot of time, effort and training on the part of its handler and others who support its career. The environment provided is "nurture" to the dog's genetic "nature." A great show dog may have the genes to endow his puppies with a perfect coat, but it is up to those who feed and groom to develop that potential. Those human talents can also help a less-than-ideal coat pass muster. Similarly, in performance events a dog's innate talent must be developed and guided by trainers and handlers. A genetically excellent dog with poor training or management can lose to a less talented one with the benefit of partnership with skilled people. Putting too much emphasis on wins or titles may not get you where you want to go with your breeding program - environmental factors (the training, handling, etc.) aren't coded in the dog's DNA.

### Working environment

Training and handling aside, performance event titles may or may not indicate a strong genetic predisposition for the behavioral traits typical of a breed's original purpose. How true-to-life is your chosen competitive arena? The more true the better it serves as a test of the innate behaviors required to fulfill the breed's purpose. The Border Collie's open field sheepdog trials are very like the actual work the dogs were first bred to do.

In contrast, in a Schutzhund blind search the dog needs to find the guy with the stick and the padded sleeve, just as a police dog needs to find the bad guy. Any well-trained

Schutzhund dog is smart enough to know that the guy is always behind the 6<sup>th</sup> blind but it's his job to search the other five first. On the other hand, police dogs, whose daily task is the real work upon which Schutzhund and similar sports are based, must think for itself and ignore the handler's commands if it knows the handler is wrong. Real bad guys are often armed with more than a stick and won't hold fire while the dog searches "empty blinds."

Alaskan Huskies are the breed of dog that dominates competitive sledding events. They are bred solely for their racing ability and are not restricted to a closed breeding pool; breeders occasionally employ crossbreeding to improve performance. This practice was typical in purpose-bred dogs before the era of studbooks and kennel clubs. A genetic study of Alaskan Husky performance in both short- and long-distance races published last July revealed that researchers could distinguish sprint dogs from distance runners by their genetic profiles. Admixture of genes from dogs of the hunting group could be found in successful sprint teams while dogs whose profiles revealed arctic heritage were better at the endurance events.

#### Without a thought...

Sometimes we wind up making breeding decisions without any conscious consideration of what we are doing. A number of years back a study of market hogs revealed that hog farmers who raised large numbers of animals inside barns and their colleagues who pasture-raised their stock inadvertently selected for different dispositions. All hogs in the study were of the same breed, but the barn raised animals were much less aggressive. Since excess aggression can be a management problem in a high-density environment, those farmers tended to keep less aggressive animals for breeding simply because they were easier to manage. Over time and generations this had a measurable effect on the animals' disposition as opposed to that of their more traditionally raised cousins.

It is very probable that dog breeders do the same thing. A dog whose behavior does not suit your particular circumstance isn't likely to remain in your home or kennel. Similarly, in a wide variety of breeds the divergence we see, not only in behavior but in body morphology, between working/field and show strains is in part influenced by the differing circumstances surrounding each set of breeders' approach to dogs and dog breeding. For example, Australian Shepherds were once all ranch dogs

whose ability to guard the home, ranch buildings and their owners' pick-up trucks was as highly valued as their utility in managing livestock. Today, most Aussies live in urban or suburban environments where a sharp-tempered guardian can be a liability – in the legal sense as well as in general. This reality has shaped a shift in temperament in a large part of the breed, though the original character can still be found in stockdog lines.

An unconsciously selected benefit working/field strains may enjoy is better overall health. Dogs which cannot withstand a highly active physical regimen wash out early and are never considered for breeding, not because the breeders are necessarily health-conscious but because meeting their selection goals requires that their dogs remain in superb physical condition.

The divergence in show and working lines has also contributed to the loss of the full suite of working behaviors in a variety of breeds. This is not because show breeders deliberately select away from those behaviors, but because most are not making a conscious and consistent effort to maintain them. Complex traits cannot be maintained without consistent selection, generation after generation.

A final example of "thoughtless" selection factors can be found in how some breeds dogs look. Considerable research has indicated that humans, and especially females, are attracted to babyish features in animals. Human nature, and perhaps the fact that the preponderance of people involved in dog breeding are female, has probably lead to the reduced size, larger eyes and shorter, blockier muzzles seen in several breeds today that did not exhibit these features a few decades past.

#### Ties that bind

Some traits go hand-in-hand, either because they are genetically linked, arising from genes that close together on the same chromosome, or biologically linked, with changes in one thing impacting the other. Not a great deal is known yet about genetic linkages for important breed traits, but examples of biological linkage between desired breed traits and undesirable things abound.

Many breed standards state that the dogs should have dark brown eyes, but in some of those breeds acceptable coat colors include brown (variously described as liver, chocolate or red) or dilute (as in blue or fawn Dobermans or any Weimeraner.) However, neither brown nor

dilute dogs can have dark brown eyes. The pigment diluting effects of the genes involved (commonly referred to as "B" and "D" by dog breeders) also dilutes pigment in the eye, resulting in eyes that are light brown to amber in shade.

Recent study of the gene responsible for very small size in many breeds revealed that small dogs are genetically predisposed to excitability. So, if you want a small dog but don't like dogs that are yappy or hyper, you may not be able to find what you are looking for, at least in those breeds whose smallness stems from that particular gene.

Selecting for body structures that vary considerably from the canine norm can also have unintended side effects. Some of these are well recognized and you can take steps to avoid them. Others you may not even be aware of. A recent study of short-faced breeds like

Pugs and Boston terriers found that, because of their extremely short muzzles, the skull has altered to the point that their brains have rotated forward. As a consequence, the olfactory lobe, which process the dog's ability to scent, has migrated to the lowest portion of their brains, probably to remain in reasonable proximity to the nose. At this time any effects this might have on these dogs' behavior or sense to smell haven't been determined.

While all of these secondary, environmental, and unconscious selection factors may not have a huge impact on any individual breeding, cumulatively they do influence the population genetics of a breed. By being aware of what these factors are and how they influence not only your decisions but those of other breeders you will be able to make more informed decisions for your own dogs.

### **Setting Driorities**

## **Bad Genes, Babies and Bathwater**

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Everyone has heard the phrase, "Don't throw out the baby with the bath water." But do dog breeders ever stop to consider how this admonition applies to them? Certainly not the novice who righteously declares that he will never, ever, keep anything that has even the possibility of producing the smallest genetic defect. Not the experienced breeder who refuses to consider an otherwise excellent line because it sometimes throws cataracts. And most definitely not the individual who declares that all DNA-tested dogs found to be carriers of recessive disease mutations ought to be removed from breeding. This tendency toward genetic over-kill not only culls dogs that might have something to offer, it can exacerbate the very problems breeders are trying to avoid. The following is a real life example of what can happen when breeders exercise short-sighted culling in the name of genetic disease control.

In the early 1970s, breeders of Basenjis launched a campaign to wipe out a fatal genetic disease called pyruvate kinase deficient hemolytic anemia (HA). HA is caused by a recessive gene. Dogs with a single copy of the gene are healthy, but those with two copies die. A screening test was developed that would indicate carriers as well as affected animals. Breeders zealously screened their dogs, eliminating not only affected animals but the healthy carriers from the breeding population.

Today HA is rare in Basenjis, but the incidence of Progressive Retinal Atrophy is significantly higher. As is yet another fatal disorder, a kidney problem called Fanconi's Disease. At the time, neither of these diseases had a screening test that would indicate carriers. (A DNA test for Fanconi is now available.) Had breeders been less fanatical in their pursuit of HA, they might have retained the healthy

carriers in the breeding population, breeding them only to non-carriers so they could avoid producing HA- affected puppies. By such a method they could have retained the good aspects of those carriers, including freedom from genes for PRA or Fanconi's, while gradually lowering the incidence of the HA gene. Now that a Fanconi test is available, they can use this approach for that disease.

Fortunately for the Basenji, there is still a native population of the breed in Africa. The Basenji club prevailed upon the AKC to allow them to re-open the stud book to admit some African-born Basenjis. This badly needed source of new genetic material comes at great trouble and expense for those breeders who make the effort acquire one of these imports. This option isn't even possible in some breeds, and even where it is, convincing a large registry like AKC to accept undocumented foreign imports is itself a daunting task.

In spite of what happened with the Basenji, this should not be viewed as an indictment of screening tests. The problem wasn't the HA test, but the drastic culling process that breeders undertook when using it. If there is a test which can identify carriers, make use of it. This is especially true of DNA tests which not only reveal the dog's genotype, they are not subject to the false positive or negative results of other types of testing. Breeders need to know as much as possible about the genetic potential of their breeding stock. Ideally, they should be willing to share the results, whether good or bad, with other breeders.

Knowledgeable dog people know there is no perfect dog. Even the best of them have faults. The faults are not only those

conformation or behavioral problems you can readily observe, but also bad genes. Dogs have around 25-30,000 genes. No matter how high the standards for selection of breeding stock or how strict the culling of offspring, every dog will have genes for unwanted traits. Experts agree that every individual--dog, human or cauliflower--probably carries, a few "lethal equivalents" as well as a batch of genes that are merely suboptimal. This may leave you wondering why we aren't seeing dogs and cauliflowers, not to mention each other, dropping like flies all around us.

Under normal circumstances, lethal genes remain rare. Natural populations breed randomly, maintaining a varied mix of alleles, or forms, of genes. Only occasionally will the right combination of bad alleles match up to produce an affected individual. In addition, the lethal nature of these diseases limits the ability of affected animals to pass them on to their offspring because affected individuals often don't live long enough to reproduce. But the breeding of purebred livestock, including dogs. is not natural or random. It is selective based on the wants and needs of breeders. As a result. the number of lethal equivalents in most breeds exceeds the average of three, the problem genes having been inadvertently concentrated through the standard inbreeding practices used to maximize production of desired traits. Two examples in Australian Shepherds are Pelger-Huet Anomaly and merle. Genes with lethal effects are only the tip of the iceberg. There unknown numbers of those suboptimal genes whose effects are anywhere from minor to extremely bad.

Breeders routinely evaluate breeding stock by studying conformation and/or performance attributes in minute detail. Virtues are weighed against faults and compared to the virtues and faults of prospective mates. If the overall analysis is positive, the breeder will proceed. Hereditary diseases and defects need to be given the same kind of consideration, in and of themselves and in combination with all the dog's other traits.

Some faults are severe enough to eliminate a dog from breeding consideration entirely, but even genetic defects and disease may not necessarily fall into this category in some circumstances. Remember the case of the Basenjis and HA. Dogs proven to be

carriers of traits in which only homozygotes (those with two copies of the gene) are affected, can be used if care is taken never to mate one carrier to another and not to use them extensively. If a DNA test is available, preference can be given to the clear-tested offspring of carriers for the next generation. In time the number of carriers will be reduced.

If the mode of inheritance for a trait is unknown or complex, identifying carriers can be difficult. Individuals that repeatedly produce traits like hip dysplasia, Cushing's Disease, or severe allergies should be pulled from further breeding because of the serious and debilitating nature of those diseases. But their healthy relatives may be used if care is taken to select mates unlikely to carry the same defect. If at any point an individual proved to be a repeat producer of the defect, it could then be removed from the breeding program.

Many faults are variable in expression. This includes such genetic defects as hip dysplasia (HD) and missing teeth. In Clumber Spaniels, where HD was once almost universal, elimination of all affected animals was not an option if the breed was to be preserved. By selecting away from the most severely affected dogs, Clumber breeders have managed to improve their overall situation, producing more non-dysplastic dogs and fewer which are severely affected, even though HD is still common. A similar situation has occurred with Collies and Collie Eye Anomaly.

In the case of missing teeth, a fault common to show line Australian Shepherds, something similar could be done. There are sufficient quality dogs available with full dentition that dogs missing multiple teeth ought not to be bred. However, those missing one or two teeth could be bred to mates with full dentition which are out of families with full dentition. In the 1970s, missing teeth in Aussies were almost unheard of. Twenty years from now the situation could be to nearly its starting point if breeders would be conscientious about screening and mate selection--and none of he good traits those dogs have need be lost along the way.

The overall size of a breeding population must be taken into account before making final decisions on whether a dog exhibiting or carrying a defect ought to be bred.

Australian Shepherds are numerous, but certain sub-sets of the breed are not. In North America there are thousands of Aussies, but in some parts of the world national populations may number only a few hundred breeding animals at best. Opportunities to add new stock can be limited by the expense of importing, strict quarantine laws, or import restrictions. Even in North America a breeder's selection of potential mates may be limited if his breeding goals are very specific, such as producing a particular type of stock dog.

In small populations, breeders may have no choice but to use some defective animals. The only alternative is to resort to increased inbreeding which will narrow the available gene pool even further and bring other, possibly worse, defects to the fore. If defective dogs are to be used, breeders should take special care to avoid subsequent inbreeding on those dogs. Neither should such a dog be bred extensively. Among its offspring, only those which do not exhibit the defective trait should be considered for further breeding.

If breeders approach genetic disease with an objective eye and if they are honest with themselves and each other about the potential for producing genetic diseases and defects in any given cross, they can obtain healthy babies while the bath water full of bad genes drains slowly away.

## Flavor of the Month Avoiding the fickle finger of show ring fashion

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"The Australian Shepherd is...of **medium** size and bone...**Slightly** Longer than tall...coat of **moderate** length...the topskull length...**equal** to the muzzle...Lips are **close fitting**...The ears lift...one quarter (1/4) to one-half (1/2)...break forward **or** slightly to the side..."

Breed Standard, Australian Shepherd Club of America

"The Australian shepherd is...**slightly** longer than tall, of **medium** size and bone, has a coat of **moderate** length...Head is...**dry**...muzzle is equal in length or slightly shorter than the back skull...The Ears...break forward...or to the side..."

Breed Standard, American Kennel Club

[Emphasis above the author's.]

"Long and low!"
"Gobs of bone!"
"Dripping with coat!"

(names withheld to protect the guilty)

Breed standards are the template breeders use to guide their selection of physical traits. Ideally, the dog as described should have the physical attributes necessary to perform whatever its original task might have been. Ideally, the big winners in conformation events should be stellar exemplars of their breed standards. Ideally.

We humans find the novel irresistible. We are fascinated by what's new or different. We crave what's "hot." This is true not only of clothing trends and electronic gizmos, but also our dogs. There are breeds or types of dogs that have been "in" because something – frequently a popular movie or television program – presents them to the public at large in an appealing manner. While we who are "in dogs" generally cringe when our favorite breed shows

up in a major entertainment vehicle, we are no less prone to flights of doggy fashion than the average person sitting in a theater or on the couch.

Media-driven breed popularity surges are largely beyond our control. Despite the best efforts of clubs, rescue groups, and concerned fanciers, there are always plenty of unscrupulous commercial breeders, large and small, who are willing to produce volumes of "product" to meet consumer demand leaving us to pick up the pieces. But we also have our own fashion cycles that we need to beware.

#### Subject and History

I'm using my own breed, the Australian Shepherd, to illustrate my thesis not because it is the most blatant example (it's not) but because it is the breed I know best. Look at your own breed – its standard, what's winning today, and what won in times past – and you can probably find your own examples.

The Australian Shepherd is a recently developed breed. First studbook registration was in 1957. The dogs derived largely from indigenous ranch dogs of the American West in the early to mid-20<sup>th</sup> century. These dogs frequently exhibited blue merle coloration and bobbed tails. The smooth-faced variety of the Pyrenean Shepherd and herding dogs of unspecified breed but of generic collie type from Australia have been documented as contributing to the breed. The breed has about 300 founders - far more than many breeds - and for most of these dogs there is no known background, they were simply useful ranch dogs with duties including guarding and varmint control as well as moving livestock.

Type was highly variable in the mid-20<sup>th</sup> century as to size, coat, color and earsets, but the majority would conform with modern breed standards. The Australian Shepherd Club of America (ASCA) standard developed in the mid-1970s is the ancestor of all standards in use today. The guotes above from the current ASCA and AKC standards conform very closely to that original document, with one exception which will be discussed later. The development of that ancestral standard and the booming popularity of a show bloodline that closely met that standard lead to a more uniform appearance across the breed. Even breeders of working and trial dogs, whose main concern was performance traits, were less prone to breed those dogs that varied most widely from the standard.

#### Setting an Example

Fashions in appearance tend not to plague working and performance dog breeders. Their focus is elsewhere. The conformation ring, however, puts primary focus on how the dog looks. Movement is important, but the dog must first look right; a dog that moved like an Aussie but looked like an oversized Sheltie would not (and should not) win in any show with competition. The nature of any competition is to identify the individuals that are better than others. And once you've found those, the next batch need to be "more better."

Since appearance is key, there is a tendency to exaggerate any trait that is considered desirable: Size, coat, bone, muzzles, ears, even movement. Biggest, smallest, longest, shortest, highest, lowest, densest, curliest, or whatever - any "-est" can become the focus of the siren song of show ring fashion. As a result breed appearance can change. Look at photographs of the top dogs in your breed of the 19th century, the 1920s or 30s. and today. In most breeds you will see marked differences between those early "greats" and current quality dogs. My breed doesn't' have such a long history, but even in the Aussie's short span you can see a difference. The first dog to win an ASCA National Specialty was a blue-merle male named Wildhagen's Dutchman of Flintridge. He was used by Dog World Magazine as their exemplar of the Australian Shepherd standard in the 1970s. He still conforms well with the standard but the breed fashion has changed and he would no longer outdistance his competition as he did in his day.

Most Aussies, particularly those that are shown in conformation, meet the standards. However, fashion trends do occur and some fly in the face of what the breed standards describe. A current favorite in some circles is "long and low." A dog that is distinctly longer than tall due to a somewhat shorter leg than is proper. The standards are very clear in stating the Aussie should be *slightly* longer than tall. The look has been around for at least twenty years but of late it has gained popularity even though it does not meet the standard.

With very few exceptions, dog breeds developed to move groups of livestock in more-or-less open terrain are slightly longer than tall and have sufficient leg to move quickly and efficiently over distances, across rough ground, and possess the physical dexterity for maneuvering around potentially dangerous stock. For this reason the ASCA standard

states: "The point of the elbow is set under the withers and is equidistant from the withers to the around."

Fashions arise because some dog or line of dogs with a particular look does a lot of winning, or wins a major event. Everybody else wants to win, too, so they try to breed dogs that look like the big winner, generally by breeding to it or its relatives.

#### **Smoke and Mirrors**

Fashion sometimes can lead to skullduggery in order to meet it. The taping and gluing of ears to achieve a particular set is clearly artificial but not sufficiently so to spur action by those who administer the rules. There are unscrupulous individuals, however, who will resort to surgery if less draconian measures don't work. This is blatantly against the rules though the perpetrators generally won't get caught.

Australian Shepherds are supposed to have moderate sized ears that break forward or to the side (rose ear.) The ASCA standard specifies that the break should be a quarter to a half along the length from the base. The AKC standard only notes that they break. Very high-breaking ears are not considered desirable so aren't an issue. However, the rose ear – totally acceptable under both standards – is also generally disliked, hence the taping, gluing, and occasional skullduggery.

The problem with all this dedicated effort to get what is considered the prettier earset is that if significant numbers of people are altering whatever Nature gave the dog, it becomes impossible to breed for the preferred set.

#### Window dressing

Some might ask whether it really matters if people's preferences change the look of a breed. It's all cosmetic, and if someone likes one look over another, why shouldn't she breed for it? Obviously, there is nothing stopping people from doing just that if they want to because it has been going on for a long time; probably from the time people first started showing dogs. But every breed has its own unique history and traditions, and the physical appearance of the dogs is part of that tradition. People are attracted to a breed for what it is, not what they can turn it into. In theory, anyway. But some changes have more than cosmetic

impact on the dog and on its owner. Sometimes they may even impact health and soundness.

In years past the Australian Shepherd was not particularly prone to over- or undershot bites. They happened, but not with great frequency. The Aussie's muzzle is traditionally tapered and approximately the length of the back skull. This allowed for the full set of teeth in classic canine orientation necessary in any function-bred dog that uses its mouth in the course of its business. A number of years back a fashion for shorter, blocker muzzles arose. The AKC breed standard reflects this new preference, stating that the "muzzle is equal to or slightly shorter than the back skull." While this muzzle type isn't by any means universal, it's still frequent enough that someone unfamiliar with the breed historically might think it normal. Because a relative few generations of dogs have been selected for this look this fashion trend may have impacted the frequency of bad bites in the breed. The Australians Shepherd Health & Genetics Institute's breed health survey, completed in 2010, found that 3% of the dogs were reported as having bad bites.

In the worst case, the pursuit of fashion can result in significant health and other issues for the dog. Your breed may find itself painted into a corner that is difficult to impossible to get out of. If you doubt me, locate photographs of Bulldogs from the mid-19<sup>th</sup> century then consider the Bulldog today.

#### The conservative approach

Another problem with fashions is that they can change. What's hot today may not be a few years down the line. The wise breeder will choose a conservative path, studying the standard and selecting for traits that don't push the boundaries. Where variety is allowed, as with earsets in Aussies, she will breed for what she wants and keep the dogs that fit her criteria rather than cosmetically improving those that don't.

Breed standards exist to guide breeders and judges in selecting the proper sort of dog for the breed. When not overly revised, they help transmit the vision of the breed's early supporters, who knew the breed as it should be, down through generations of breeders. The task of their heirs, the breeders and exhibitors of today, is to preserve and continue those traditions.

