Genetics and the Working Border Collie

The following article was published by the United States Border Collie Club in 1995, just after the Border Collie was recognized by the AKC. At that time, the AKC stated that it would close its studbook to non-AKC Border Collies three years after it began registering the breed. In the end, it never did close the studbook to Border Collies registered with the working registries. However, this article is a good explanation of the effect that setting a conformation standard and breeding to it has on the working ability of the breed.

1. Basic Genetics

We all learned basic genetics in high school biology, but most of us who don't use it every day have forgotten the details, and those who remember it don't necessarily see the connection between Mendel's work and the practical questions of dog breeding. In order to understand the genetic basis of the USBCC's objection to AKC registration of the Border Collie, we may have to start back at the high school biology level and work forward.

In the first place, heredity, the information passed from one generation to another, is contained in discrete particles which Mendel called "genes." These genes are carried, somewhat like beads on a string, along microscopic bodies called "chromosomes." Genes are arranged in constant linear order along a chromosome; each has a specific "address" (called a locus) on a specific chromosome. Chromosomes occur in pairs, one from mother, one from father. Each member of the pair has, at the same address, the same gene, but possibly a different form of the gene (called an allele). There are probably close to twenty thousand genes altogether in the dog. The only ones that are important to us are the ones that have identifiable alternative alleles. In other words, the only reason we are aware of the E gene which causes black hair is by the existence of its allele, e, which, if present on both chromosomes, causes red hair. The vast majority of the genes that make up the whole dog are not even identified; at most, we deal with a few dozen.

2. Recombination and Linkage

Behavior traits which make up the Border Collie:

A. Eye
B. Crouch
C. Balance
D. Power
E. Speed and agility
F. Interest in moving targets

All come from different sources: A. setter/pointer; B. spaniel; C and D. unknown (maybe some older breed of sheepdog; E and F. greyhound/whippet. How did they all come to be assembled in the modern sheepdog?

During the production of eggs and sperm, the chromosomes are scrambled, and one of each pair is passed to each germ cell. There is no preference for those originating from, say, the mother, to be passed together as a group. Furthermore, actual genetic material of those chromosomes is regularly exchanged between members of the pair during the process, so that in the end there is no such thing as "mother's chromosome." All of the chromosomes carry genes that originated with both mother and father. This is the very important process called recombination.

The closer two genes occur along the linear order of the chromosome, the more likely they are to remain together in the next generation. In fact, we can actually "map" chromosomes in organisms where we can do large numbers of breeding tests, (fruit flies, mice, etc.) simply by measuring how often particular alleles of two genes remain together from one generation to the next. In extreme cases, where this "linkage" is as close as one molecule to the next, they may only separate in one case in millions. It is very rare, however, for two recognized, identifiable genes' addresses to be so close together.

What are the implications of recombination and linkage? First, it would have been impossible to assemble into one breed all the different genetic traits that go into Border Collie instinct if they were very tightly linked together. Suppose, for instance, that the spaniel has the traits "crouch; no eye" and the setter has "no crouch; eye." If the "crouch" gene were tightly physically connected to the "eye" gene, the two original combinations of alleles would still be together. We would have dogs which crouch, but have no eye, and dogs which have plenty of eye, but do not crouch.

If we couldn't recombine those alleles, we could never arrive at the "crouch; eye" combination we treasure in our working dogs. The original litters in which spaniels and setters were crossed with old-fashioned sheepdogs, and the next few generations as well, must have included the whole spectrum: crouch, eye; crouch, no eye; no crouch, eye; and no crouch, no eye. From those, the ancestors of the Border Collie were selected: those which had eye and crouch together. The more completely random the recombination among these genes, the easier it would have been to get these new combinations.

The flip side of this, though, is that now that we have assembled within our breed the combinations that we want, it is just as easy for them to come apart in future generations if no effort is made to hold them together by selective breeding.

3. Fixing a Combination

Genetic combinations are said to be “fixed” within a breed if every member of the breed has exactly the same combination of alleles at a particular locus. In the Belgian Sheepdog, for example, the black color gene has been “fixed”; no other alleles of the E gene are present within the breed. A single gene of this sort is easy to fix. We simply remove from breeding all individuals who either are the wrong color or ever produce puppies who are the wrong color. It is also possible to make progress toward fixing the normal allele of a gene which causes a genetic disease by such selective methods.

Generally speaking, the Border Collie does not have any fixed genetic combinations. In appearance, this is obvious. Even in herding, however, there is a spectrum in almost every one of the traits we value. A dog may have very little eye, or so much that he freezes (like the pointer who is “staunch on point”) and cannot be brought to move the stock; he may have no balance at all, or such a strong sense of it that he cannot be persuaded to move off the balance point; he may lack power and be unable to move even the lightest stock, or he may have so much that sheep flee from him the moment he comes onto the field. In all of these, the combination that produces the middle ground is the most desired; it is also the most difficult to fix genetically. After all these generations, a litter of the best-bred working Border Collies still may have perfect working dogs, mediocre dogs, and useless dogs.

Alleles are not always dominant or recessive; often the combination in which the two alleles are different (heterozygous) is intermediate between the two homozygous combinations. Some of our most desired genetic combinations in herding behavior may well be heterozygous pairs: they can never be fixed in a population because they will always produce some of each type of homozygous pair.

Because these complex traits cannot be fixed in the breed at their optimum levels, because the alleles and combinations of alleles that produce mediocre and useless dogs still occur in the breed, the risk of losing the best combinations is always at hand. If every Border Collie carried the precise combination that produced a perfect worker–for light sheep or cattle, for trial or farm, for Arizona or Scotland, for the tough handler and the soft–we would have nothing to worry about. No matter which of our dogs were chosen to be bred for “conformation” showing, all of them would still be working Border Collies.

4. Complex Genetic Traits

The red Border Collie, like the chestnut horse, is the result of a recessive gene pair. In the case of the dogs, the dominant (black) is called B, and the recessive (red) is called b. A black dog may be either BB or Bb; the red dog is always bb. Two black dogs may have red puppies if both of them are Bb; the b can come from each parent to produce bb in the pups. Statistically, one out of