## **Making Genetics Work For You**

## In the Mode How traits pass in dogs, lines and breeds

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What dog breeders do is not breeding dogs; normal, healthy dogs can do that without any assistance from us. Breeders manipulate genes, encouraging some to pass on from generation to generation while at the same time trying to prevent others from doing so. With somewhere around 25-30 thousand canine genes to work with and, for most of them, no way to know for sure exactly which versions, called alleles, a particular dog carries we are not doing much more than rolling dice unless we develop a thorough understanding of modes of inheritance: How genes flow from a dog to it's offspring, as well as down through generations of a line or breed.

Single-gene modes of inheritance

Inheritance from parent to offspring is the most basic and easiest to understand form of gene transmission. Every dog has two copies of each autosomal gene. (Autosomal genes are those that are not on the sex chromosomes.) One of these copies came from its father and the other from its mother. What combination of alleles it has is its genotype. How the alleles interact with each other, other genes and the environment will determine what traits you will see in the dog, referred to as phenotype.

The most basic mode of inheritance is simple dominance. Black vs. liver color is a classic example. The allele for black is dominant; the allele for liver is recessive. If a dog has at least one copy of the dominant black allele, it will be black. For a dog to be liver, a color produced by the recessive allele, it must have two copies. A black dog might produce liver puppies if it carried a recessive allele, but a liver dog cannot produce black puppies unless bred to a black.

You cannot tell from appearance whether a dog exhibiting a dominant phenotype like black is also carrying a recessive allele. However, knowing the phenotypes of dogs in its pedigree can give you an indication of whether it might carry the recessive. If a black dog has a liver parent you know that black dog is heterozygous, meaning it has two different alleles. Such a dog will produce liver if bred to another dog with at least one copy of the liver allele. It has a 50/50 chance of giving a liver allele to each of its pups.

When looking at pedigrees and thinking about autosomal dominant or recessive traits. the breeder should follow the pedigree back step by step along each path of ancestry and note where he first encounters a dog he knows was either one or two copies of the recessive allele. In most cases 4 or 5 generations will be sufficient. The closer up an ancestor with the recessive is, the more likely it will have been inherited. Dogs which exhibit the trait have two copies of the recessive allele and will always pass the trait but a carrier which has only one may or may not. If you don't know the genotype of the dominant phenotype individuals that lie between that ancestor and your dog, you can't know for sure if the recessive was passed along or not. The farther back the carrier is, the less likely the gene will have passed on.

By knowing how many and how far back are the ancestors that you know carried a recessive trait, it is possible to precisely calculate the probability that a dog has inherited the recessive allele. Even the math phobic can have a good idea of what could happen just by studying the pedigree. However there is one factor those who don't want to mess with math need to keep in mind. We tend to think of probability being halved with each generation:

Half the genes come from each parent, a quarter from each grandparent, an eighth from each great-grandparent, and so on. This often leads people to the erroneous conclusion that the offspring of two carriers of a recessive trait all have a 50-50 chance of carrying the recessive allele. This is not the case.

Matings of carriers can produce four allele combinations: Homozygous (two copies) dominant, homozygous recessive, paternally inherited dominant heterozygote (one copy) and maternally inherited dominant heterozygote. Three quarters of these are phenotypically dominant. In our black/liver example, that would be three black puppies for every liver. Among the black puppies, two out of three will be carrying the liver allele. Therefore, the odds for carrying liver in any black pup out of such a cross are not 50/50 but 2 out of 3.

If a recessive trait is something you want, you can use this process to determine how likely you are to be able to produce it in a litter. You can increase the likelihood that it will happen through your mating choices. Conversely, if you do not want to produce the trait you can eliminate the risk of producing it by breeding known or possible carriers to dogs you know are homozygous dominant.

Some genes have an incomplete dominant mode of inheritance. In this case each genotype will have a distinct phenotype, with the heterozygote being intermediate to the dominant and recessive phenotypes. The merle color pattern is an excellent example of this. Dogs with two recessive alleles are not merle, heterozygotes will be merle patterned, and those with two dominant alleles not only have merle patterning but frequently have considerable amounts of white markings and almost always have serious eye defects and deafness. Since the phenotype always indicates the genotype, the breeder will know what alleles the dog has by looking at it.

If alleles are co-dominant, their traits will both be expressed in the heterozygote. The genes in the Major Histocompatibility Complex, which governs important aspects of the immune system, are co-dominant. Both maternal and paternal alleles will be active. In the case of MHC genes, it means the dog has a bigger arsenal to protect itself against disease than a dog whose MHC genes are mostly homozygous.

Some genes have more than two alleles. Dominance between them may be simple, co-dominant or incomplete. The gene that produces golden/yellow coat color and black

masks has a clear dominance hierarchy among its alleles. The most dominant allele will not produce mask or yellow, so dogs that have one copy will have coat color determined by other genes. The most recessive allele, when homozygous, results in hair that is yellowish, as in Golden Retrievers and yellow Labradors. The middle allele in the series is for a mask. Any dog that has at least one copy of his allele and does not have a copy of the most dominant allele will have yellow hair with a darker mask on the face. The color of the mask will depend on what other color genes the dog has.

If the multiple alleles are incompletely dominant, heterozygotes will be intermediate in phenotype to the two alleles resulting in a continuum of phenotype expression with the phenotype of a specific individual dependent on which pair of alleles it had. (White markings were once thought to be due to this type of inheritance, but recent genomics research has found that there are at least two major and distinct white marking genes.)

Not all breeds will have all alleles possible for a particular gene. Knowing which ones your breed has can be important. For years Australian Shepherd breeders have been selecting against yellow, dilute, and sable because the colors are disqualifying though they did occur early in breed history. As a result sable is absent or nearly so because it is dominant to tan trim and most Aussies have tan trim. Both yellow and dilute (blue or Isabella) do occur but are rare.

There are two other proposed single-gene autosomal modes of inheritance:
Dominant with incomplete penetrance and dominant with variable expressivity. With the former, a dog can have the genotype but sometimes will not. In the latter, when or how trait presents can vary considerably. As more and more is learned about how genes interact with each other and the environment, as well as how specific genes are structured and function, it is apparent that neither of these modes of inheritance actually arises solely from a single gene.

Inheritance of genes on the sex chromosomes differs from that of autosomal genes because the sex chromosomes come in two different forms: X and Y. Female mammals have two X chromosomes while males have an X and a Y. The Y chromosome contains only a very few genes, all of them are related to specifically male traits. The X chromosome contains a normal number of genes that produce

a wide variety of traits not related to the sex of the individual. However, only one copy of the X can work in any given cell, so females are a "mosaic." Which X operates in each cell is randomly determined during development. This can be most clearly seen in calico cats, in which black and orange are phenotypes produced by different alleles of the same X chromosome gene. Whether a calico cat has a black patch or an orange one at some particular spot on her body will depend on which of her X chromosomes was turned off in an embryologic ancestor cell. The exact pattern will have no bearing on what she might produce beyond being an indicator that she has the potential to pass on both black and orange alleles.

If a particular X allele produces a disease, like hemophilia, it will occur most frequently in male offspring who have only one X. Females' mosaicism provides them with sufficient normal cells that they will be healthy. (In the unlikely case of a female homozygote for hemophilia, whose father would have to be a hemophiliac himself, she would in most cases die during development. If she did make it to birth she would hemorrhage to death no later then her first heat cycle.)

If a male has an X-linked disease, this is the only time (other than with traits that are clearly autosomal dominant) that a stud owner can truthfully and accurately insist that her stud was not responsible for the problem. These diseases are inherited from mother to son. Each daughter of such a mother has a 50/50 chance of herself being a carrier. The mother of a carrier is probably also a carrier and the health status of her sons should be examined. However, genes for hemophilia and some other X-linked diseases mutate with unusual frequency so one cannot assume that all the bitches on the direct female line were carriers.

Another form of inheritance in which sex plays a role is imprinting. With imprinted genes, the phenotype will be determined by which parent the gene was inherited from. This mode of inheritance is not common and all imprinted genes discovered thus far are involved with development or reproduction. There is also evidence that epigenetics, a form of gene regulation that can be influenced by environment, may play a role in these traits.

### Multiple genes

Unfortunately, most traits are not inherited in a simple, single-gene fashion. Many are polygenic, resulting from the action of

multiple genes. Often environment can influence these traits to some degree. At the present time, there is no way to know the genotype of any particular dog for any polygenic trait. The best the breeder can do is make an educated guess. Phenotypes in polygenic traits represent a continuum, rather than a series of similar but more or less distinct types. Canine hip dysplasia (HD) is a prime example. Dogs can have hip joint conformation that ranges from superior to abysmal. Two sound dogs can produce dysplastic offspring and dysplastics can produce sound pups.

With polygenic traits the parental contribution can be unequal. A parent with just one or a few genes that produce the trait may have offspring that exhibit it if mated to a dog that has all the rest. Or the trait may show up after many generations of absence because the right combination of genes finally happened to fall together. With polygenic traits a breeder must consider the history of the trait in the family, rather than in the pedigree. Dogs that have a family history of the HD (affected siblings, cousins, aunts/uncles or nephews/nieces) are more likely to produce HD than dogs which do not. The more affected relatives there are, the greater the risk.

This kind of family analysis can be useful for producing desired traits as well as avoiding those not wanted. For example, if a dog has an excellent front and comes from a family of excellent fronts, it is less likely to produce incorrect fronts than a similar quality dog that has unusually good front conformation for its family.

Sometimes genes that do not interact with each other produce traits that are nearly always found together. Such genes are linked, occurring close together on the same chromosome. Chromosomal near-neighbors are unlikely to become separated as the genes are shuffled prior to formation of sperm and eggs. If a breeder observes that she cannot find a dog that has a trait she likes without it also having some other thing that she does not like, it may be that the traits are linked. She may have to live with the one if she wants to have the other.

The genetics of the immune system are both polygenic and linked in an extreme degree. The Major Histocompatibility Complex (MHC) is a set of linked genes that inherited as a unit called a haplotype. The higher a dog's level of inbreeding and the more recently that inbreeding has occurred, the greater the probability that the MHC haplotypes will be the same or very

similar. This can result in an impaired immune system, autoimmune diseases, and reproductive problems. Risk of producing affected offspring is greatly reduced if the breeder makes an effort to produce heterozygous haplotypes by monitoring the degree of inbreeding through the use of coefficient of inbreeding (COI) calculations on proposed litters and opting for suitable mates that will produce lower COIs.

#### Environmental effects

Genes do not act in a vacuum. The environment a dog experiences in the womb and throughout its life impacts the action of its genes. Even things experienced by parents may have epigenetic effects on the offspring. Dogs are born with a certain genetic potential. Whether and how much that inheritance comes to fruition depends on where it and its parents live and what it experiences, both mentally and physically. The genetic contribution is often described as the "heritability" of a trait.

Heritability is a measure of how much phenotypic variation in a trait results from genes, rather than environmental effects. Heritability estimates for hip dysplasia vary by the type of exam used, ranging from 54-76% depending on the focus of the exam (PennHip's distraction index was 61% and OFA's extended hip joint radiograph was 76%)<sup>1</sup> meaning most of what you see in your dogs' hip joint conformation is the result of genes rather than diet or exercise, the two most important environmental factors. The higher the heritability, the more control the breeder has over the trait.

Some inherited traits, notably chronic autoimmune diseases, require an environmental trigger. The dog must have the genes before it will have the disease; however it is possible that a dog will never develop disease if it never encounters something that triggers the immune system to start attacking its own body. Such conditions are said to be genetically predisposed. As with traits of high heritability, the genes must be there in order to produce the trait, no matter what the environmental conditions.

#### Lines and Breeds

Understanding inheritance in individuals is only the first step a breeder needs to make. Each individual dog is part of a larger population from which its mates will be selected and of which its offspring will become a part. Not every breed will have every allele possible for each gene. MHC haplotypes are an example of

this. The genes tend to have far more alleles than do other types of genes. Pure breeds have fewer haplotypes than do mongrels because the breeds are a closed subset of the species. How few haplotypes a breed has depends on its history and how much it has been subject to the effects of popular sires and prominent kennels.

Selection criteria need to be sufficiently broad, encompassing not just physical attributes, but health, behavior and temperament. Strong selection for or against a particular trait or a few traits can skew a gene pool and inadvertently result in the lowered frequency or elimination of some alleles while at the same time increasing or "fixing" others. (An allele is fixed in a population if it is the only one present; color gene alleles causing solid black body color are fixed in the Schipperke.) Fixed genes may be good or bad, depending on what those alleles happen to do.

Breeding according to the current fashion via selecting for this year's winning "look," or the excessive use of a popular sire or the output of a prominent kennel can likewise skew a breed gene pool and result in unintended consequences. The smaller the breed population, the greater the effect narrow selection criteria and breeding for fashion will have.

A line is an extended family of dogs. It is developed by some degree of inbreeding and thus will necessarily lead to a sub-set of the alleles present in the breed. The composition of this sub-set can be altered through the same things that will alter allele frequency in the breed. Since a line is necessarily a smaller population the effects can be more drastic. Desired traits can be made fairly uniform in a relatively few generations, particularly those that are easily observed and not much influenced by environment. However, unwanted traits may become intractable features: Epilepsy became just such a problem in show line and some working line Australian Shepherds in little over a decade.

New (or lost) alleles can be brought into a line by the simple expedient of outcrossing. On a breed-wide scale, however, this can be difficult in our current system of closed registries. For breeds with wide geographic distribution, imports may provide sources of fresh genetic material provided the exporting country's registry is considered acceptable, the populations are not already substantially related, and the populations have not diverged in type to the point that breeders in each country consider

the dogs in the other unsuitable. In a few cases the American Kennel Club has allowed significant additions of fresh stock at the request of member clubs, most notably the admission of a few African tribal Basenjis and the acceptance, via the Field Dog Studbook, of some Salukis of recent desert origin from a small US registry. But for some breeds, there is nowhere to go without cross-breeding with another breed of similar type. This was done with several European breeds after they were pushed to the brink of extinction by one or both World Wars.

The breeder's task is to effectively utilize what is known about modes of inheritance for breed traits, both positive and negative, in order to produce quality dogs that not only meet his

competitive or performance goals but which are also physically and mentally healthy. He must at all times remember that he does not act in isolation. Whatever he does will have an impact on breeders that follow. The greater his success, the greater his impact for good or for ill will be.

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## Of Babies, Bathwater and DNA Tests The use and misuse of a new technology

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The old saying "don't throw out the baby with the bathwater" warns us not to lose track of what's important by overemphasizing a negative detail. Anyone who achieves lasting success in the dog game learns that it is the totality of an individual dog that must be considered. While there are specific faults and defects that are deal-killers for any responsible breeder, most need to be evaluated in the larger context of the breed, a breeding program, or the dog's collection of vaults and virtues. However, in recent years a technological advancement has sometimes made the bathwater so murky for some of us that we forget there is a baby in there somewhere.

### If some is good, more is better...

...is another oft-cited truism. This phrase might even be hardwired into the human brain. We are endlessly fascinated by extremes of all types which we often view as 'better" than the normal run of things. DNA screening tests are proving to be one of those things.

Until very recently, the only way we knew to prevent producing something unwanted was to avoid it. If a particular thing was very bad, avoidance might mean eliminating a whole group of related dogs from a breeding program or even an entire breed. Not every one of those

dogs would have the potential to produce the unwanted trait, but there was no way to tell who did and who didn't. The risk of breeding those individuals and possibly producing the bad thing wasn't worth it despite whatever good traits the dog might possess.

Since the completion of the canine genome in 2005, science has been able to pinpoint individual genes responsible for particular traits. When those traits are diseases, a DNA screening test is soon developed and made available to the public. These tests are of tremendous benefit: For the first time in dog breeding history, a breeder can know with absolute certainty what every one of her breeding dogs' genotype is for various inherited diseases, as well as a few physical traits like coat color.

Since dog breeds' genetic backgrounds differ, the diseases common in one will vary from those common in another. Therefore, each breed has its own set of tests. In Australian Shepherds, we commonly do DNA tests for MDR1, a drug reaction mutation; HSF4, a gene with mutations that cause cataracts, one of which causes most of the cataracts I Aussies; as well as Collie Eye Anomaly and the progressive rod-cone degeneration form of Progressive Retinal Atrophy. About a half dozen other tests

are offered for the breed, but the diseases are sufficiently rare that they are used only by those whose lines have those diseases in or because a related individual has been diagnosed.

All this testing is a good thing: With it we can prevent producing puppies that have those diseases. But sometimes our pursuit of best practices can lead to overkill. In a classic example of more is better, there are people in dogs who decided that mutations ought not to be tolerated at all, even when carrier dogs are healthy. This viewpoint appears to be especially prevalent in Europe, but there are breeders in North America who also subscribe to the philosophy.

#### Perfect is the enemy of good

Anyone who's been in dogs for any length of time will have encountered someone, usually very new to the game, who proudly declares that she would never, ever breed anything with any sort of fault. Experience soon teaches us, if we didn't know already, that there are no perfect dogs any more than there are perfect people or anything else. Living beings have flaws. It is the breeder's task to evaluate those flaws and decide how she will minimize their effect in her breeding program. In most cases this means, among other things, breeding away from any unwanted traits a dog may have. If the dog's faults are sufficiently numerous or especially bad it may not be bred at all.

DNA tests have provided yet another factor for breeders to consider. All of them will tell you what variants of a specific gene a dog has. If it is clear, so is the "bathwater" and nothing need be thrown out. But the presence of a mutation sends some people running to dump not only the water but the baby and the bathtub. too.

When DNA test results indicate the presence of one or even two copies of a mutation, the breeder must consider what the presence of that mutation actually means for the dog and her breeding program. Striving for perfection – in this case no mutation – is a lofty goal, but only if eradication of the mutation does not also cause major harm to a breeding program or, worse yet, the breed. This does not mean the breeder can simply shrug off the results and do whatever she wanted to do anyway. She must give serious consideration to test results, but within a wider context than the test result alone.

What's the point?

When it comes to health issues, the point is to produce healthy puppies. The removal of affected dogs from the breeding pool has long been and remains an important form of prevention; the affected dog necessarily has genes for whatever disease it has and will pass them to its offspring. DNA tests allow breeders to make use of healthy carriers with no risk of producing affected pups.

People tend to use the term "carrier" loosely and some testing labs use it incorrectly. The mode of inheritance for a particular mutation determines whether there are – or are not – carriers:

- Dominant even a single copy of the mutation will lead to disease, there are NO carriers with this type of inheritance.
- Recessive a dog must have two copies of the mutation to develop disease, those with only one are carriers and will remain healthy.
- ➤ Polygenic specific variants of multiple genes, which individually may be any mode of inheritance, are required for the dog to develop disease. The specific collection of gene variants a dog has will determine whether it is affected or not. (There are no DNA tests at present for this type of disease.)
- Incomplete penetrance the mutation may be dominant or recessive, but not every dog with the disease genotype will actually develop the disease, probably due to environmental factors or the actions of other genes. These genes are often said to confer a "risk factor."

### Caveat Foecunduque Canes

(Let the dog breeder beware.)

The presence of a mutation, in and of itself, is not a reason to eliminate a dog from breeding: *Every* dog has mutations. The only way to get rid of them all is to cease breeding dogs. If a mutation is common in a breed, excessive culling may narrow the breed's gene pool. Heavy-handed culling can also lead to problems far worse than the one being culled. The Basenji offers an example of how this can happen.

Before there were DNA tests breeders were occasionally lucky enough to have a blood test for a disease that revealed carriers. Such was the case with the lethal recessively inherited disease, pyruvate kinase hemolytic anemia (PKHA.)

In the 1980s, armed with the carrierrevealing blood test. Basenii breeders launched a campaign to wipe out PKHA. They zealously screened their dogs, eliminating not only affected animals but the healthy carriers from the breeding population. PKHA became extremely rare in Basenjis - a breed with an already tight gene pool - but formerly uncommon late-onset Progressive Retinal Atrophy (PRA) and Fanconi's Disease, a lethal kidney ailment, both became prevalent. Had breeders been less fanatic in their pursuit of PKHA, they might have avoided the increased frequency of those other diseases by keeping the healthy carriers in the breeding population by not breeding them to each other.

At present, we can test for only a tiny fraction of the disease-related mutations that exist in the canine genome. To make constructive use of these tests we need to make the production of disease-free puppies the goal rather than the total eradication of the causative mutations.

#### How-To Manual

If a mutation is a simple dominant and the disease has serious associated qualityof-life or financial impacts, removing every dog with at least one copy of the mutation makes sense.

In the case of recessive mutations, affected dogs – those with two copies of the mutation – should not be bred if there are serious quality-of-life or financial issues because all offspring will have at least one copy of the mutation. With less serious simple recessive diseases breeding of affected dogs should be avoided if at all possible. Exceptions might include a very high-frequency disease (CEA in Collies), a breed with a very low population or extremely tight gene pool, or a dog from very rare bloodlines. If a dog affected with a recessive disease is bred, it should be bred only to clear-tested mates.

Carriers should be bred only to clear-tested mates with preference given to using clear offspring to carry on with. By doing this the mutation can be reduced to extremely low levels within a relatively short time while the carriers' genes for desirable traits can be passed along. If carriers are eliminated, 20 thousand other genes are tossed out in true bathwater-plusbaby fashion, without for whether the dog's variants of those 20K other genes are good, bad, or indifferent.

If the tested gene confers a risk factor, as is the case with the Aussie HSF4 cataract mutation, the situation becomes anything but black and white. There are a variety of issues that need to be taken into consideration before firm breeding decisions are made.

#### Risky Behavior

Risk factor genes really muddy the bathwater. From here on out, most of the disease genes to be identified are going to be for risk factors. We in dogs need to develop a constructive approach to controlling those diseases. A risk factor gene increases the probability that the individual will develop disease, but not every dog that has the disease-causing mutation will become ill. Who does and who does not is determined by other genes, environmental factors, or both. At present, we rarely know what these other genes or factors

An Aussie with the dominant mutation of the HSF4 gene may get cataracts with only one copy, but it also may not. Even dogs with two copies don't always develop cataracts. The degree of risk varies in genes like this, but with this particular mutation it is very high: A dog with it is 17 times more likely to get cataracts than one that doesn't have it.

How much extra risk a particular mutation brings needs to be considered. A gene variant that conferred a 2x risk is less likely to lead to disease than something which is 17x. However, the breeder needs to consider the average risk of that disease in the breed as a whole. If a condition is very rare, even a 10x risk factor may not be significant. Such a test might not be worth the cost unless a near relative has been diagnosed with the disease. Where a disease is common, even a small increase in risk may prove significant. Cataracts are the most common eye disease in Aussies, with 3-4% becoming affected at some point in lives. With a disease like this, even doubling the risk would be a reason for concern. 17x puts the test in the "must do" category.

#### Frequency

Another thing that must be considered when evaluating DNA test results is the frequency of the mutation in the breed. The MDR1 mutation, which causes sometimes lethal drug reactions in most of the collie-type breeds, as well as some others, varies in frequency from breed to breed. In those breeds where frequency is very high, like Collies, Australian

Shepherds, and Miniature American Shepherds (aka Mini Aussies) it is vital that all dogs be tested. Dogs with just one copy of this mutation can react to certain drugs. However, eliminating every dog with the mutation is extremely short-sighted and even dangerous in those breeds where it is common. (Remember the story of the Basenjis.)

The MDR1 mutation presents a problem to the dog and its owner only if the dog is given too much of certain drugs, something that would never occur in nature. Rather than being viewed as a reason to cull, this mutation should be treated as a fault. If the dog has many other faults, the combination of those may indicate it shouldn't be bred. However, if it is otherwise a very good individual, even dogs with two copies of this particular mutation might be bred to clear-tested mates. Over time the frequency of the mutation would be reduced without, in the case of Aussies, eliminating over half of the breed.

In some instances, as with Collie Eye Anomaly in Collies, the frequency of the mutation is so high that most dogs have two copies of the mutation and only a tiny percentage are clear. In such an instance, breeding double-mutant dogs to each other cannot be avoided. Even so, an effort to reduce its frequency should be made through careful use of quality dogs that have only one or no copies. It would take many generations, but eventually the frequency of the CEA mutation in that breed would be reduced. Breeders would need to beware turning clear or single mutation males into popular sires based simply on their CEA testing status. There is ample evidence of the harm done by popular sire breeding and

doing so based on the status of a single gene is especially risky.

The Aussie HSF4 mutation is also very common. About a quarter of the breed has at least one copy. Because the mutation can lead to cataracts sometime in life (age of onset varies greatly) some advocate not breeding any of these dogs. This author did so before large numbers of dogs had been screened. I have revised my opinion because a quarter of the breed has at least one copy. Immediately removing that many dogs could have dire consequences.

Cataracts are undesirable and no breeder wants to produce them, but quality of life issues for this disease are minimal to moderate. The only significant expense that might be associated with the disease is for cataract surgery, which is optional. Until such time as the frequency of the mutation has been significantly reduced it will be necessary to breed quality single-mutation dogs. They should be bred only to clear-tested mates and high preference should be given to retaining quality clear-tested offspring to carry on with. Dogs with the mutation should not be bred extensively. Three or fewer litters should be sufficient to provide sufficient clear replacements.

#### Clean Babies

DNA tests are with us to stay, and with good reason. It is up to us to use them wisely. If test results foul the bathwater, pull the plug and drain it away through careful and informed breeding practices so the "baby" – your dog's good qualities – will be with you to stay.

# Pedigrees The Breeders Road Map

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Road maps tell you where you've been and where you are going. If you are breeding dogs, pedigrees are your roadmap. They don't just tell you what has been. Used properly, they can give you a good idea of where you need to go. This doesn't mean that your goal is to make something that looks pretty on paper. You're breeding dogs, not documents but those documents contain a wealth of data and can

point you toward additional information. Analysis of pedigrees and supporting information will aid you in making informed breeding decisions.

#### **Pedigreed Dogs**

The use of written pedigrees is so intrinsic to the breeding of purebred dogs that the general public views them as synonymous.

Pedigree = purebred. But a pedigree isn't just part of the documentation you send along with a pup when it goes to its new home.

The standard pedigree format, sometimes called a horizontal pedigree, is a listing of recent ancestors ranked by generation. Along with the names of the ancestors you may or may not find additional information on such things as registration, appearance or date of birth. Most printed pedigrees show only three to five generations. The deeper the pedigree and the more supporting information it contains, the more useful it is as a research tool. However there are limits to what can be put on a single piece of paper. A great deal of vital information about the listed dogs cannot be found on a printed pedigree.

Vertical pedigrees show not only your dog, its parents and grandparents, but all of their full siblings. It may also contain information on things like hip status and can be useful in determining whether polygenic traits like HD may run in the family. Like the horizontal pedigree, the information listed is limited by the size of a piece of paper.

There is another type of pedigree, often called a genealogy chart, flows the opposite direction—from one or more ancestors down through their many descendants. These are commonly presented as a chart with squares and/or circles representing individuals and lines indicating mates and offspring. The number of generations shown depends on the reason for constructing the chart. Genealogy charts may also contain a limited amount of information about the individuals listed. Researchers use this type of pedigree to demonstrate patterns of descent for hereditary traits. A breeder might use it to show important descendants of one of her dogs. If you are a visual person, sketching out a pedigree descent chart and noting what you know about individuals in it may help you understand how a trait is flowing through your line or indicate how significantly a particular dog has impacted breeding program.

For the most part, we rely on the classic listing of the dog, its parents, grandparents, and so on. Generating pedigrees once required hours of tedious typing or hand copying. Thanks to computers, kennel management and pedigree software are readily available. Paper pedigrees can be printed as needed or posted on websites, sometimes customized to include additional information on the dogs, kennel logos, or photographs. Considerably more data can be

stored on each individual dog's record than was ever possible on a paper pedigree.

#### **Keeping Track of Business**

Whether you use paper records or a computer, you should record as much information as possible about all your dogs and as many of their relatives as possible. Also, record important facts about dogs that you may use in your breeding program. Don't neglect their relatives, either. The better kennel software will calculate the coefficient of inbreeding (COI.) You can use COI to monitor the level of inbreeding in your dogs or proposed matings.

Beyond the standard descriptive data and titles, note the strengths and weaknesses of each animal. If it has been bred, what do you know about its offspring? Have any of its relatives ever had an inherited disease or disqualifying fault? With this kind of information readily available, you may be able to focus on the best additions to your kennel or prepare a short list of potential studs for your bitch without ever leaving home or picking up a telephone.

For example, if your bitch's weakest point is straight stifles, you could pull up data on dogs you think might be complementary. What do your notes say about their stifle angulation? Remember to check their offspring-knowing the dog has consistently produced proper angulation will be a further point in his favor. Look at his siblings, parents, cousins, and other relatives. The more of them that have correct stifles, the more likely the dog is to produce them. If your bitch has been bred before. records on her offspring will indicate how frequently she throws her faulty stifles and whether any prior crosses resulted in improvement. If straight stifles are something you have been dealing with for several generations, look back at records from prior breedings. Which crosses resulted in consistently correct angulation? You can determine how likely you are to get a particular trait so long as you have enough pedigree data. This can be done for any trait, good or bad. It's no quarantee on the outcome, of course, but the point of the exercise is to map out the route that is most likely to get you to your desired destination.

#### **Number Crunching**

If a trait is due to a single gene you can calculate the probability that a dog will have the trait, based on which individuals in the pedigree

have exhibited or produced it and what is known about how the gene is passed.

Liver is recessive to black. If your bitch is liver and you want to breed her to a black dog, you know that all the pups will at least carry liver because your bitch only has liver versions of the gene. The probability that they at least will be carriers is 100%. But what is the probability that you will get liver pups?

If you don't know whether or not the sire is carrying liver, you need to look at the pedigree. If one of his parents was liver, he does. There is a 50% probability that a given pup will be liver. If both his parents were black, but the dog has a liver littermate, both his parents are carriers and there is a two in three chance he is carrying liver himself and a one in three that you will get a liver pup if you breed him to your bitch, or 33.3% probability.

What if you don't know the mode of inheritance for a trait or it is polygenic? The best way to calculate whether a given cross will produce a trait of unknown or complex inheritance is with Best Linear Unbiased Prediction (BLUP) analysis. BLUP has been used successfully to maintain high levels of marketability in livestock. It enables breeders to determine Estimated Breeding Values (EBVs) for herds and flocks as well as individuals, compensating for management and other environmental differences. The EBVs of one individual or group can be meaningfully compared to those of others. This helps breeders select stock that will best meet their goals, whether they are breeding commercial production animals or bloodstock.

BLUP can be applied equally well to dog breeding: It is used by Canine Companions for Independence and Seeing Eye, as well as some European breed organizations. At present none of the commercially available kennel pedigree software will perform BLUP analysis. The math and statistical analysis required to do it by hand are enough to make ordinary mortals tremble. It also requires a data set far more comprehensive than most breeders will be able to pull together.

A more practical pedigree analysis technique for the average dog breeder is a modification of percentage of ancestry. Percentage of ancestry, sometimes called percentage of blood, is used to determine how much an individual ancestor contributed to the pedigree. Each parent will have given your dog 50% of its genes. Statistically, each grandparent will have given 25%, great grandparents 12.5% and so on. There comes a

point when the probability is very low that your dog will have inherited any significant number of genes from that particular ancestor unless the ancestor appears in the pedigree multiple times. If a single dog is four times a great grandparent, he will have contributed about as many of his genes as did either of the actual parents—50%.

#### Examples:

Oso is a grandsire and a great grandsire: 25 + 12.5 = 37.5% ancestry.

Lady is great grand dam twice and great-great grand dam three times:

 $(2 \times 12.5) + (3 \times 6.25) = 43.75\%$  ancestry

Oso falls between a grandparent and a parent in his potential contribution to the pedigree. Lady is very nearly at the level of a parent. I say "potential" because we cannot know exactly which genes came down through the intervening generations. It may be everything the individual passed to its immediate descendant or it may be nothing. Dogs have around 20-30,000 genes, so in most cases the actual number of genes passed down will hover somewhere around the percentage of ancestry.

Percentage of ancestry cannot exceed 50%. No bitch or dog could be behind more than half of the possible lines of descent in a pedigree. If you come up with a number greater than 50, you have made an error. Percentage of ancestry allows you to determine how much individual ancestors may have contributed to your dog's genes. With minor modifications, the same technique and can be used to determine how likely you are to get a particular trait.

#### **In-Depth Analysis**

To determine the risk you will get particular traits, review the pedigree for dogs that have had or produced the trait. Except in the case of X-linked or single gene dominant traits, both parents of any individual exhibiting the trait will be carriers of genes for that trait. I usually go a step farther and note grandparents of individuals that had the trait. With polygenic traits, it is highly probable that the grandparents also carry genes for it. It's also very likely that you won't have full information, especially if the trait is one people don't want to admit their dogs have. However, you might know about one or more affected grandpups. I refer to grandparents of affected dogs as "suspect" carriers. Suspects should not be given the same weight as dogs that actually exhibited the

trait or produced it, but including them in your analysis is as a technique for incorporating the breath of pedigree necessary for evaluating risk of producing polygenic traits like hip dysplasia. You can effectively include vertical pedigree information without actually creating such a pedigree.

The farther back you go before you find a dog connected to a trait, the less that dog contributes to the risk. Dogs that have a trait are the ones most likely to pass it on, parents less so and grandparents even less. More distant ancestors are less likely with each subsequent generation to have contributed the necessary genes.

How many generations to analyze behind your dog or a proposed cross depends on a number of factors. If the trait is easily recognized (color, coat type) or commonly discussed (dentition) or if information is readily available (performance records), three generations might be enough. However if recording of the trait is inconsistent or it is one people may not mention for fear of stigma, five generations may be more revealing. Beyond five or six generations, likelihood that the trait could have been passed down without someone noting it becomes more and more improbable

When analyzing a pedigree, start with the first generation and move back along each line of descent. Once you find an individual connected to the trait you are looking for, note whether it was affected, carrier or suspect. Once you find something, do not proceed any farther along that particular line. If you note more distant ancestors behind that one, you will inflate your result. For example, if you find that the paternal grandsire has produced the color you are interested in, it does not matter that his sire was that color; the grandsire is the closest dog that you know had the gene, so skip to the paternal grand dam. If she has no connection to the trait, go to her sire, and so on. Continue with each line of descent until you either find something or reach the last generation you are searching.

The next task is to determine the likelihood that you will get the trait. These are not precise probability calculations. Rather they are a method of consistent comparative ranking. Who's higher and who's lower, with lower being preferable for undesirable traits like hereditary disease. If you were looking at desirable traits, you would want high numbers.

I assign a value of 10 to an affected dog that appears as a parent on the pedigree, 5 to a

carrier and 2.5 to suspect. For each generation back, I divide that score in half.

#### Example:

A suspect appears as a great grandparent -

2.5 (base score for a suspect) divided by 2 (grandparent) divided by 2 (great grandparent): 2.5/2/2 = .625 (which I round to 1)

Since gender isn't an issue in this kind of search, values over 5 (equivalent to 50% in a percentage of ancestry calculation) can easily result. It's also possible to get values over 10 (e.g. if the sire was affected and the dam a daughter of a carrier, the score would be 12.5.) Even though this procedure is similar to and based upon percentage of ancestry, the result is not a percentage calculation. I deliberately moved to a 10-point scale to help avoid confusion with percentages.

A pedigree I reviewed recently for epilepsy had a suspect parent (2.5), a carrier great grandparent (1.25), and five suspects on the 5<sup>th</sup> generation (5x .157). This resulted in a score of 4.535. Since the fractions of a point are not significant to the final result, I generally round to the nearest whole or, for anything under 1 up to 1. For similar reasons, I have made 10 my maximum score—once you have reached 10 points you are extremely likely to get the trait. If you prefer to use fractions, scores over 10, or a 100-point scale that is certainly acceptable so long as you are consistent in what you are doing.

#### The Good Stuff

The above system works best for traits you don't want and those that you do which are rare. For good things that occur more often than not you will want to determine the consistency of that trait across a horizontal pedigree or, better yet, in a vertical pedigree.

The modified percentage of ancestry method would work well for determining risk of hip dysplasia, but if what you want to determine is the likelihood you will get good or excellent hips in a cross, you would do better to look at hip scores across a vertical pedigree. Having all excellent hips behind your dog is no guarantee he won't produce HD. But if there is little or no HD in the vertical pedigree going back two or three generations and the scores were largely good or excellent, odds are your dog will produce good or excellent hips in his offspring.

### **Completing the Map**

Follow the procedures outlined above for every trait you consider important. You can use the results to determine potential weaknesses in a particular pedigree and map out a course for improvement. A pedigree is

very much your road map as a breeder. Whether it helps you find your way or not will depend on how well you have taken note of the road signs and landmarks you've discovered along the way.

## **Science and Your Dog**

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# Alphabet Soup Why do genes have such weird names?

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Remember the good old days when genes were so simple for a breeder to understand? They were dominant or recessive and occasionally incompletely dominant. We figured there was a gene for almost every trait, though a few were polygene so there wasn't much you could do about them. And the names! The names were easy: *A*, *B* or maybe *M*. A really fancy one might be *Tw*. Then came all the genome research and scientists found that dogs don't have a hundred thousand genes, but 20-30 thousand. And the names! ALX4, EPM2B, and HSF4 – where do they come up with these?

The truth is it was never simple. But years ago science new only a little and the average person—which included most dog breeders—knew even less and most of what was known didn't have a direct bearing on what we do. Now that science is able to read the genetic code and has started figuring out exactly what genes do, there's a huge amount of information out there that actually can be applied to not only breeding in general, but to specific breeds of dog.

It turns out that genes are part of a complex interconnected network. This network links not only genes, but other parts of the DNA and molecules within the cell that regulate and control the activity of genes. The names of the genes can tell us something about what roll they play in that network. Most of this knowledge isn't something we are going to apply to our dog breeding efforts on a daily basis, but understanding why the old terminology we used to describe specific genes has changed can help us absorb new scientific findings that may have a direct impact on our dogs' health and the choices we make as breeders. The more we understand, the better equipped we will be to breed better, healthier dogs.

Actually those old single-letter gene names were themselves short-hand for more descriptive terms. The names were almost always related to traits like coat color that were easy to identify. *A*, *B*, *M* and *Tw* were abbreviations for gene names: **A**gouti, **B**rown, **M**erle and **Tw**eed (the gene for a variation of merle sometimes called "harlequin" in Australian Shepherds.)

I'll be using bold capital letters to help the reader understand how the abbreviations are derived from the gene names. In actual usage they are not bold. The abbreviations, however, are always in caps – unless the researcher works with mice, in which case they capitalize only the first letter. (Some people just have to be different.) For our purposes as dog breeders we will go with what the dog researchers use, which is the all caps abbreviations.

Gene names in *Drosophila*, the fruit fly favored by generations of geneticists, often describe mutations associated with those genes: Buttonhead, wingless or hunchback. ALX4 (Aristaless-like homeobox 4) is a gene dogs and many other species share with *Drosophila* and the name is used for all these species even though the trait the name describes applies to insects: "Arista" refers to the bristle-like appendages on the end of the flies' antennae.

As you can see with ALX4, abbreviations are still in vogue, though they've gotten longer. Our old favorite *A* is now ASIP, short for **A**gouti **SI**gnal **P**eptide. We can still use the old short-hand as a convenience amongst ourselves, but if we want to look up recent research on a particular gene we need to know the current scientific abbreviation.

So how is it they come up with these weird names? One of the most common naming conventions is to use the protein the gene

produces. Remember *B*? It's short for "brown," or what we in dogs call variously liver, red, or chocolate. That gene is now called TYRP1 (**TY**rosinase **Related Protein 1.**) Another example among canine coat color genes is MLPH (**MeL**ano**PH**ilan) which we are more familiar with as *D*, or **D**ilute. A third example is AP3 (**A**daptor-related **P**rotein complex **3.**) A mutation of AP3 causes cyclic neutropenia, or "grey collie syndrome," a lethal congenital blood disorder in collies that also features an unusual grey coat color.

You may have noticed that several of the genes mentioned so far have numbers as part of their names. There is a reason for that: Some genes belong to "families," groups of genes with similar but slightly different end products. The number signifies which one of that family it is. The IGF (Insulin-like Growth Factor) family includes two genes. In humans these genes may be associated with eating disorders. A particular variation of IGF1 in dogs is associated with small body size. IGF2 also has an interesting canine connection: Whippets with one copy of an IGF2 mutation tend to be faster than those that lack it. However, having two copies makes the dog hyper-muscled, a trait referred to in the breed as "bully whippets" for their resemblance to the more muscular bully breeds, like American Staffordshire Terriers.

Remember the fruit fly gene ALX4? It's part of what might be considered a sub-family (Aristaless-like homeoboxes) of a larger group of homeobox genes. Homeoboxes are a type of DNA sequence that regulates developmental patterns. Here's another example from canine coat color: MC1R (MelanoCortin Receptor 1.) You'll note that the numbers sometimes appear before the "R" for "receptor," but this isn't consistent. MC1R is our old familiar "E" (Extension,) variants of which can give a dog a facial mask or yellow color.

Genes may also be named for what they do: Remember the odd fruit fly gene names, like "headless," mentioned earlier. HSF4 (**H**eat **S**hock transcription **F**actor **4**) is a member of a gene family that activates another group of genes called heat response genes under conditions of heat or other types of stress. In dogs, we know HSF4 best for its association with cataracts in Boston Terriers, Staffordshire Bull Terriers and Australian Shepherds. Another function-named gene, MITF (**MI**crophthalmia **T**ranscription **F**actor) is associated with abnormally small eyes (microphthalmia) in some species though not, apparently, in dogs. In dogs

it is another color gene, producing some, but not all, white spotting patterns. Our old favorite S (**S**potting) isn't a single gene after all. MITF is but the first to be identified.

Genes may be named because of association with a disease. EPM2B (Epilpesy Progressive Myoclonus 2B) causes a particular type of epilepsy specific to wirehair Dachshunds. The human version of this gene causes Lafora Disease, a lethal neurological disorder. However, the name of the human version of the gene, called a "homologue," is NHLRC1, or NHL (Non-Hodgkins Lymphoma) Repeat Containing 1. In humans the gene is also associated with a particular form of lymphoma, hence the name.

Genes like EPM2B/NHLRC1 wind up with different names in different species because they were discovered independently by researchers. In some cases the researchers may have been investigating different problems in the same species. In many cases scientific bodies have designated a particular form of the name as official in a given species. Ultimately, to save confusion, this will probably be the case with all genes across species.

Most genes do have homologues in different species, particularly those that are closely related. It isn't surprising that you would find homologues among different mammals, but we and our dogs share some genes with species that aren't closely related at all, like insects. (Remember ALX4?) Merle color – the old color gene designation is M - is the result of a version of the SILV (**SILV**er) gene. The name derives from the mouse, where it was determined to be the cause of the color variation of the same name. Because of the mouse origin of the name, you will often see it noted as "Silv." SILV is associated with diluted color in a variety of species, including horses, cattle, and even chickens. But not all genes are shared, even among related species. Homologues of SILV are found in a number of species, including the chimpanzee, but it may be absent in humans.

For once we have a short, simple name in SILV. However, there is another name that is becoming less used but will be found in older research on this particular gene: PMEL17 (Pre-MELanosomal protein 17.) Melanocytes are pigment cells, so the gene is involved with the development of those cells. In the case of merle, something interrupts the developmental pathway and diluted pigment is produced on some areas of the body.

Merle dogs aren't silver (or not completely, in the case of some blue merles) but if a gene is originally named in a different species – in this case the mouse – the name may be related to what was observed in that other species. In the case of SILV, it was the silver color of the mice. For our old friend ALX4 it was the lack of bristle-like features on the fruit fly's antennae. Merle and silver are different, but similar, traits. However, I think it's safe to say none of our dogs have bristles on their antennae!

A gene's name may be changed as science learns more about it. There is one that we in Australian Shepherds are very familiar with that has recently undergone a name change. A mutation of MDR1 (Multi-Drug Resistance 1) has a mutated form that can cause severe reactions to some medications. A DNA test has been available for several years and is commonly used by dog owners in several collie-type breeds, including the Aussie, and a couple of sighthounds. The gene's original name stems from cancer research. It was found to confer resistance to chemotherapy drugs. This is why it had the, to us, confusing name of Multidrug resistance 1 when we associate it with increased drug sensitivity in our dogs.

Research on this gene is ongoing and scientists have recently discovered that what we call MDR1 is actually a member of a superfamily of genes called ATP (adenosine tri-phosphate) Binding Complexes, or ABC. ATP is the fuel for cell operations. No ATP, nothing happens. Our old familiar MDR1 is now ABCB1, for ABC family, B subfamily, Gene 1. The lab that offers

the test is keeping the old MDR1 name to save confusion among the dog-owning public.

On a related note, even chromosomes have names. Different species have different numbers. While many genes are shared across species, the arrangements of genes on the chromosomes can be very different, so science has developed a short-hand method of describing chromosomes. Dog chromosomes are designated by CFA, followed by the number of the chromosome, or X and Y for the sex chromosomes. So why CFA? Canis FAmiliaris, the scientific name for the species. Therefore, dog chromosomes might be identified as CFA1. CFA32 or CFAX. The same system is used for other species: HSA - Homo Sapiens or BTA -**B**os **TA**rus (the cow.) So if you read something that says ASIP is on CFA24, you know that A, the agouti gene, is on your dog's 24th chromosome.

All this alphabet soup can seem confusing, but there is a logic and purpose to it. Knowing a bit about how these names arose and what the abbreviations stand for can help us better understand the genes we manipulate when breeding dogs and, for those so inclined, make it easier for us to do deeper study of genes that are of particular interest to us.

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# Canine Genetic Counseling A Discipline in Gestation

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It started with an e-mail, but could as easily have been a phone call or letter. Upon occasion, it takes place face-to-face. The writer told me his dog had been diagnosed with an inherited disease. He wanted to know what this meant not only for that dog, but for related dogs in his kennel and that those he'd sold to other

people. He gives me his name, Kevin, and that of his dog, though not everyone who contacts me is comfortable providing this information. Even if they don't, I'll do what I can to help them.

I'm not a vet, so I didn't offer Kevin treatment advice for his dog. I did ask how the dog was diagnosed and what testing had been

done. The disease is one that can be misdiagnosed. It also can be difficult to treat. I suggested he seek a second opinion from a specialist. Kevin was unfamiliar with the disease, so I gave him some basic information about it. I also told him it is genetic and how it is inherited.

I discussed the breeding implications for the dog and its kin. Since Kevin volunteered detailed information, including his dog's pedigree and relationship to his other dogs, I was able to tailor my response to his specific situation. Since there is a DNA test for this disease, I told him what it is, how to have it done and which dogs he should get tested. I explained what type of results he might receive, what they would mean and how to apply them to his breeding decisions. I suggest that after he has the test results, he get back in touch with me so we can discuss them in detail.

Kevin and many others like him have come to me because I have a high level of knowledge of canine genetics and hereditary disease. I am particularly knowledgeable about my own breed, the Australian Shepherd. Like Kevin, most people who contact me have Aussies. If you asked them why they consulted me, they'd say they wanted help or advice. What I have provided for them is genetic counseling.

Genetic counseling for humans has been an established discipline for many years. For dogs and other animals the practice is in its infancy and, for the most part, has yet to attain professional status or structure. Dog owners' need for effective genetic counseling is growing along with the ever-expanding knowledge about the canine genome.

Current sources of information about genetic issues in dogs are largely informal. A breeder with a problem often seeks advice of other breeders. An owner might ask her veterinarian. Club members may ask questions of their breed club's health committee. The motivated will embark on a self-education process, using libraries, the Internet and, where available, classes and seminars. For the most part, these sources will not be able provide advice tailored to the person's specific situation. Sometimes what the concerned dog owner acquires is misinformation that helps neither her nor her dog.

Self-education can be helpful, but without someone knowledgeable to mentor the process, the do-it-yourself student may find it difficult to determine which sources are the most up-to-date and accurate. Some apparent information sources may be aimed more at emptying your pocketbook than providing something that will actually help your dog.

The most frequently contacted sources of advice are probably breeders. The quality of advice gained in this fashion can vary tremendously, depending on the knowledge level and of the person being asked. While the insight of an experienced and knowledgeable breeder can be invaluable, not every breeder has attained that level of expertise and some, unfortunately, offer feedback colored by their personal attitude toward the dogs or persons involved.

It isn't unusual for a troubled breeder to seek advice from several people, leaving her with two or more conflicting opinions. If any of the advisors are personally vested in the dog under discussion, their emotional reactions can color their response. This can leave the breeder with more questions than answers, not to mention an emotional burden that only exacerbates her situation.

When a dog is diagnosed with a disease, the vet may offer breeding advice. However, unless the vet is particularly knowledgeable about not only genetics of that particular disease as well as its frequency in the dog's breed, the advice may amount no more than a warning against breeding the dog and, often, its relatives.

This does not imply that veterinarians aren't doing their job. Their specialty is the diagnosis and treatment of disease and injury. They treat multiple species. In dogs alone there are over 400 identified genetic diseases and a similar number of different breeds. Though a few vets specialize in genetic disease, for most of them genetics is secondary information that often does not play a large role in their practice. Human doctors, who deal with only one species, also generally do not to have a strong background in genetics. That is why genetic counseling exists as a separate, but complementary, discipline in the human medical field.

The science of genetics has been growing exponentially in recent years. The sequencing of the canine genome has lead to rapid advances in the identification not only of disease genes, but those associated with traits like coat color. All this research is leading to the development of a growing number of genetic

screening tests. The abundance of new information can be overwhelming and the need for advice tailored to specific breeders' concerns is growing.

Breeders need to know what genetic diseases and inherited faults they are likely to encounter in their breeds and how they are inherited. If there are screening tests, they need to know when and how those tests should be used. They also need to know what they can do to minimize the risk of things going awry in dogs of their breeding. The need for canine genetic counseling services provided by qualified individuals is obvious.

Human genetic counselors get their clients by referral from doctors or other medical professionals. Their advice is most frequently offered to individuals or couples who are concerned about future children. More recently they have also counseled individuals about DNA tests they may have had or are considering. Generally, only one disease is at issue. The counselor will explain how that disease is inherited. If screening tests are available, they may or may not have been done when the counselor first sees the clients. If the clients have not been tested she will explain what the tests are and what they can reveal, leaving the decision to test or not to the clients. If tests have been performed, she will interpret the results and explain what level of risk the clients might have for themselves or for giving birth to an affected offspring. Human genetic counselors will have access to the medical documentation pertinent to the case.

The canine genetic counselor may or may not be presented with documentation. If the person seeking counsel has learned that his dog's sire has produced a genetic problem, he may not have direct access to records or the treating veterinarian. The counselor must then make it clear that the advice given is based on the information as provided by the client. ("If your dog's brother has hereditary cataracts, then ...)

If the client is the owner of the affected dog, the counselor needs to determine whether the client's understanding of the situation is accurate. Educating the client or suggesting further consultation with treating vets or specialists may be necessary before any breeding advice can be given.

Because human genetic counselors are dealing with questions surrounding human

reproduction, they do not tell clients that they should or should not have offspring. They enable their clients to make an informed decision for themselves. The situation with purebred dogs is somewhat different; dogs' reproductive lives are very much under human control. While the ultimate decision of whether or not and how to breed remains with the breeder, it is my personal feeling that the canine genetic counselor would be remiss not to offer an informed opinion on whether or not to breed a particular dog under the circumstances presented. The counselor should let the client know what precautions should be considered in mate selection should he decide to breed the doa.

Even canine reproductive choices can be fraught with emotion. It is vital that anyone offering canine genetic counseling services remain objective and non-judgmental, remembering the ultimate decision belongs to the owner of the dog. One test of the counselor's objectivity unlikely to be encountered by those in human practice, is the possibility of having more than one client seek advice about the same situation. This has happened to me more than once. Ideally, the counselor should refer the second client to someone else. Unfortunately, there often isn't another person to refer to who has the necessary combination of disease and breedspecific knowledge. In addition, even admitting that "someone else" has already requested services can lead to a breech of confidence and possible repercussions toward the counselor or between the clients.

Since there are no formal guidelines for me to fall back on, my practice has been to treat parties that are openly cooperating with each other as a group once I have asked each, in turn, if they are willing to participate in a joint discussion. In other cases I've had two or more people contact me about the same circumstance with markedly different sets of "facts." In such an instance I may or may not know what the actual case is. Even if I have an opinions about which person is playing straight. I take each client at face value. I base my responses on the assumption that what the she has told me is accurate. I also go to great lengths to make sure no one is aware of other contacts about the case.

Another challenge faced by canine genetic counselors is the vast array of breeds, each representing a distinct population. I suspect that most people offering counseling

today are, like me, single-breed specialists. If someone is to adequately serve people with different breeds of dog, the advice must be appropriate for the breed.

Advice that is accurate for a Standard Poodle owner might differ from that given to someone with Mastiffs. The genetics of a particular disease may differ between the breeds, so an approach that could help with one might be unhelpful or even detrimental for the other. The frequency of the disease in the breed under discussion is also important. The breeding advice one might give for a rare problem can differ from that for dealing with a common disease.

While I am periodically contacted by people in other breeds, I make sure they are aware up front that I am not a specialist in their breed and can only offer general advice based on my own experience and what I am able to glean from reference works, which I cite so they can follow up themselves if they wish.

At this point in time there are no formal ethical or professional standards for genetic counseling on canine issues. As time goes by, the need for these services will grow. When the demand is sufficient, formal training programs should be established. Professional organizations will need to form, with the responsibility for developing and maintaining standards of performance and ethics. Once genetic counseling for canines and other animals becomes accepted, cooperative referral networks can be developed with the veterinary profession. Our dogs' health will be the better for it.

In the meantime, I and others like me will continue to study and keep up-to-date on research so we can share that knowledge and, most importantly, the application of the knowledge to dog breeding with Kevin and his fellow breeders for the benefit of their dogs.

# When Worlds Collide Purebred Dogs and Genetic Research

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An experienced breeder gathers volumes of data and writes to a researcher; she never gets a response. An owner with a sick dog and disposable income offers thousands of dollars for research and no one seem interested. A breed health advocate documents a common inherited problem only to be told by a board certified veterinarian, "That doesn't happen in your breed."

These true incidents exemplify the frustration dog people sometimes experience interacting with the research community. The author has worked with a number of researchers in canine genetics and hereditary disease and networked with numerous breed health advocates. The discussion that follows is drawn from our successes as well as our failures. I hope it will help you understand the research process and enable you to avoid common pitfalls that lead to misunderstanding between the research and dog communities.

#### Worlds apart

The dog world and the world of genetic research revolve around stars sometimes light years apart. If we in dogs are to interact successfully with geneticists and research veterinarians we need to study their world so we can understand how they work, what issues concern them, and how we can best facilitate their efforts.

Things that seem important to us may not be fruitful avenues for current research. The minutiae of breed type and behavior often cannot justify the time, effort and expense required to determine their inheritance on a molecular level. Adequate research funds are not likely to be available for the esoteric details of canine structure and breed type. Most researchers don't breed, work or compete with dogs. They may be utterly baffled by why we think some things important. Just as we don't always understand their jargon, they may be confused by ours. No one will investigate a trait

he cannot understand and may be unable to recognize.

Even a disease won't always provoke immediate scientific enthusiasm. In order to gain acceptance scientific discoveries must be published in peer-reviewed journals or, in recent times, result in patents or produce a marketable test. We in Australian Shepherds knew by the mid-1980s we had Collie Eye Anomaly in our breed. However, no researcher had yet investigated and published it. Lack of a reference led some veterinary ophthalmologists to tell breeders CEA didn't occur in Aussies.

Even after publication the process isn't necessarily final. Other experts may disagree. When a study of CEA in Aussies was finally published (Rubin, et al 1991) there were those in the research community who disagreed with some of the conclusions. The article provoked a spirited rebuttal in an editorial appearing in the same journal issue. Researchers debate canine genetics with as much fervor as any group of dog breeders. Until the scientific dust settles on a disputed point, breeders must rely on their own experience and the best current scientific information available. However, we must resist the temptation to subscribe to the viewpoint that is most convenient to our personal breeding plans.

One of the hallmarks of scientific investigation is that no theory, no matter how well established, and is immune from reasoned debate. Given sufficient compelling evidence, a generally accepted view will change. Sheila Schmutz of the University of Saskatchewan, studied a gene called melanocortin receptor 1 (MC1R) which is the same gene Clarence Little, an early researcher in canine color genetics. referred to as "E." Little proposed that in addition to versions of E that resulted in black or yellow (fawn) dogs, one type produced the brindle pattern and another produced black masks such as are seen in Mastiffs and Great Danes. This is the model that dog breeders have long used for the inheritance of mask and brindle. But Little worked long before scientists were able to pick apart the molecular structure of individual genes; his conclusions were based on breeding trials. Using DNA from a litter of Great Danes and their parents, Schmutz found that a certain version of MC1R could clearly be tied to the mask pattern, but none correlated with brindle. Little was right about mask but erred concerning brindle. As of this writing no one knows what gene causes it.

Sometimes these winds of scientific change slam hard against the monolith of purebred dogdom. We operate in a world steeped in tradition. Our actions spring from a body of knowledge handed down from one generation of breeders to the next, often in the form of oral history. We must be willing to set aside what we have believed for decades when new science demonstrates our viewpoint has been flawed. This isn't easy to do. Recent discoveries about gene function and interactions as well as the long established but unfamiliar principles of population genetics stir heated debates and fierce resistance from dogdom's traditionalists. Likewise scientists tend to put more stock in published findings than breeder information and if the two don't match, the breeder's idea will not be taken seriously. We need to do our homework. Acknowledging that "although Smith (1980) found that....., our data suggest this is not always the case in our breed" may make scientists take breeders more seriously from the outset.

The current state of genetic science may impede getting answers to some of our questions. After nearly a century of canine research, many single gene traits have been described and their mode of inheritance established. Any dog breeder worth his salt understands the genetics of this type of trait and can apply that knowledge to the betterment of his breeding program. But many issues of inheritance that puzzle us today are tougher nuts to crack.

Recently, researchers at the University of California, Davis, asked a group of breeders what we wanted to have studied with the ultimate goal of developing screening tests. Our list consisted of diseases that are polygenic or which result from an interaction of genes and environment. Complex traits are difficult to pin down at the current state of the art; screening tests will be a long while coming. Even so, we should keep asking because someday science will be able to tackle them. In the meanwhile, we need to concentrate on issues that are genetically simple and with which the researcher can have reasonable hope of success.

As with so many other aspects of life, money can be a problem. Research goals often include finding the responsible gene. Doing so bears a big price tag. A huge donation, from our point of view, might be totally inadequate. Mark Neff, of Davis' Veterinary Genetics Laboratory, pointed out that laboratory consumables for a single researcher can cost

about \$2000 per month. Salaries of the people working on the project raise the monthly cost even higher. A successful research project, from initiation to publication, costs around \$200,000. While the work is expensive, there are ways to maximize your contribution and make effective use of your money that will be discussed later in this article.

Prior to starting a project, the researcher will find out what is already known. He will determine whether anyone else is working on the topic. Another project in progress won't necessarily mean the end of yours, but knowing who else is interested may suggest a different approach or lead to collaboration. Or sound the gun on a race that can generate as much excitement as any contest between coursing hounds. Parallel genome sequencing projects conducted by the publicly supported Human Genome Project and Celera Genomics, a commercial lab, generated scientific enthusiasm and considerable media attention.

With groundwork laid, data gathering can commence. If the mode of inheritance is unknown, necessary data may be pedigrees of animals exhibiting the trait combined with photographs, screening reports, lab results, or other documents. These will be used to develop genealogies that illustrate patterns of inheritance. If a probable mode is identified, test matings may be done to confirm it. For decades, all studies of inheritance used this type of data and analysis. Today it's possible to find the responsible gene and this is often the preferable approach.

To accomplish this breeders and owners must provide not only demographic, pedigree and diagnostic information, but DNA samples as well, usually in the form of a cheek swab or blood draw. The samples cannot come from just any dog, but must be drawn from affected animals and their closest kin. Which dogs qualify depends on the protocols developed for the project. Submitting samples that don't meet the protocol wastes time and money.

Science is unpredictable. As a project develops it may hit a snag or some finding may require a change of focus. The researcher will not be able to provide a hard estimate for a completion date and may or may not be able to provide an estimated timeframe. If the project isn't very complex and everything goes smoothly, a year might suffice but it may also take many years. Researchers don't want to discourage people with a lengthy projection or mislead with one that is overly optimistic. They

may be uncomfortable committing to a schedule which circumstances could prevent them from keeping. Finding genes involves elements of looking for a needle in a haystack. Even with a big magnet, the researcher may need to probe from several directions before he finds that needle.

Most dog studies won't be the only thing a researcher is doing. Higher priority projects may be the ones that support your researcher so she has time and resources for yours. No one in academic (university based) research works solely on a single project. Most will have administrative, supervisory or teaching duties that occupy a significant part of their time.

Even with the best minds and material and plenty of money, not every effort will be successful. If things don't work out as you hoped, be philosophical about it. Negative results mean there is one more thing you know it is not. Somewhere down the road someone may have a new idea or turn up some other information that will help complete your abandoned puzzle.

As in the world of purebred dogs, every once in a while research goes awry for reasons having noting to do with the matter at hand. A project may languish because of people or politics. Employees come and go, student researchers may quit, leave or otherwise abandon their work, and politics within a company, university, or professional organization may delay or terminate a project. Sometimes people die. In such instances, it may not be possible to pick up the pieces.

Once research is successfully concluded, the funding source and the researcher(s) who did the work will have a bearing on what happens next. If it was done by a commercial lab or funded by a corporation the results may be kept private or subject to patents. Dog breeders complain at the high cost of some tests, especially when they or their club provided some of the funds. Commercial interests expect to recoup their expenses and make a profit. If dog people provide significant funding, the club or other canine organization spearheading the effort should discuss the ultimate financial impacts with the researchers ahead of time.

Many universities now patent findings as well, primarily to protect the intellectual property of their researchers but also in hopes of generating income that will help underwrite future research. But university research is almost always published and becomes available to those who want to build upon it. Articles

detailing findings are submitted to peer reviewed journals. Peer review is a process by which other scientists not directly involved with the project give critique. If the article doesn't pass muster, it doesn't get published. Before publication, the researchers must limit public discussion of their findings. Saying too much may prevent publication or jeopardize the entire project.

Another danger of releasing preliminary results is that as the project progresses, early interpretations may prove incorrect. The researcher does not want to confuse or mislead you by saying too much too soon. While you will not be able to get detailed reports prior to publication, major contributors should be offered periodic progress summaries.

#### Luck be a lady

Even in science, pure chance can play a role. Shortly after Dr. Schmutz located the canine brown (liver) gene, John Potter, a breeder of Dexter cattle contacted her. He believed his cattle were brown and, in spite of several prior rebuffs, persisted in seeking someone who would look at what he had. Receiving Potter's information at that particular time enabled Schmutz to do a follow-up study on the cattle version of the gene.

#### Get the ball rolling

Successfully instigating research on your own is a daunting task. It requires many hours, incredible effort, and meticulous record keeping. You also need well-developed people skills and a degree of obsession that might justifiably be called crazy. A better approach is to gather a group of like-minded individuals to share the load.

Ideally, such a group would work under the auspices and with the support of a breed club or breed health organization. Sometimes that is not possible. If so, an ad hoc group will do so long as everyone involved is clear on the common purpose and goals and is willing to share in the work.

A project must stir interest within the breed community to generate data. Health surveys can be an excellent tool to determine what breeders and owners feel is important, as well as provide some indication of what data might be available. Statistics from health registry and veterinary school databases can also point you toward potentially fruitful topics.

Education is everything

Once a subject is identified, the first task is to convince others that research is needed. Do some homework to get the facts. Learn what research has already been done on other species in addition to other breeds of dog. Have some idea of how frequent the trait is in your breed. Put your findings to work in an education campaign designed to inform the average owner or breeder. Use the Internet and e-mail discussion lists. Put articles in club newsletters and breed magazines. Talk to regional clubs and other interested groups. Set up an information booth or hand out pamphlets at major breed events. Reach out to dog owners as well as breeders. Those not involved with the breed mainstream may have dogs valuable to research.

If information is too technical, you lose people. Keep initial efforts at a level most people will comprehend. Append a list of references and resources for those who want to learn more. Be positive. The danger to the breed should not be soft peddled, but launching a campaign with an accusatory tone will turn people off.

Present the problem. If necessary, indicate that some people (not all) are acting in a manner detrimental to the breed. If you do this, also provide suggestions for positive action. Do not propose punitive measures against malefactors; witch-hunts are counter-productive.

Keep the issue current. The more people hear—and the more sources they hear it from—the more likely they are to acknowledge its importance. If you find articles by people from other breeds or, better yet, people well known for their involvement with canine genetic issues, ask for reprint permission in breed publications or on a website. Most writers are happy to cooperate.

If you have an interested researcher who can effectively communicate with lay people, arrange a speaking engagement. Make sure you handle your end professionally. Determine whether you need to provide transportation and housing, have an appropriate venue, and make sure you can get any necessary equipment or supplies. Be organized and ready to start on time. Poor hospitality and sloppy event management may burn an important bridge. Don't oversell: If you promise a huge audience and only five people show up, you risk discouraging and embarrassing your speaker.

The right man (or woman) for the job

Once people are aware and concerned, it's time to start looking for a researcher if you don't already have one lined up. Know who is currently working in the field and whether they might be interested in dogs. Initiate contacts. E-mail is the most convenient and effective route. Universities often have faculty directories and commercial labs will have contact information on their websites. If you can't find an e-mail address try a letter or phone message, but be aware these sometimes don't reach the intended recipient or might get set aside in the press of other business. Follow up after a while if you don't receive a response, but don't be rude or become a pest.

However you make contact be brief, businesslike, and to the point. A breeder who had a unique and valuable set of data sent a long, detailed letter to a researcher and never got an answer. She was understandably unhappy at being ignored after all her effort. Coincidentally, I happened to attend a meeting at which that researcher was present. In a discussion about dealing with dog people, the researcher mentioned receiving a letter so long and detailed she didn't have time to plow through it. The researcher's frustration is also understandable.

Be patient and persistent; finding the right researcher can take time. I spent several years trying to locate someone interested in reviewing the Collie Eye Anomaly data on Australian Shepherds. A number of researchers acknowledged I was on to something, but they were involved in other projects or it wasn't in their area of interest or expertise. I kept at it, eventually obtaining introduction to Lionel Rubin of the University of Pennsylvania, who ultimately wrote journal article.

If you meet with researchers, set aside any preconceptions you may have about what a scientist should look like. There is no conformation standard requiring white lab coats, pocket protectors and horn-rim glasses. If you are accustomed to conducting discussions and negotiations in formal business settings, don't assume the researcher must wear a three-piece suit to be worthy of consideration. If you meet at or near the place the researcher works and she shows up looking like she spent the night in a barn, she may have had to do just that. What is important is not what researchers look like, but their training, prior research and willingness to consider your project.

Good people skills on the part of the researcher are helpful, but not vital. A friendly, accessible researcher will engage people. Our dogs are our friends and family members. We want to feel the researcher cares. However, the best researcher for the job may not be good at schmoozing or have the time to engage in public relations efforts. At a meeting between researchers and breeders at the University of California-Davis in January 2003, researchers bemoaned the level of involvement and response often demanded by dog people. A board member of one of the major grant funding agencies remarked, "We are a needy bunch."

And so we are, but we need to be aware that not every researcher will be comfortable with intense relationships with dog owners or being bombarded with calls, letters and e-mails. Stick to business unless invited to do otherwise. You should not expect a researcher to engage in social chit-chat or extended back-and-forth about minor details. That is not to say that you might not develop a friendship, but this is not your purpose.

Try to determine researchers' comfort level with owner contact. If it is low, arrange for some kind of intermediary who can answer simple questions for dog owners and offer a sympathetic ear, succinctly restate information and pertinent questions for the researcher, and then relay the response back to the owner in language she will understand.

Some labs and universities have research coordinators who field all contacts from breeders and dog owners. The University of Missouri and the commercial lab VetGen have both done this for their separate canine epilepsy research efforts. They currently have excellent individuals filling those positions. The right person makes people feel comfortable and confident in their dealings with the project. This facilitates communication and provides positive word-of-mouth promotion. Financial constraints may render some university research programs unable to support this kind of position. In that case, the club, breed foundation or other group should offer to help.

#### Brass tacks

Once researchers are involved, get organized so you can facilitate their efforts. Someone should be designated as the group's liaison with the scientists. Preferably this should be a person with the knowledge and expertise to handle technical issues and render them into lay terminology. The dog world has its own jargon

and every breed has additional unique language; the liaison may also find himself translating "dog speak" for the researcher.

Someone should be the primary public spokesperson to club members and other dog people. This might be the researcher liaison but can also be someone else. Whoever has the job should be experienced with public relations or education, both as a writer and speaker. This individual must be able to present information in a manner understandable to the average person. Avoid assigning anyone highly controversial. People may refuse to cooperate with the research because they are in disagreement with the spokesperson over some other issue.

For both liaison and spokesperson, people skills are key. Polite and friendly but professional individuals will get better response from dog people and researchers alike.

Avoid discouraging volunteers. If someone wants to help, try to find something for him to do. If he offers a good idea, be sure to include him in its execution. If the idea isn't workable, politely decline and try to involve him in another way.

#### Pulling it all together

Gathering sufficient samples is the key to success. The journal article on CEA in Aussies got written because I had previously accumulated the pedigrees and CERF forms that made up the data. Where DNA samples are necessary, the cooperation of numerous owners and breeders is required. Samples will in most cases be gathered after the commencement of the project, though a gene or DNA bank is an excellent tool for storing samples against future need. Lacking sufficient data or banked samples, it takes publicity, loads of encouragement, and maybe a few samplegathering clinics to get enough to do a study.

Make sure both you and the researcher are clear about how samples are to be handled and what is to be done with them. Someone needs to maintain a log of all samples received, including their current location and status. The researcher should see that this is done, but if a club or other group is responsible at any level it should keep its own log. Know what will happen to the samples once the project is complete. In some cases, they will be discarded. In others, they might be kept and stored for use on future projects. Determine this up front and make sure donors are so advised.

Confidentiality is key. Hereditary disease is an emotional issue. People must feel that the data they provide will not be abused or used in a manner they didn't intend. People who are actively breeding or competing should not have access to detailed information on who has participated, if at all possible. This includes researchers who happen to be active in the breed under investigation. One such breeder-researcher set up procedures that assigned someone else to review incoming samples and maintain the full data files. That individual assigned an ID number to each sample. When the researcher got them, she had no idea what specific dogs they came from.

People take pride in the fact that they and their dogs contributed. They want to know how it all comes out. Study results should be made available on websites or in breed publications at the conclusion of the project. People who contribute may want to know specifically what was learned from their own dog's sample. Donors need to be informed that this information is rarely available.

#### \$\$\$\$\$\$\$\$

Data alone is not enough. Locating a gene can cost \$50-100 thousand dollars or more. Dog groups should be willing to provide at least some of the funds but few clubs have the ability to raise that much money. Even fewer have the expertise to review grant applications and oversee recipients. Clubs, breed health organizations, and groups of concerned individuals can team up with organizations like Morris Animal Foundation and AKC's Canine Health Foundation to provide more financial support than your group could gather on its own. The foundations have trained staff that provides the necessary review and oversight. Joint fundraising efforts, like CHF's "Donor Advised Funds" also provide a method of making donations tax deductible.

Financial benefits can flow two ways. If your club or health organization will provide most or all the data for research likely to result in a screening test or marketable product, you might want to bargain for some level of return for your effort, like a certain number of no-cost screenings for those individuals who provided data or a testing fee discount for club members over a defined period of time. Such negotiations are best done up front.

The end of the road

Not every research project will be fruitful. Not every relationship between researchers and dog people works out. If it doesn't, accept the situation without blame or recriminations and resolve to do better next time. Choose your subsequent effort carefully. Too many spectacular failures chill response.

The road to a successful collaboration between dog people and researchers may sometimes be long or difficult. Understanding how research works and what is required of you can smooth over the rough spots. Make the effort to build that bridge from our world to theirs. The journey will ultimately benefit both science and our dogs.

# DNA Fatigue Why Keep Giving?

November 2006, Rev. May 2013

Hurricanes, tsunamis, earthquakes—the devastating series of major disasters in the recent past spurred world-wide calls for donations to relief organizations. As time went by and more crises occurred, relief organizations worried that "donor fatigue" would hinder their ability to respond. There is only so much people are willing, or able, to give.

While difficulties generated by canine genetic issues have little impact on the world at large, the scientific and technological explosion in the field of genetics may be producing its own variety of donor fatigue. These advances provide great benefit to dogs, so their owners and breeders are recipients of a barrage of requests for canine DNA samples. Major registries offer DNA parentage verification services that, in some instances, require DNA to be submitted before registration can proceed. A growing number of research projects actively seek donations of DNA samples through messages to canine chat lists and advertising or notices in dog-oriented magazines and newsletters. Some researchers set up DNA collection clinics at canine events.

One canine geneticist recently lamented that, where once he had found breeders and owners not only willing but anxious to supply samples, more recently he met rebuffs. Some people told him they had "already donated." How much DNA donating is too much? And how often should we be expected to provide samples?

Understanding what you are donating for, the type of sample you are providing, and who it is going to is important. All may have a

bearing on whether you do—or don't—wish to give and whether a previous donation may apply to the current request.

Dog owners sometimes ask if the DNA they sent to a registry's DNA program can be used by a researcher who is investigating a breed health issue. The purpose of the registryoperated programs is to establish positive identification for individual dogs and verifying parentage. These programs help maintain studbook integrity for the registry and give owners and breeders a vehicle by which a dog can be positively identified and its parentage assured. The sample is sent to a commercial lab that determines the DNA markers for that dog and issues a report for the registry and the owner. Samples, and even test results, cannot. in most cases, be utilized by other registries. In the United States, both the American Kennel Club and the United Kennel Club have DNA programs. They use the same lab to process the samples. UKC will accept AKC results, but the reverse is not true. For Australian Shepherds, accepted for registration by both UKC and AKC, there is also a third DNA program administered by the Australian Shepherd Club of America. Those results are not accepted by either UKC or AKC. ASCA cannot utilize the results of the all-breed registries' programs because ASCA's program uses a different marker set, tailored specifically to Aussies.

Programs in other countries may or may not use the same marker sets and the registries may or may not accept results from US registry programs. If you are importing or exporting a dog and either DNA-ID or parentage verification on the dog will be necessary, you will need to check to see what might be required and whether results from the country of origin are acceptable to the registry in the receiving country.

Samples sent to registry programs generally will not be used for any other purpose. Depending on the sample type, they may be too small to be helpful in a research context even if transfer were available. In general, you should consider DNA samples submitted to a registry program to be for that purpose only.

DNA samples for research are usually submitted by the owner directly to the researcher. Usually these samples will be used only for that particular project or other projects done by the same investigator or at the same institution. Only occasionally will samples be forwarded from one institution to another unless they are cooperating on the same project. Most research institutions are not maintaining DNA "banks" openly available to the entire research community. When you give a DNA sample on your dog, you should be sure you understand what the sample will be used for and whether it will be retained for future use. You should be able to find that information in the project's literature or on its website. If not, you should ask.

When you give a sample to research, the type of sample has a bearing on whether any of it might be available for further projects. The most typical sample types are whole blood and buccal (cheek) swabs. For the most part, buccal swabs contain only enough DNA for the project at hand and do not store well. The University of California-Davis has been successful at extending the use of this type of sample, but thus far most other institutions have not. Blood samples are preferred if multiple uses or long-term storage are anticipated. However, researchers realize that it is easier to get samples if swabs are an option, since they don't require a vet visit or special shipping.

Occasionally other types of samples, like tissue taken during surgery or necropsy, might be accepted for a research project. However, you need to verify that the study can accept them, then make timely arrangements for the proper preservation and shipment of these atypical samples. Tissue samples your vet sends to a pathology lab may or may not be stored after testing and should not be assumed to be available for transfer to researchers unless prior arrangements have been made.

DNA sent to commercial labs for any of the various DNA tests for diseases or coat color are very unlikely to be made available for any other use. Most such labs have a working relationship with the research institutions that did the basic research leading to the tests they offer. They may share a particularly interesting sample with that institution, but this would not be standard practice for all samples received, nor will they make samples openly available for other purposes. These tests frequently use buccal swabs for test samples, limiting additional-purpose use. The DNA samples sent to commercial labs should never be looked on as a repository for future use unless the lab has specifically stated they will be retained for such purposes.

In general, every DNA sample you submit on a dog should be assumed to be for one-time use unless you have been specifically informed otherwise. When new research projects start up or new tests are developed, expect to give new samples.

Research projects are largely dependent on voluntary submissions of DNA samples and data by dog owners. If you adopt an "I already gave" attitude, you might stifle research vital to your dogs' health and wellbeing. It is vital that breeders and owners continue to provide appropriate samples to pertinent research efforts.

"Appropriate" samples are those that fit the protocols of the particular study. If you offer a sample from your dog and it is turned down because it doesn't meet the criteria for the project, don't take it personally. The researchers simply want to get the samples most likely to help them reach their research goal. Samples from dogs diagnosed with whatever disease is under study are always welcome, though some studies are limited to one or a few breeds or the dogs sampled may need to meet particular diagnostic criteria to fit the study. Sometimes relatives of affected dogs are wanted. The most current techniques require a set of unrelated. healthy "controls" to compare to the affected dogs. In most cases, the affected dog needs to be part of the study before samples of healthy relatives are wanted. The definition of "relative" will vary from one study to another. You need to make sure your dog meets the researchers' needs. Sending unsuitable samples is a waste of time and expense for the researchers.

If, for any reason, you are unhappy with a particular researcher, don't refuse to support research in general. Researchers are every bit as individual as dog people. Your feelings about one individual or research group shouldn't extend to the entire profession.

Research takes time and not every project will be successful. The failure of a particular effort to produce short term results—or any results at all—does not necessarily mean the researchers weren't doing their jobs. The samples you send to their next project may be the ones that give you the answers you need.

If you've been in dogs for a long time, you've probably felt the disappointment of not being able to assist a research project that comes along after you lose a dog to the disease, putting a potentially valuable sample beyond reach. DNA banks are an attempt to overcome that problem. This type of "bank" is a collection of DNA samples linked to pedigree and health data. Some research institutions maintain reference sample collections for one or more breeds. Some laboratories offer sample storage programs to breed groups, generally for a fee which may require periodic renewal. Most of these programs are limited to one or a few breeds. Samples may or may not be accessible to the donor after submission, making them unavailable for screening tests developed after the dog is gone. If you submit a sample to one of these programs, be sure you are familiar with the scope of the program, who has access to portions of the samples, and under what conditions.

The program that may offer the greatest potential for long-term storage of a comprehensive set of purebred dog samples is operated by the Canine Health Information Center (CHIC.) After a trial program with the

cooperation of the Golden Retriever Club of America, the CHIC DNA Repository was opened to all CHIC breeds in April of 2006. Over 150 breeds are participating in CHIC, which was established by the Orthopedic Foundation for Animals and the AKC Canine Health Foundation to promote the open exchange of health information. The contact networks of these two organizations in the canine research community make CHIC ideally situated for providing a high-profile, widely accessible DNA storage facility.

CHIC's DNA Repository accepts both blood and buccal swab samples. Blood is processed and stored by the Animal Molecular Genetics Laboratory at the University of Missouri. The Veterinary Genetics Laboratory at the University of California-Davis maintains the cheek swab collection. Information on the program can be found at http://www.caninehealthinfo.org/dnabank.html.

Your dog has plenty of DNA to spare, even after you've submitted to registry programs and had breed-appropriate DNA screening tests done. When you become aware of a research project to which your dog might contribute, send in a sample. Buccal swabs are easy and generally aren't going to cost you more than the postage to mail them back. Even blood samples aren't that difficult, though the shipping requires more care. Vets often waive fees if they know the draw is for research, especially if it doesn't require a special appointment. If a clinic is available at a show or other event, make use of it. And remember the importance of setting aside for the future and get samples from your dogs into a DNA bank.

Most importantly, never tell a researcher, "I already gave." The sample you refuse may hold the key to solving a serious breed health issue.

# What *are* they looking for? Research Data

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"I told them all about Blue, and offered to give them any kind of samples they want. How come they told me no?" In three decades of providing genetic education and counseling for people with Australian Shepherds, I've heard this lament more than once. Rejection hurts and it smarts all the more when you have a dog that is sick or

you've just discovered offspring of your top ranking stud dog have been diagnosed with a genetic disease. Even so, you shouldn't take it personally nor view it as an unreasonable roadblock to returning a sick dog to health.

There are many reasons why researchers may not be able to use information or samples from your dog. Simply stated, the dog must fit the protocols for the study. Protocols are the rules that define how a project will be conducted and what constitutes appropriate data. To make an obvious example, if a study focused on hip dysplasia, researchers would not be looking for samples from dogs with cataracts. Few people would think of offering information or samples for a study when their dog has a different disease, but oftentimes the needs of a particular project may not be obvious to the concerned owner or breeder.

There are many different types of studies designed to investigate different aspects of canine hereditary disease. Clinical research aims at discovering how the disease presents or progresses or whether a particular treatment will be effective. Epidemiology studies focus on how illnesses arise in and are transmitted through populations. Cellular and molecular research investigate nano-scale disease processes, sometimes on the level of the DNA. Other DNA studies may try to locate responsible genes or things which change the function of genes.

All studies have particular needs when it comes to data. What works for one may not work for another, even if the studies seem similar in type or target the same disease. However, there are some things that will apply in most cases and understanding that will help you determine whether or not you may be able to provide useful data to a particular study.

#### **Clinical Studies**

A clinical study might focus on all breeds or only one, but the goal will be to improve diagnosis or treatment of the disease.

A hip dysplasia study investigating the value of a new surgical technique might accept dogs of any breed, including mixes. They wouldn't want healthy dogs, even if a healthy individual had numerous affected relatives. The point is to see if treatment works. Even an affected dog might not be accepted for a study if the specifics of its condition didn't meet study protocols or if it had other health issues that could cloud the result. Dogs with bad hearts, for example, are poor surgery candidates, and would likely be rejected for an investigation of a

surgical technique. If the study was investigating a drug that could reduce degenerative joint disease in dysplastic dogs, researchers might want only dogs that had HD and degenerative joint disease.

A clinical study focused on prevention might be confined to a single breed or even a strain within a breed. Having a genetically consistent study population to draw from enables researchers to better evaluate environmental influences on the disease. Dogs who are already ill might or might not be needed.

Some diseases, like Progressive Retinal Atrophy, can be genetically different from breed to breed. It may be necessary to work only with a single breed to discover what screening and/or breeding practices will best reduce disease incidence. Results may or may not apply to other breeds with the disease.

Age of the dog may be a factor, if the research focuses on initiation and early development of the disease. Treatment studies may seek young individuals, looking to prevent or cure the disease, or at least mitigate its effects down the line. Other studies might focus on geriatric aspects of a disease.

#### **Epidemiology**

Epidemiological studies aren't just for infectious disease. Genetic diseases also arise and are transmitted through a population. The source of a disease may be a mutation in an individual or via the introduction of one or more carriers to the population. A study population might include an entire breed (Australian Shepherds), a single line or segment of the breed (show line Aussies), or a geographically isolated group (show line Aussies in Australia.)

Environmental influences on a genetic disease might limit what dogs will be acceptable study subjects. If sun exposure played a roll, researchers might want dogs only from the desert southwest or Alaska. Maybe even both, depending on what specific effect sunlight--or the lack of it—has. In a case like this, they might not be able to use samples from an Irish Setter in Iowa or a Newfoundland in New York even though both were diagnosed with the disease.

Pedigrees are vital to genetic epidemiology research. Because of this, rescues and other dogs of uncertain ancestry would not be eligible.

#### Cellular, Molecular and DNA Research

Cellular and molecular studies of disease processes, as with similar clinical studies, will need participation from affected dogs. However, the dog itself may never need to leave home. It might even have died.

Researchers use tissue samples or products derived from them. One study might require freshly drawn blood, where another can use blood that has been properly stored. "Proper" storage will be defined by the needs of the study, as will the specific tissue type. In addition to blood, biopsy samples of skin, internal organs, muscle or bone might be needed. Occasionally hair or nail samples or semen will do, but this is rare.

The only definitive way to diagnose some diseases is on necropsy. In such cases, appropriate tissue from deceased dogs needs to be submitted in the manner specified. Despite what you might think after a steady diet of TV forensic crime dramas, samples from interred or cremated remains generally are not useful for research. Samples usually need to have been removed from fresh corpses for immediate use or be properly chilled, dried or treated with a preservative agent for later use. No single study is likely to accept all of the above.

DNA research conducted to locate genes often requires blood samples or cheek swabs. Some studies, as with CEN (above,) will accept other tissues. DNA studies may require samples from individual affected dogs or from family groups. If a family is needed, what relatives do and don't qualify as "family" will be defined. Pedigrees on sampled dogs must also be submitted so the researchers can develop genealogies, which serve as research tools and provide supporting documentation for their findings.

### **Longitudinal Studies**

A longitudinal study follows a group of subjects, called a "cohort," over a period of time. The period may range from months to decades depending on the focus of the study. These studies seek to find out what happens as time goes by...after diagnosis of a disease, after administration of a drug, or throughout a long-term course of medication. Some longitudinal studies are designed to follow an age group cohort of individuals, gathering data on their

environment, genetics, and unfolding health or behavioral history.

Subjects for longitudinal studies must fall into a specific class: Dogs with hip dysplasia, survivors or a specific cancer, individuals who received a particular drug as juveniles, or Golden Retrievers aged 1-3 years. Length of the study will depend on the questions the researchers expect to ask of the data they gather.

Study subjects (or their owners, in the case of dogs) usually need to submit information on a regular basis, report to the research group or a cooperating local vet or clinic periodically for exams and/or testing, possibly conform to specific dietary or other personal practices, and agree to stay in the study for its expected duration. If your dog doesn't meet the criteria for the study cohort or you aren't willing to comply with study requirements, your dog will not be accepted into the study.

#### What to do

If you have a dog that you think might be suitable for a study, do some homework before you contact the lab or university involved. If outside animals are needed for the work, researchers will often have information posted on their institution's website. Granting agencies, like Morris Animal Fund and the AKC Canine Health Foundation, routinely issue informational press releases and post supported studies on their websites. Read the information carefully to see whether your dog actually fits the needs of the study and whether you will be willing and able to do what is asked of you as owner. Follow directions for sample and data submission carefully and completely. If the study requires cases to be referred by a veterinarian, don't bombard the researcher with calls and e-mails. Ask your vet to look into it for you.

Having an understanding of the needs of different types of studies, doing your homework on research projects of interest, and following directions is the best way to have your dog participate. If a particular researcher still tells you he doesn't need samples from Blue, hank him and keep looking. You may find another with a project Blue can contribute to.

# Not Always Clean and Simple Science can help, but the rest is up to us...

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In a perfect world, once people in a particular breed recognized an inherited health problem, they would find an interested scientist who would then provide the breeders with specific information on how the disease is inherited and a DNA test that allows them to screen breeding stock for carrier status. Unfortunately, life is seldom so clean and simple for dog breeders or scientists.

Recently I had a conversation with a breeder who was unhappy because a particular researcher "didn't come through" on a project of great interest to her. She was skeptical that any project done at this researcher's institution should be supported. His failure to develop a wanted study made him untrustworthy in her eyes and put an onus on the university where he works.

The rapid advance of canine genetics and veterinary medical research in recent years has put the worlds of purebred dogs and scientific research in much closer proximity than ever before. Most of us know little about the inner workings of the scientific research community. It is easy to become frustrated when a much-wanted project doesn't flow smoothly, or perhaps fails to flow at all. It is the breed community's responsibility as individuals and, most importantly, clubs and breed health organizations, to make sure we have done everything we can to move research projects of interest toward successful conclusions. That means not only supporting the research effort but doing our homework before making commitments.

Research projects can go astray for numerous reasons beyond our control. However, if we become better informed about how the process works we can avoid investing too heavily in a project that is iffy. We also need to gain better understanding of our responsibilities as providers of data, samples and financial support.

#### What Happened?

A research project may not produce the outcome we desire for any number of reasons. Some problems prove extremely difficult to

solve, no matter how much data and samples, money and effort have been invested. Hip dysplasia is a common and serious problem in many breeds of dog. Thus far, science has given us a better understanding of the environmental influences on the disease and provided surgical treatments that can improve quality of life for the more severely affected dogs. However, we still have no way to screen breeding stock for HD genotype so we can stop producing dogs that have it. The complex genetics of HD and the influence of environment on gene action has thus far been too tangled a knot for researchers to untie, but not because they haven't tried.

To date, the only polygenic puzzle that has been partly put together is cardiomyopathy in Dobermans. Researchers were able to determine which genes were functioning abnormally in the heart tissue of affected dogs. Even so, there still isn't a screening test. The disease develops in adult dogs when genes that formerly worked properly cease to do so. Science must discover *why* this starts to happen before Doberman breeders can hope for a test that will identify affected dogs before they become ill or let them know which healthy dogs are carriers.

Our expectations of science must be reasonable, based on the current state of knowledge and technology. For some things we need to plan for the long haul. Someday we will have genetic screens for HD and cardiomyopathy, but it isn't likely to happen in the near future.

# **Money Money Money**

The best research project in the world won't go anywhere if the researcher can't get the money she needs to do the work. Good science is expensive. Even the most dedicated dog organizations may not be able to fully fund an important project. If other funding isn't available the work may not go forward. Marshaling our forces by forming alliances with other breeds or organizations may enable us to achieve our own breed's goals.

With luck, the researcher may already have financial support from government,

corporate or other private sources. If not, or if she has only acquired a portion of what she needs for the project, your breed may need to go into the fundraising business.

For a small project, this may be simple. Breed groups can rely on the same tactics they have successfully used to raise money for rescue or scholarships. Larger or more complex projects will be more expensive. Adequate funding might be raised if your breed organizations team up with their counterparts from other breeds that share your interest in the subject under study. For a common canine disease, like hip dysplasia, forming partnerships shouldn't be too difficult. However, if the subject of research is found only in a few breeds or, worse yet, is specific to your breed alone, you may need to look beyond single-breed organizations to larger organizations with broader focus.

Working cooperatively with granting agencies is an excellent way to increase not only the ability to fund a project, but to better ensure the viability of the project itself. There are numerous private and public granting agencies. Most focus on a particular area of science, a single disease or one or more species. The AKC Canine Health Foundation and the Morris Animal Foundation are both are dedicated to funding animal research, with CHF devoted exclusively to dogs. Working with a granting agency can help you assure that the money your breed provides is used as you intended it should be. In a recent conversation I had with an executive of a high-profile canine health organization, the executive remarked that even his organization teamed with granting agencies because they lacked the staff and inhouse scientific expertise to review and administer grants. This is the case for almost all breed clubs and breed health foundations, too. Recognizing your own limitations and arranging for qualified professional assistance makes it much more likely your hard-earned dollars will bear fruit.

#### A sample of this, a sample of that

When it comes to research, one of our primary jobs in the purebred community is to provide the researcher's raw materials—data, samples, and sometimes even dogs. Exactly what is needed will depend upon the researcher's needs. Surveys, clinical studies and molecular genetics research have different requirements.

Surveys will be data-based. Breed organizations frequently conduct their own surveys, either in-house or by hiring a researcher to gather and analyze the data. Sometimes a researcher will initiate his own data-based project. Information may come from vet schools, private practice vets, breeders and owners, other sources, or some combination of these. If data is requested of dog clubs or individuals, the breed club and health organizations should do what they can by way of education, promotion of the project, and encouraging participation to assure the data provided is as complete and accurate as possible. Results of a data-based study will be of greater value if the number of responses is statistically significant, either through percent of total breed population represented, or through its demographic breakdown.

Getting large numbers of responses can be difficult, particularly if the amount of information required is voluminous or complex. Mailers may be ignored for as many reasons as there are people on the mailing list. Internet-based submission can increase response but may skew it toward a particular group of respondents. Before a project begins, a great deal of thought needs to be given to how to reach potential respondents most effectively.

Demographic issues can go beyond the computer savvy of the breed community. If a club publicizes a study only among its members and the majority of owners and breeders are not members, the resulting data may be limited in volume, represent only a subset of the breed population, or, worst of all, not be sufficient to provide useful information. If a breed has significant subdivisions, as between function-bred and conformation lines, odds are the enthusiasts of one sort do not pay much attention to the publications aimed at the other. Promotion of studies needs to encompass media that will reach as much of the breed as possible.

One of the most frequent research requests nowadays is for DNA samples, usually in the form of blood or cheek swabs. Swabs are easiest to accumulate because almost anyone can do them. Blood samples provide much more genetic material and almost indefinite storage capability, but they usually require a vet visit and that the dogs undergo an invasive procedure, albeit a minor one. Depending on the needs of the research project, a particular form of sample submission may be required. If blood samples are called for, the club or health

organization may need to mount an education campaign to explain why swabs cannot be used. Participation can be encouraged and improved by hosting and promoting clinics at major breed events.

If there are multiple genetics projects involving your breed, people may tire of providing samples over and over again. The ideal way to address the need for samples is a long-term DNA "banking" system. With a DNA bank, samples and associated pedigree and health data can be kept on file against future need. Only one sample per dog will be needed. The stored sample will remain available indefinitely. A dog may provide a significant contribution to an as-yet unanticipated study many years after it has died.

Several individual breeds have set up their own DNA banks, but this system depends on regular and on-going breed club administration of the sample collection. Most clubs are governed and operated entirely by volunteers and do not own any permanent facilities where records or samples may be stored. An in-house program can be subject to shifts of interest or the loss of one key individual. A system that does not require frequent involvement by already busy club officials is preferable. The Canine Health Information Center, jointly operated by the Orthopedic Foundation for Animals and the CHF. established a DNA Repository for all CHICqualified breeds. Participation in CHIC is voluntary and open to any breed club, including those that are not AKC recognized. At present there are over 150 breeds participating.

The program is relatively new, but made the first distribution of samples in 2007 for a study of mast cell tumors in Golden Retrievers and more have followed since. Into the future, more grants of samples from individual breeds and groups of breeds will be provided to researchers and the fruits of this effort will benefit our dogs, both through improved clinical practice to aid the ill and more DNA screening tests that will enable breeders to avoid producing affected dogs altogether.

Anyone with a CHIC breed can submit samples (blood or swabs) to the CHIC DNA Repository. Ultimately, they plan to have an online password accessed database that will allow owners to update their dogs' records at need. The long-term benefits of this effort, to both the dog and research communities, are immense. By encouraging participation, clubs and breed health foundations can make sure needed

samples are available while at the same time greatly reducing "sample fatigue."

An additional benefit of the CHIC DNA Repository—something that can be difficult or impossible to achieve with samples submitted to individual researchers—is long-term participation by the breed club in decisions on when and how samples are to be used. Samples given to specific researchers may be used up during the course of the study. If there is residual material, it may be discarded once the work is complete. Material gathered may not be kept if the researcher cannot get sufficient samples to proceed and moves on to something else. If submissions are abundant, the researcher may choose to use them for other projects or send them on to other researchers either before or after working on the project of interest to your breed. Your club may not even be aware this is taking place. If this is going on and circumstances prevent the researcher from tackling your breed's issue for an extended period of time, the sample collection may dwindle to the point where more will be needed before work can proceed.

Unless your club or health organization has a specific contractual agreement with a researcher about how the samples are to be managed, there is little you can do should a project fail to materialize or bear fruit. Use of a DNA bank can prevent this from happening because each researcher will only receive the specific samples required for her project and only after review and approval of a request for those samples.

Improved technology has reduced the need for donation of live animals to research. However, clinical studies of a particular disease may require participation by live dogs. Often the dog may remain with its family, making periodic visits to the research site or the office of a cooperating veterinarian. If the project requires donation of live dogs to the study, the breed club or health organization should know how the animals will participate and what their fates will be upon completion of the project before agreeing to support it.

# **Personnel Management**

Like most of us, scientists are usually employees. They have to answer to superiors who may rearrange their priorities. They will also have other projects and duties that may take precedence over your breed's project.

Thanks to the wonder of Internet search engines, it isn't difficult to learn something about

a scientist whose work you may want to support. You will often find a bio and list of a researcher's citations (published research) on the website for the university or laboratory where he works.

How long has the researcher been working in the field? Long experience, including canine projects, is a plus. However, a young doctoral student or post-doc (someone who has recently completed a PhD but is still studying) may have enthusiasm and a will to prove himself that an older, established researcher may lack.

If the researcher has been around for a while, what projects has she completed? Did it concern dogs? Is that work recent, or has it been several years? Was she principle investigator, or a collaborator? If a principle investigator, how recently did she serve in this capacity? If a collaborator, can you determine whether she did substantive work on the project or merely provided previously gathered samples or data? If she works in an academic setting, has she advanced to the level one would expect for her years of service? Answers to these questions may indicate how viable the efforts of that person are apt to be.

Another thing to consider is whether a researcher has made a major mark in his field. Having a star performer involved in your breed's

project can be a very good thing. The top people attract the best staff and work at the best institutions. They attract funding. But they also have their pick of projects, are very busy, and may not find your project compelling. There are many excellent researchers who have not achieved star status (or may not want it!) who could give your project more time and attention than a Big Name is able to provide.

# **Due Diligence**

Science has provided tremendous gifts for the benefit of our dogs and will continue to do so into the future. It is our job, as members of the purebred dog community, to make sure we understand the process, get to know the players and provide the necessary resources, in the form of data, samples and/or money, that will allow science to continue giving us the answers we need for breed health issues.

Complaining about a particular study that went awry and laying blame on a researcher or institution will not produce results. When a study doesn't pan out, we need to take a dispassionate look at how it happened, what we could do better, and carry on in our effort to find and support a project that will bear fruit.

# **Controlling Genetic Disease**

# From the Top Down The role of dog organizations in canine health

First published in Double Helix Network News, Spring 2011

Nothing happens in a vacuum. No matter what the cause, progress doesn't happen without the passion, effort, and commitment of many people and the involvement of the organizations they form. This is no less true of canine health efforts than anything else. Everyone, from the hierarchy of the major kennel clubs at the top on down to the membership of your local breed club, needs to recognize that the health of our dogs is a high priority and take steps to insure that our practices promote not only dogs with sound minds and excellent structure but longevity and health, as well.

#### The major players

There was a time when health issues weren't given much attention. Even so, there were some shining achievements like the Golden Retriever Club of America's involvement in the founding of the Orthopedic Foundation for Animals (OFA.) For the most part health issues were on the back burner, if not in the closet. However, over the past couple of decades there has been a major shift in attitude and action at all levels of the purebred dog community, including at its highest levels: The national kennel clubs.

These clubs wield tremendous influence over a wide range of canine activities. Their programs provide the events we enjoy with our dogs while their studbooks maintain vital records of the heritage of the breeds they support. They are looked to by the public as well as dog enthusiasts as supreme authorities on almost anything pertaining to dogs. That authority can go a long way toward setting standards for health.

For many years a number of European kennel clubs have maintained open health

registries. The Swedish Kennel Club has one of the longest standing and makes for review health records available (kennet.skk.se/hunddata/) via their web site. Most of these registries include at least the results of eye, hip and elbow exams. Screening for diseases prevalent in particular breeds may also be included. In the past few years DNA health screening tests have been added to the mix. The various clubs publish the information in print form and sometimes, like the Swedish club, make it available via searchable on-line databases. The motivated breeder can use these resources to track breed health issues so they can make informed breeding decisions.

The Kennel Club in the United Kingdom, the first of its kind, set the standard for all others that followed. A few years ago they launched a health and welfare program which all kennel clubs could emulate. The Assured Breeder Scheme

(http://www.thekennelclub.org.uk/breeding/exper ienced-breeder/assured-breeder-scheme/) provides certification to breeders who meet certain standards of breeding practice, including breed-specific health screenings requirements and recommendations.

The American Kennel Club has also become much more active in health-related issues. In 1995 it established the AKC Canine Health Foundation (CHF, www.akcchf.org/) The AKC underwrites some of CHF's operating costs and provides generous contributions toward CHF's research grants. Since its founding, CHF has, among other things, provided stellar breeder and club education programs and millions of dollars for research on a wide range of canine health issues. The findings of those projects have lead to improved diagnostics and treatment, DNA screening tests for breeding

dogs, and better understanding of canine diseases.

### Get specific

The major kennel clubs can do the heavy lifting and set the tone in the campaign for improved health, but our many breeds are no more alike in health concerns than in appearance or behavior. Those best acquainted with the issues in any given breed are those who dedicate themselves to the preservation and improvement of each breed in the form of national breed clubs.

Most national breed clubs are affiliated with a major kennel club. However, a few, like the Schaferhund Verein (German Shepherd Dog Club) in Germany or the Australian Shepherd Club of America in the US, maintain their own studbooks and offer a variety of competitive programs. The extra responsibilities the club/registries shoulder due to their independent status does not exonerate them from responsibility for breed health. If anything, their responsibilities are greater. This is especially so those which are the only organization for their breeds.

Whether affiliated with a kennel club or not, breed clubs are ideally positioned to promote breed health initiatives. Every club has a multiplicity of programs, events and member services its board must oversee. If effective progress on the health front is to take place, the club must have a motivated and focused health and genetics committee peopled with individuals who are passionate about their breed and make health a high priority.

H&G committees conduct surveys to identify breed specific heath issues and track progress in reducing their frequency. They develop educational material for members and, just as important, everyone who owns their breed. The committee should serve the club board by providing information, suggestions and advice on health-related items that come before the board for review or action.

Research is vital to developing prevention and better treatment for disease, as well as providing breeders with tools that will enable them to avoid producing affected dogs. A good H&G committee will identify research projects focusing on its breed's concerns and, where needed, raise funds to support research, and organize sample and data collection where needed. Finally, H&Gs can encourage best health practices by developing recommended health screening protocols, creating recognition

programs for breeders who meet high health standards, and by organizing and operating screening and/or research clinics at the annual national specialty and other key events.

The national breed clubs should be their breeds' primary advocates for health. Every breed community has its own unique culture and quirks, but if the breed club "walks the talk" when it comes to health, member health awareness will be heightened. If a club fails to do so, the less motivated of its members will see little reason to make health a priority.

#### Laying a foundation

There are some things a breed club cannot easily accomplish because of their legal status as membership associations. Fundraising for research is vital if a club wants to make serious inroads on key breed health issues. But, in the US at least, donations made to clubs for charitable purposes cannot be deducted from the donor's taxes. For this reason, many clubs have set up charitable foundations. Ideally, the breed foundation should work with the breed club's board and H&G committee to identify areas where the foundation can contribute toward improving breed health. Stellar examples of breed foundations that have had a major impact on canine health include the American Boxer Charitable Foundation (www.abcfoundation.org/)and the Golden Retriever Foundation (www.goldenretrieverfoundation.org/.) Between them they have contributed hundreds of

thousands of dollars to canine health research.

While not every breed community has the numbers or the resources to achieve at the level established by the Boxer and Golden foundations, even a relatively small breed

foundation can take major strides ifs board is

motivated.

#### The grass roots

Regional breed clubs and their members are the grass roots of a breed community. Whatever plans and programs the major kennel clubs and national breed clubs devise, they succeed or fail on the acceptance by the breeders who make up much of the regional club membership. Major initiatives, in health or anything else, are spurred by the demands of those with "boots on the ground." If managing breed health issues isn't a significant concern for the regional club membership, not much is likely to happen on the national scale.

Aside from making their health-related wants and needs known at higher levels, regional clubs can take an active part in he process by educating members and the public about breed health issues and responsible breeding practices, developing a local resource list for owners of dogs afflicted by common diseases, sponsoring health screening clinics for breeders, and raising funds for breed health programs or research.

# No playing around

There are also hundreds of multi-breed regional clubs that are activity-based:
Obedience, agility, field trials, herding, and so on. While the focus of these groups is on their particular area of activity or competition, they, too, should become more proactive in matters of health. Sports injuries are an obvious concern, but activity club members should keep in mind that only a sound, healthy dog can have a full and successful performance career.

Like the regional breed clubs, activity clubs can provide pertinent health education to members and others and raise funds for research in sports medicine or diseases which impact participants in their sport. They can also let whatever national organization they are affiliated with know their concerns and push for national action to address those health issues.

# Let's get together

There is strength in numbers and no club or individual can do alone what many clubs and individuals might accomplish together. Most diseases are not unique to a single breed. Share ideas or join forces with other clubs whose dogs share your breed's issues.

There are also organizations outside the formal purebred dog club structures that can assist clubs with their health programs.

Consider partnering with non-affiliated health

organizations. Some of these groups focus on a single breed, like the Australian Shepherd Health & Genetics Institute (ASHGI) education, resources, and funds research. There are allbreed organizations, like the OFA (www.offa.org.) originally a hip registry but now offering not only an expanded semi-open health register but a wide variety of programs of value to breeders and clubs. Some groups serve multiple species, including dogs. The prime example in this are is the Morris Animal Foundation.

(www.morrisanimalfoundation.com/) MAF is a major source of research grants for companion animals and wildlife and has many active grants focused on canine diseases. Joining with groups like these can help dog clubs achieve health goals they might not be able to accomplish with only their own resources.

Over the past couple decades the world has become a much smaller place. Not only can we easily communicate with far corners of the world, our dogs can more readily travel to new homes in different countries. The health concerns for a particular breed in one country are apt to be much the same in another. Cooperation, exchange of knowledge and ideas, and partnership in disease control programs or research efforts doesn't need to stop at national borders.

Finally, every club depends on people, whether it is a membership organization or a "club of clubs" like the AKC. It is up to each of us as individuals to individually and collectively act and advocate for improved canine health. The dog world is replete with organizations, from Kennel clubs down to local groups with a dozen members. The more of us who take active steps to improve canine health, the more effective our organizations will be to the benefit of all dogs.

# The Importance of Community Working Together Toward Better Breed Health

First published in Double Helix Network News, Winter 2009

The mythic heroes of yesteryear singlehandedly performed mighty deeds: Paul Bunyan felled entire forests with one swing of his ax, Beowulf slew Grendel, and Hercules cleaned the Aegean stables (a dirty but doubtless necessary job.) But outside of myths, few great tasks can be accomplished by a single person; they require a community. A community might be a large as a nation or as small as a handful of cooperating dog breeders.

Genetic health issues are a significant concern in purebred dogs. Lack of focus and discord within our ranks are the biggest impediments to major progress in reducing the incidence of these diseases. No one person, in a single breed or within dogdom as a whole, can counter this trend. It takes a community.

#### Facing facts

Genetic health issues exist in all species, including dogs. We in purebreds need to accept that every dog has a few "bad" genes. The difference between mixes, which the public mistakenly believes are paragons of good health, and our purebreds is that if we work together and do our homework we can reasonably predict what traits we are or are not likely to get in a particular litter. This allows us to take effective steps toward reducing the frequency of unwanted traits, whether in conformation, behavior or health. This isn't true of mixes whose random and unplanned breeding makes any but the most obvious physical traits difficult-to-impossible to predict.

Because it is possible to predict which purebreds are more likely to produce particular traits – including genetic diseases – we can, through breed-specific communities, take effective measures toward reducing the frequency with which they occur. Cooperation between members of a community, be it a handful of like-minded breeders or the membership of a national breed club, can make this possible.

Breed health issues affect everyone, so the more people are encouraged to pull together, the better off that breed is. The Portuguese Water Dogs and Bernese Mountain Dogs are fortunate to have a significant number of breeders and owners focused on health. Their breed clubs make health a priority. Both these breeds are small in population and struggle with small gene pools. Even so, their dogs are better off than those in more populous breeds where health issues are not a priority. The PWD and Berner clubs achieve this through positive efforts.

Getting compliance with health initiatives can be difficult and frustrating, but draconian measures will only drive people out of your community, if not out of your breed. In fact, that is probably why Incorrigibles, who militantly

discourage discussion of health issues, resort to bully tactics: They make people who point out inconvenient truths sufficiently uncomfortable that they shut up or go away. The Incorrigibles may then continue business as usual with no bother about pesky health issues. It only takes a few well-placed and persistent Incorrigibles to wreck havoc on a breed's health. On the other hand, if health advocates enact restrictive rules or engage in loud public finger-pointing to enforce compliance, they may encourage people to give up or cover up to avoid hassle. Positive approaches and peer pressure are far more effective at making meaningful and lasting changes in attitude than bullying or by-the-book rules enforcement.

The "10-Steps" program operated by the Australian Shepherd Health & Genetics Institute, Inc. (ASHGI) was created as a positive approach to dealing with health issues. It not only suggests proactive steps individuals can take but discourages punitive behavior toward others. While the steps were written with Australian Shepherds in mind, they could easily be adapted to other breeds of dog or even other species.

There are also positive purebred health programs that cross breed barriers. The AKC Canine Health Foundation and Morris Animal Foundation have funded millions of dollars of research. No small amount of that money has come from dedicated breed people and clubs. The Canine Health Information Center (CHIC) program of the Orthopedic Foundation for Animals (OFA.) which currently includes over 120 breeds, is a major high-profile effort to make health information publicly available. Only with the open exchange of information on health traits can breeders can make informed decisions that will reduce the frequency of inherited disease. Breed clubs should make every effort to encourage participation in CHIC. If CHIC certification becomes as much a given as hip and eye exams are for many breeds, our dogs will be much better off.

#### **Teamwork**

Hereditary disease is everyone's problem: Breeders, competitors, pet people, and the trainers and handlers of working and field dogs. Each of us we must cooperate within our separate disciplines and areas of canine interest, with our local and national clubs, and between breed clubs. We must foster cooperation with non-breeders because they are the ones who have most of our dogs. We

should all be on the same team. Knowledge of the genetic status of non-breeding dogs is as important as that of those which are bred. Owners need to be made aware that the control of genetic disease is as important to them as it is to any breeder and encouraged to share health information on their dogs.

Breeders should strive to maintain open lines of contact with all their puppy people. Stud dog owners need to keep in touch with the owners of bitches that the stud has covered. Groups of breeders often form informal cooperatives for a wide variety of reasons; one of those reasons should be minimizing the frequency of genetic health issues through information exchange.

Regional clubs are an ideal resource for networking, distributing important information and educating members and the general public. Putting on quality trials and shows is important, exciting and fun, but health events and member health and genetics education are equally important. Regional clubs can offer health clinics and other events. Members can join together under the club banner to present their views on important health-related issues to their national clubs.

National clubs' health & genetics committees should take the lead in public education about breed health issues and provide their membership and affiliate regional clubs with up-to-date information on breed health issues. If affiliated with AKC or other all-breed clubs, they should interface with those organizations regarding health issues. The AKC established its Canine Health Foundation (CHF) to provide education, assist breed clubs with health initiatives and support important health research. AKC member clubs should make a positive working relationship with CHF a "must." Individual breed clubs also need to form alliances with other breeds to tackle common health issues. Few breeds have the sort of large well-funded foundations that benefit the Golden Retrievers or the Boxers, but by working cooperatively, smaller breed organizations can achieve important results.

Most breeds have only a single national breed club and, perhaps, a related foundation. But for those with multiple national clubs or registries, and for all breeds which exist in more than one nation, cooperation on health must be pursued even if there are other issues upon which neither side is likely to agree. To present one very convoluted example: The Australian Shepherd has two strong national breed clubs in

the US, the breed's country of origin. Each club has a foundation. There are also two independent health organizations in the US and somewhere upwards of a twenty other established clubs in other countries, most of them affiliated with or seeking affiliation with their national all-breed registry. Obviously all these groups have their own priorities and concerns. Some are to some degree antagonistic toward each other, but a few years ago when epilepsy was recognized as a serious breed health concern an international grassroots effort lead to a large collection of research samples and cooperation among key clubs. foundations and organizations to provide funding for a research project that involves an international collaboration. Is all now peace and bliss in the Aussie kingdom? No. But if we have put our differences aside to meet a common health goal once, we can do it again. And so can people in other breeds far less fragmented than ours.

#### Develop a game plan

You can't make progress unless you know where you are going. Health and genetics committees and breed health organizations need to identify the most significant health concerns then develop a set of best practices for breeders to follow.

Experienced breed people will know which health issues are most common, but without conducting a survey, important information may not be available. Long-time Aussie breeders recognized that cancer has become a more frequent occurrence, but since there are so many different cancers and so many potential environmental causes no one felt cancer was a breeding concern. In 2007. ASHGI completed a cancer-specific survey. While numerous types of cancer were reported, nearly half the dogs entered had either hemangiosarcoma or lymphoma. Subsequent examination of pedigrees demonstrated that these two cancers were familial in the breed, indicating some degree of heritability.

Targeted surveys like ASHGI's cancer effort can be useful, but periodic comprehensive health surveys are vital in every breed. The surveys need to encompass a broad range of health issues and might also include other problematic inherited traits, like non-standard colors or other disqualifying faults. Surveys need to be part of a club's long-range planning: Because populations change over time,

comprehensive surveys should be conducted about once a decade.

Putting a survey together, gathering the data and ultimately doing statistical analysis can be a daunting task. Contact other breed clubs or breed health organizations that have done surveys to ask about their experiences. CHF provides guidelines to help with survey development and OFA offers a survey service. In addition there are individuals and universities which will help develop surveys and analyze the data for a fee. (Ask for references.)

One of the greatest difficulties with surveys in the past was getting sufficient response. Today, web-mounting improves access and response. Paper copies can also be made available for those who cannot or will not utilize a web-mounted survey. Unless your breed has a small population and is largely in the hands of club members, encourage survey participation by non-members. The broader range of responses you get, not to mention larger numbers, the more useful the data will be.

Collecting the data electronically via a web-mounted survey form also saves a lot of time and effort during the data analysis phase. Proper statistical analysis is vital and if your club does not have someone with that expertise, you may need to hire someone to do it for you.

Once key areas of concern have been identified, get the word out to the entire breed community, including pet owners. Let breeders know what health issues are of greatest concern, how to recognize them and how best to avoid producing them. Provide up-to-date information on screening and diagnostic testing. For owners, make treatment information available. Encourage participation in research on those diseases and provide financial and logistical assistance to the scientists doing the work.

Develop a recommended <u>testing and screening protocol</u> and promote it to breeders. ASHGI has developed one for Aussies It could readily be adapted for other breeds.

#### Stay on message and on target

We must, within our various communities, reach a consensus on important health related goals and set priorities. Once that is accomplished we must work together to communicate the importance of those goals to the wider breed and canine communities with which we intersect.

The message should not be confined to a single health issue. Eye disease may be the

primary health concern now, but in a decade it might be epilepsy or cancer. What is important isn't the detail, but the approach. If your message lays the groundwork for constructive action for individuals as well as groups, it can be translated to new concerns as they arise.

The message cannot be overly optimistic. Building unreasonable expectations does only harm in the long run. Solutions to entrenched health issues are never quick or easy. Science takes time and may require multiple studies before a breakthrough is made. Health issues will never disappear, but they can become infrequent given sufficient scientific knowledge of the disease and a willingness among breeders and owners to do what is necessary to reduce disease frequency.

If the message is not clear or continually shifts focus it will not hold the attention of those who most need to hear it. And it can provide ammunition to our critics.

#### The Devil in the Dark

Purebred dogs are under assault from extreme animal rightists who would like to see all domestic animal populations managed to extinction. Some of the major extreme-AR players have mounted well-funded PR campaigns proclaiming all purebred dogs to be hopelessly riddled with disease. They market their views to large numbers of well-meaning but ill-informed people who have come to believe that every purebred is hopelessly riddled with disease while mixes and cross-bred dogs are uniformly healthy. This message resonates with a public that, rightfully, disdains those who cause needless suffering to animals. No matter the inaccuracies in the argument, it makes us look bad.

AR extremism is a purebred dog issue: The extinction they plan for all our dogs is the ultimate health problem. Genetic diseases exist in mix-breeds and whatever-doodles, too. But our detractors will grab onto breed health information and beat us about the head with it if we let them. A recent, scathing (and, in this writers' opinion, biased) BBC documentary on purebred dogs used individual animals with significant health issues to imply that all dogs of those breeds had horrible health issues and, by extension, all other purebreds as well. Our breed and all-breed organizations must stand ready to answer these accusations with facts. Have proactive health programs in place to demonstrate your dedication to improving health.

Our failure to adopt substantive and proactive health initiatives in some breeds and a lack of public education about what is being done in purebred dogs leaves us open to criticism. We need to work toward getting a more positive picture before the public and at the same time put our houses in order.

We may have to accept that some of our traditional practices, like cropping and docking, may need to be abandoned. These surgeries once served practical purposes but today are largely cosmetic. John Q. Public and his kids aren't buying long tradition as an acceptable defense. This gives the AR extremists a handy bat to bludgeon us with.

Breeds with extreme features that are associated with health issues are under attack already. I don't personally feel that any breed should be mandated into extinction. However, the breeders and breed clubs for those breeds may need to think long and hard about a return to historic standards of breed appearance dating to the time before high-quality veterinary surgery and professional dog groomers were available to maintain those breeds in their current state.

Health issues are our Achilles' heel. If we aren't diligently and publicly making efforts to

improve health, from the big all-breed registries down to the individual breeder, we will remain open to AR attack and the restrictive legislation it encourages.

# Hang together or hang separately

The harshest critics of purebred dogs and the Incorrigibles within our own ranks are adept at divide and conquer. It is vital that we who care about the future of purebred dogs work together, despite breed differences, never mind what registry we use, across international borders and without regard to the thousand and one political wrangles among ourselves. We must not tolerate Incorrigibles, who would prefer to keep hereditary disease in a closet. We must continue to build on the successes achieved so far.

To paraphrase Benjamin Franklin, we'd better hang together toward meaningful and substantive improvements in purebred health or the AR extremists will be only to happy to hang each one of our breeds separately. And extreme AR sensibilities aside, we owe no less than good health to our dogs.

# Trust but Verify The Advantage of Open Health Registries

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Susan, an established and respected breeder, mated one of her bitches to a stud owned by David, another established and respected breeder. She kept a male pup who turned into a top competitor and an excSusant sire. A few years later Susan started receiving reports that some of her male's offspring had cataracts. Susan had rarely experienced cataracts in her bloodline but, since it was a common breed problem, she had requested a copy of a current eye exam form on David's stud and had asked about eyes and other health issues in his line before she bred her bitch. David had denied having any problem with eyes.

Susan started talking to other breeders, trying to find out where the problem had come from. In the process, she heard rumors that numerous offspring of David's male either had

cataracts or had produced them. Some people claimed the dog himself had developed them when he was 6 years old. After more investigation she learned that some of these rumors were true. David had lied to her. When she confronted him, he refused to give her a current eye report on his dog and threatened to sue her if she pursued the matter further.

Susan was furious and frightened. David had put her breeding program and those of others at risk. His actions had caused avoidable misery to dogs and their owners. She knew remaining silent would only add to the problem, but what could she do?

The story of Susan and David is a composite of several incidents. Sadly.

scenarios like this have played out time and again in breed after breed. Concerned breeders like Susan work with each other and share information. They place ads or write articles aimed at educating other breeders. They may develop lists of dogs known to have had or produced a genetic disease. And sometimes one or more "Incorrigibles" like David force them to abandon their efforts. For many years, the culture enveloping purebred dogs has provided more support to the Davids among us than it has for the Susans.

Conscientious breeders lament that they can never be entirely sure that people they have depended on for information have been straight with them. This problem is especially difficult for those who are relatively new to a breed. Novices often ally themselves with mentor breeders while they are learning the ropes, but not all mentors are created equal when it comes to knowledge and honesty about genetic health issues. Even experienced breeders can be lead astray by people they thought they could trust. Fortunately for us and our dogs, things are beginning to change.

For twenty years or more, until his death in 2005, Dr. George Padgett, a veterinary pathologist and writer/lecturer on canine genetic health issues, has been telling breeders that if they want to control genetic disease they must start talking about it openly. Aware of the huge difficulties facing individuals in collecting useful genetic disease data, he and other scientists. veterinarians and breeders who were members of the International Elbow Working Group took a bold step. In 1990 they founded the Institute for Genetic Disease Control. GDC, modeled in part on the previously established open health registry operated by the Swedish Kennel Club, offered the first open disease registry in North America.

Initially, GDC tracked only hip and elbow data. As time passed it expanded to include eye exam results and a variety of breed-specific issues, taken on at the request of and in cooperation with concerned breed clubs.

Open health registries and published screening test results have been commonplace in some European countries for a number of years. All the Scandinavian countries and Great Britian have open health reporting for orthopedic, eye, and some other diseases plus

DNA test results. In the Netherlands and some other countries, breed clubs publish results of standard health screening tests. (The Dutch kennel club is in the process of establishing an open registry.)

Unfortunately, too many in the purebred dog culture in the US were indifferent if not actively opposed to the open registry concept. GDC remained largely unrecognized and little used. Within a decade of its founding it was at risk of shutting down. To preserve what it had accomplished, GDC turned most of its data over to the Orthopedic Foundation for Animals (OFA) in 2002.

Historically, the Orthopedic Foundation for Animals provided a closed registry that publicly listed only phenotypically normal dogs. During the 1990s, OFA came to recognize the importance of open reporting and initiated a "semi-open" system. They expanded their services from hip dysplasia and other orthopedic diseases to a broader range of health concerns. With the owner's permission, results of affected dogs will be listed along with identifying information about the animal, including the names and registration numbers of its parents and their own test status, if available.

OFA now registers screening results for hips, elbows, patellas, Legg-Calve-Perthes Disease, cardiac abnormalities, thyroid disease, congenital deafness and sebaceous adenitis. With genetic research leading to DNA tests for various canine ills, OFA will no doubt be adding other conditions to the registry in the future.

About the same time that OFA developed it's semi-open registry service, the AKC Canine Health Foundation initiated the Canine Health Information Center with a view to developing, in cooperation with breed clubs, an open source of genetic disease information on specific dogs CHF combined forces with OFA to bring CHIC on line. CHIC now provides information on almost three dozen breeds. It automatically receives all OFA open data and CERF normal data. Other information (failed CERF exams, PennHip reports, etc.) must be submitted directly by the owner of the dog.

When a dog has completed its breed's CHIC requirements, the owner will receive a certificate and the dog and its test results will be listed on the CHIC website. Minimum CHIC

requirements for Australian Shepherds are hips, elbows and eyes. Thyroid is optional. Other OFA registered tests (heart, patellas, etc.) which have been submitted and designated open by the owner would also appear in the CHIC record. Should additional tests be recognized as essential they could be added to the Aussie's CHIC protocol.

CHIC is voluntary. If an owner elects not to allow OFA affected data to be shared or declines to send in other kinds of information, CHIC does not pursue the matter nor are there any sanctions against those who decline participation. The hope is that as more people become involved peer pressure will bring some of the reluctant into the fold.

OFA and CHIC are not the only available open registries. In some cases, individual breed clubs or organizations in the US maintain their own programs. Bearded Collies, poodles and Portuguese Water Dogs are a few breeds that have single-breed health registries. These may or may not be open and may or may not be associated with the national clubs. Aussies in North America do not have any official disease registries outside the all-breed organizations.

European databases vary, but most record at least hips and eye screening results. In Europe, publication of results—even bad results—may be mandatory.

There is already a great deal of information available to the breeder who is willing to look for it. OFA and CHIC have easy-to-search on-line databases. The Canine Eye Research Foundation also has one, though it lists only phenotypic normals. With the open registries you can discover what dogs have failed a particular test, what results are recorded for a particular kennel and which offspring of a particular stud have results recorded. The OFA site is especially easy to search. Some European databases are also searchable, though you will need to understand the language or find a translator to help you.

Since the US databases do not list negative results without the active consent of the dog's owner, some information may not be available. While you cannot know the status of an unlisted animal, conspicuous absences (i.e. no hip listing for the 4-year-old stud, Ch. Bigwinner Last Year) can suggest questions you will want to ask if you are interested acquiring or breeding relatives of the absent dog. Beware sires or kennels that have many missing names for a screening test that should be standard.

Active participation in open registries needs to become the gold standard for serious breeders. Only through the open exchange of genetic information will we be able to make serious impacts on the incidence of genetic diseases in purebred dogs. Certainly the long and commonly held opinion that all Aussies should have their hips and eyes screened has made those exams near universal among breeders who wish to be taken seriously. Breeders who are discovered to have failed to do standard screening on breeding animals are risk being held up ridicule. They may find their dogs are avoided by their peers.

In a perfect world, everyone would willingly share information about genetic issues in their dogs. But the world isn't perfect and there will always be those who will not come clean. Listing with the open health registries should become just as much a hallmark of responsible breeding as the faithful screening of eyes and hips. If conscientious breeders make it a regular practice and actively promote the idea, others will follow. When they do, the Susans of the future won't have to rely on trust, they can verify.

### What ASHGI has done to help:

Since inception, the Australian Shepherd Health & Genetics Instituted has been an advocate for the open registry concept and the open exchange of health information on breeding dogs. To that end ASHGI has established the following programs:

Ten Steps to a Healthier Australian Shepherd Breed (10-Steps)
http://www.ashgi.org/home-

page/programs/10-steps International Directory for Australian Shepherd Health (IDASH)

http://www.ashgi.org/openhealth-database-search/idash

# **Testing Ground Hosting a Health Clinic**

First published in Double Helix Network News, Summer 2011

Health clinics for genetic disease screening are one of the most valuable services a dog club or other motivated group can offer to purebred dog breeders. Holding a clinic, especially one that offers multiple types of screening, makes testing convenient and economical for dog owners. Test results are important tools for making informed breeding decisions. Holding a clinic might seem like a daunting task, but with a little planning and organization anybody from an individual breeder to a national club can host a successful event.

### Proper Prior Planning...

One of the best ways to make any event successful is to lay out a plan of action well before even the first deadlines loom. For a small event you can start the process a few weeks ahead of time; a large one with multiple offerings might require that you get your dogs in a row four to six months out. If you write your plan down and keep a copy, it will be there to guide you the next time you host a clinic or when you pass the responsibility on to someone else.

There should be one individual in charge of the event. If that's you, you may be able to handle everything for a small clinic yourself. However, for a large multi-offering event you're going to need a team of key volunteers. However many people you involve, your team's initial chore is to determine the what, where, and when for the clinic.

# First things first

Everything else hinges on what you will be offering at your clinic. Offerings may include one or more of the following:

- X-rays for orthopedic conditions
- Eve exams
- BAER tests for hearing
- Semen collection
- DNA health screening
- Registry DNA parentage evaluation sampling
- DNA bank sample collection
- Research sample collection
- Anything else that may be useful to participants

Once you know what you plan to offer, look at possible dates and operation times. You might want to start with two or three possible dates and whittle down the list as you identify conflicting events and verify availability of facilities and key personnel. A major competitive event will draw a ready-made pool of participants if you can piggy-back your clinic health. In this case, the date and time will be determined by the other event's venue and schedule. A small, simple clinic will require only a few hours operation, but a large one held at a national specialty or other major event might extend over several days.

#### Human Resources

There are a plethora of tasks to complete before, during and after any clinic. The "personnel roster" that follows describes key volunteer positions for a large clinic. For smaller events use the list as a guide and combine areas of responsibility to suit your needs.

Operations guru – The go-to person for all things

clinic: Puts a team together, supervises preevent planning, and directs the clinic itself Lab liaison – Point person for all communication with labs and research groups Bean counter - Handles all money matters Paper shuffler - Performs all non-financial clerical tasks Volunteer wrangler - Recruits and oversees warm bodies to help on event day Publicist - Creates and distributes all promotional items Web Geek - Responsible for website/page, Facebook, Twitter, etc. Supply guy/gal - Acquires, stores, and distributes all necessary clinic supplies Logistics wizard - Gets all clinic materiel where it needs to be when it needs to be there The Pros - Veterinarians, animal health technicians, and any other specialists needed.

Location, location, location

Decide where to hold your clinic. The venue needs to have adequate facilities and be conveniently located for participants.

Every event will need a check-in and waiting area which may or may not be in the same room as the exams/sample-collection. If you plan to offer x-ray-based screening you will either need enlist the cooperation of a local veterinarian or find an examiner who has a portable unit. If the latter, you must find a space that meets the vet's specifications. Eye exam require a room which can be made very dark and is big enough to hold an exam table, a small work table or desk, a chair or two, and two or three people. If you will be collecting blood samples, you'll need one or more exam tables with adequate work space around them plus a supply staging table and someplace to safely stow completed samples. Cheek swab sampling can be done almost anywhere you can position a supply table and a few chairs plus one or more additional tables on to dry the swabs.

For single-day events a veterinary office works well A vet's office already has a waiting room, check in desk, exam rooms, adequate parking, and, if needed, x-ray facilities. The down-side is you will probably be limited to scheduling your clinic on a Sunday or after hours.

A caution about outdoor events: Make sure the actual collection/exam area is under cover. (Rain, snow, sleet, and/or hail are sure to fall if it isn't!) Make sure any outdoor venue has or can accommodate adequate sample storage facilities to prevent sample damage or contamination from things like wind, loose dogs, and rampaging children.

Make sure you reserve your space; multi-use public facilities are in high demand and may be booked well in advance. If you are doing a clinic during another event, make sure the event organizers are willing to have you there, know your needs, and will make adequate space available to you at the times you need it.

A final note on facilities: Have a Plan B in case something goes awry. Identify another nearby place which can be used in an emergency. Depending on the situation, you may need to cancel. If that happens, if at all possible you should have a way to notify participants ahead of time, particularly if some of them may be traveling long distances.

Necessary stuff

Put together a list of necessary supplies and equipment. You'll need adequate numbers of tables and chairs for staging supplies, a check-in station, waiting area, and exam/sample collection area(s.) Many venues will provide these, but you still need a count so they provide the right number. If you will have veterinarians or other professionals performing exams, they will probably bring their own supplies, but check to see if there is anything they expect you to provide.

If you will be collecting blood samples stock up on alcohol wipes, syringes with 20 gauge needles, and an adequate number of blood tubes. You'll also need coolers and cold packs. (Hint: Bags of frozen peas are a cheap, easily obtained, lightweight "cool pack.) If a wash basin won't be available, get hand sanitizer so sample collectors can clean their hands. This is especially important for those collecting with cheek swabs because they are working in the dogs' mouths.

For cheek swab items, be sure you obtain sufficient kits from the lab offering the test. If the lab does not issue kits, be sure you have an adequate supply of cytology swabs on hand. Finally, it doesn't hurt to have a big box of dog cookies to reward the victims after their ordeal.

A suggested supply list for a large, multioffering clinic can be found in the appendix at the end of this article. Use it as a guide to develop your own list.

#### Time is of the essence

Create a timeline to make sure things get done on time. This section lays out key tasks and when they need to be done for each of the key volunteer positions described under "Human Resources," above. You can combine or split tasks to suit your event. All the key volunteers should have their assignments no later than 90 days before the event.

# Operations guru

90 Days out: Obtain detailed sampling and handling instructions for each clinic item from the Lab Liaison. Study them and prepare a notebook with all pertinent information for on-site reference during the clinic. (Once prepared, this notebook can be retained for future clinics, though it should be reviewed at least once a year and updated as necessary.)

Pre-registration and payment can save time and effort on event day. Decide whether you will offer this convenience and coordinate with other key volunteers to implement. If preregistration is offered, establish a cut-off date no later than one week prior to the event.

Determine whether your group will accept samples collected elsewhere. (Note: It is better that you don't. See "Handle With Care!" below.) Will your group be providing photocopying at the event for those who forget to bring copies of required documents. (Hint: Not is the better option, here, too. You'll have plenty to do without this.) Provide policy statements on these two items to the Publicist and the Web Geek so attendees will have prior notification.

Throughout the pre-event period, keep in touch with and assist other key volunteers. Trouble-shoot problems and facilitate communication between key volunteers if necessary.

If a club is hosting the clinic, discuss with the board whether or not they wish to underwrite a portion of the costs on some or all of the items. Price underwriting encourages participation and benefits your breed. If the club does decide to underwrite, try to make prices come to even dollar amounts - it'll be easier to make change during the event. Notify the Bean Counter and the Web Geek of any changes in pricing.

Serve as point-of-contact for any veterinarians or other professionals who will be offering their services at the clinic. See that their needs are met.

One Week Out: Visit the clinic site during this week to inspect the space. Make plans for how you will arrange check-in and collection/exam areas with an eye to traffic flow and efficient operation. If your supplies are being shipped to a drop-point, verify that they have arrived; have contingency plans ready if they have not.

#### Lab liaison

90 Days Out: Contact all research groups and testing labs connected to clinic offerings to provide them with clinic dates and determine current prices and whether bulk discounts are offered. If a lab/research group provides sampling kits, order those at this time. Advise the labs to ship no later than a month before the event (this gives you time to trouble-shoot if necessary.) Obtain instructions and current forms for each offering. Give the instructions to the Operations Guru. If clinic info will be made available on a website, obtain pdf files of the forms for the Web Geek; you can

often download these from the lab or research group's website.

From now until clinic time, field all communications with laboratories and research groups. Consult with or advise Operations Guru or other key volunteers as necessary.

#### Bean-counter

90 Days Out: Create or update forms for recording monetary receipts. (Hint: If you can have a computer on-site, set up a spreadsheet so all you need to do is enter the information as it comes in.) If PayPal® is used for pre-registration and you don't have access to the account coordinate with whoever does so you will receive payment information in a timely manner and decide how you will handle any refunds for those who pay but later can't attend. Refund information needs to be provided to the Publicist and Web Geek. Decide what types of payment will be accepted (checks, cash, and/or credit cards) and advise other key personnel.

<u>60 Days Out</u>: Maintain an up-to-date record of all receipts and expenses.

1 Week Out: Get a change fund for event.

#### Paper shuffler

90 Days Out: Set up a log to record all clinic activity. At a minimum this should detail who presented dogs, how many, and for which offerings. The date payment was received and payment type (cash, check, etc.) can also be helpful, Prepare check-in sheets for each clinic item with columns for the dog's call name, the owner's name, payment type, and – for multiday events – date seen. If your group offers pre-registration, keep the clinic log current. Coordinate with the Web Geek or other key volunteers to collect this information.

60 Days Out: if applicable, continue logging all pre-registrations as received. Send participants an acknowledgement notice verifying the item(s) purchased.

1 Week Out: Post any pre-registrations to the individual item check-in sheets; doing so will save time during the event. Make enough copies of these forms to accommodate walk-ups.

#### Volunteer wrangler

90 Days Out: Work with the Operations Guru to develop a list of warm bodies needed on clinic-day including someone to direct clinic traffic, one or more runners, cheek swab samplers (one person can do it but it is much easier if there are at least two at any given time,) two-person team(s) including an AHT or other qualified individual for blood sample collection, and – if required for any other offerings – veterinarians and AHTs, Once arrangements have been made with any veterinarian, refer him/her to the Operations Guru as the point-of-contact for anything they might need. Make sure the Operations Guru has the names, contact information and work assignments for professional participants. Work with the Publicist to develop recruiting announcements.

60 Days Out: Set up a work schedule for the event and start active volunteer recruitment. Get the contact info, including a cell phone number (useful during the event) for each volunteer. Determine their availability and assign a work shift. If you will be recruiting AHT's for blood sample collection, determine whether they have any special needs and communicate those to the appropriate key volunteers. Continue the recruiting process until a week out.

1 Week Out: Verify with all volunteers that they know when and where they will be working and what they will be doing.

#### **Publicist**

90 Days Out: Develop promotional and informational announcements; coordinate with other key volunteers for content. Put together a distribution list and schedule for any and all appropriate media. If your group cannot take credit cards at the event, make sure all your promotion includes that information. Encourage pre-registration, if offered.

<u>60 Days Out</u>: Initiate promotion of the clinic and preregistration, if applicable, continuing through the week before the event.

# Web Geek

90 Days Out: Prepare any needed webmounted material with input from other key volunteers and with the site's webmaster if other than yourself. A page should, at the very least, include date, time and location for the clinic, a listing of the clinic offerings, and a "contact us" for questions. You may also want to include a description of each item and its price. Have forms available for download and advise attendees to fill them out ahead of time.

If pre-registration is offered, provide an on-line payment option so you can take advantage of credit card payment. If your site doesn't have a formal storefront, PayPal ®

buttons are easy to install if your group has or can set up an account.

If your clinic will be a large multi-offering event, you might also want to provide some instructions. What paperwork must be presented? Is there any special preparation participants must make? Where appropriate, provide links to related web pages for research and other sampling/ testing offerings.

If Facebook will be utilized for publicizing the event, prepare and mount all necessary information there.

60 Days Out: Website and/or Facebook info goes live. Once they are up, monitor Facebook for comments and oversee the "contact us" e-mail. Answer questions or direct them to the appropriate key volunteer. Assist Publicist as needed with Facebook and Twitter.

One Week Out: If applicable, disable pre-pay option ASAP after the pre-registration deadline.

# Supply guy/gal

60 Days Out: If your group does not already have one, develop a supplies list with input from other key volunteers. (See Appendix for a suggested list.) Coordinate with other key volunteers to determine any special supply needs. Inventory left-over supplies from prior clinics and acquire anything needed.

30 Days Out: If kits have not been received, notify the Lab Liaison for follow-up to make sure you have them on time. Arrange with Logistics Wizard to get all supplies delivered to the site.

One Week Out: Get all supplies to the Logistics Wizard.

#### Logistics wizard

90 Days Out: Check with the Supply Guy/gal to determine what needs to be shipped to the clinic site. Get specific post-event sample shipping instructions for each lab/research group from the Lab Liaison. Check with the Bean Counter and Paper Shuffler to determine which post-clinic sample shipments must include a check. Make a check-off list to assure everything that needs to be shipped to one place gets included in a single package and sent to or from the clinic at the proper time.

60 Days Out: If needed, arrange for an incoming "drop point" on or near the event site. Identify shippers convenient to the clinic site for post-event shipping needs. Verify that they provide overnight service and that they will accept biological samples.

<u>1 Week Out</u>: Make sure everything gets delivered to the event site in a timely manner.

#### The Pros

Veterinarians and other professionals connected to specific clinic items will generally run their own show. They should be directed to the Operations Guru for anything they might need. AHTs or others who will be drawing blood should communicate their needs to the Volunteer Wrangler.

#### Handle With Care!

If your clinic includes cheek swab or blood sample collection there are a few important things to keep in mind. Every laboratory and research group will have its own specific requirements but here are some general guidelines.

Laboratories appreciate receiving properly collected and handled samples. Improper handling can render a sample useless. You must take the utmost care in collecting, storing, and shipping your samples. Not only does this ensure happy clinic customers, it helps you maintain a good working relationship with the labs and research groups.

Ensuring sample quality is the reason you should not accept samples gathered elsewhere and brought to you at the event. If you don't collect a sample, you don't know what dog was actually sampled or whether the sample was collected and packaged correctly. Nor can you rely upon people's assurances that they have the proper sampling kit. Labs can be picky about getting samples collected with another lab's kit. If the customer paid for the test and the lab rejects the sample, you may be the one stuck in the middle.

# Blood samples

Blood must be kept chilled between collection and shipping. Because temperature maintenance can be critical and qualified blood-drawers are in limited supply, if you plan a multiday event it might be best to hold the blood draw on a single day. Try to make use of a refrigerator for overnight storage and *do not* allow the blood to freeze. Blood needs to be shipped in a container that can be kept cold. Small, insulated beverage coolers are cheap and work well. If you cannot find cold packs, use bags of frozen peas. Pack any accompanying paperwork in a zip-lock bag so it won't get wet.

#### Cheek swab samples

Before sampling each dog, verify that it hasn't had food, water, or exposure to other dogs or toys for at least one hour prior to swabbing. This prevents cross-contamination with another dog's DNA. If someone brings a young puppy, make sure it isn't still nursing. Ideally, these questions should be asked at the check-in table.

Collect swab samples one dog at a time. Samplers should wash or sanitize their hands between dogs. Swabs will need to be air-dried before packaging; select a place where multiple swabs can be laid out without touching. The drying area must be safe from jostling by people or dogs. If your event is outdoors, protect the swabs from wind. Make sure the paperwork does not get separated from the associated swabs. If swabs are to be shipped back in their individual sleeves, fasten the flap on the tube *loosely* with a piece of tape. WARNING: If you seal swab sleeves tightly, moisture trapped inside can spoil the sample.

When the swabs are dry, package them and related paperwork as per the instructions of the lab to which they will be sent. Prior to shipping, store completed swab/form sets in a box kept safe from traffic and moisture. Have a separate box for each lab's completed samples and forms.

If the clinic is held in humid conditions it may be necessary to spread out completed swab envelopes indoors for a few hours to get them as dry as possible. DO NOT ship swabs in plastic containers or bubble-wrap envelopes as these trap moisture.

#### Clinic Day!

The Operations Guru and anyone else who will be helping with set up should arrive before the event (at least two hours for a blood draw, one for other items) to make sure all tables, chairs, supplies, etc. are in place. If at all possible, set up the day before. All other key volunteers who will be working the event should arrive no later than a half hour before opening. Day-of-event volunteers who have been assigned work shifts should arrive 15 minutes before their scheduled time so they can receive instruction and for a smooth transition between shifts.

The Lab Liaison, Publicist, Logistics Wizard, and Web Geek do not need to be

present during the clinic unless they have other duties at that time.

# Supply guy/gal

Collect supplies from the drop point, if shipped. Deliver them to the clinic site at least an hour before opening. Open supply containers and distribute all items to the appropriate work areas before the clinic opens. If the clinic will run multiple days and the set-up is not in a secure (i.e. indoors, locked) area, pack up and remove all supplies at the end of each day and bring them back at least one hour before start time the next morning. If something is running low or somebody needs something, go get it. At the end of the event, pack up all leftovers. Coordinate with the Logistics Wizard for transport to the place they will be stored.

### Operations guru

Supervise and direct all clinic volunteers and assist in sampling dogs. Oversee sample handling, labeling, and short-term storage (prior to shipping). Once the clinic opens, make sure all samples are handled properly to avoid contamination. Verify sample labeling for accuracy and compliance with lab requirements. Oversee proper air drying of all cheek swab samples and refrigeration of blood samples.

For multi-day clinics, at the end of each day assist the Paper Shuffler to check all samples collected against the clinic log and check-in sheets. Secure all samples overnight in a place where no unauthorized person will have access to them and where there is no possibility of contamination. Verify that the Bean Counter has balanced the day's receipts against the clinic log.

When the clinic is over, assist the Paper Shuffler and Bean Counter with a final over-all inventory of samples and an audit of at-event receipts and expenses.

#### Paper Shuffler

Oversee all on-site paperwork, including verifying that forms submitted are properly and legibly filled out. Run the check-in table. Verify any payment required with the Bean Counter. If the dog will have a cheek swab test, verify that it has had no exposure to foreign DNA. (See "Handle With Care!" above.") Assist Bean Counter with daily audit of receipts and expenses.

Bean Counter.

Maintain control of the cash fund and all monies received during event. Record receipts for walk-ups. If the event is multi-day, secure the money box overnight. With the assistance of the Paper Shuffler, audit receipts and expenses daily.

If you do not have check writing authority for the host group, provide the responsible party with a list of any checks (payee and amount) needed to go with sample shipments. if appropriate, prepare and make the end-of-event deposit and give the receipts record to the appropriate individual. If you are doing a large multi-day clinic and you receive considerable cash it may be prudent to make a few interim deposits or convert the cash to cashiers checks.

### Volunteer wrangler

Make sure all scheduled volunteers remember their shifts. Fill in for no-shows. Dragoon extra help if needed.

# The job's not over...

...until the paperwork, among other things, is finished.

#### Logistics wizard

Shipping samples to labs and research groups is job one: All samples should be shipped as soon as possible after the close of the clinic. Make sure checks for the labs are included in any shipments that require them. Provide the Lab Liaison with tracking numbers for all shipments.

Blood samples need to be cushioned when packed to prevent breakage. Place any accompanying paperwork in a zip-lock bag or other waterproof container. Samples should be shipped as soon as possible via overnight service. DO NOT ship blood on a Friday. If you do, someone, somewhere will leave the container sitting on a loading dock in bad weather until Monday. If you cannot ship the same day as the clinic or the morning after, make sure blood is properly stored and chilled until the earliest possible shipping opportunity.

<u>Cheek swabs</u> are less problematic but should reach the lab in a maximum of 3 shipping days. Ship them at the earliest opportunity after the close of the clinic.

Bean Counter

Your top priority is to make sure the Logistics Wizard gets any checks that need to be included in the sample shipments. Provide the club treasurer or other appropriate party with copies of all clinic financial records and a listing of any checks written including check number, date, payee, and amount. After the event, redeposit the cash fund. Turn expense receipts and a record of all income and expense over to the host organizations treasurer or other responsible party.

### Lab Liaison

Follow up with the labs/research groups to make sure all samples arrived in good order. (Don't depend on the shipping company's online tracking information, alone.) Trouble-shoot any problems. Being on top of shipping issues helps you build a good relationship with the labs.

# Volunteer Wrangler

Send thank-you notes to all volunteers and professionals who took part in the clinic. If applicable, make sure the club newsletter editor and webmaster receive a list of volunteers so they can be thanked in those media.

#### **Publicist**

Send out a "thank you" message to clinic participants and announce clinic totals. If the host organization is a club, send copies of the announcement to the newsletter editor and webmaster

#### Operations Guru

Verify that all key volunteers with afterevent duties follow through. Call a "post mortem" meeting a week or so after the clinic to discuss the event. Make note of things that worked and things that didn't for future reference. Serve refreshments and celebrate a job well done!

If your club or group hasn't done a clinic in the past, consider putting one on. This article tells you the things most folks have to learn by

trial and error. (The author has tried - and occasionally erred – at numerous clinics.) Putting on a clinic, particularly a big one, is a lot of work. However, the effort is well worth it.

> Clinics are a tremendous benefit to the dogs, their owners, and your breed as a whole. If you've already done a clinic, plan more and keep up the good work!

# Appendix: Supplies list

This is a basic clinic supply list. Add or delete items as needed to suit the specifics of your event.

Copies of forms for all clinic offerings Brochures or other hand-outs about clinic offerings

Cytology swabs, if not provided by lab(s) (for cheek swab items)

Clipboards, one for each clinic item #10 (business size) white envelopes for packaging individual swab sample sets Disposable tablecloths

Pens (for people to fill out forms)

Cash box (for change fund)

Note pads

Paper clips

Scotch tape

these)

Packing Tape

Stapler and staples

Small boxes/baskets for small supply items

Boxes for completed swab kits

Alcohol hand wash or wipes

Suitable shipping containers for blood tubes Sufficient 6ml EDTA (or other, as required) blood tubes (for blood sample items we (recommend you have at least 100 research groups generally do not supply

Cold packs (or bags of frozen peas) Suitable shipping boxes for cheek swab samples

Tables and chairs, if not provided Dog cookies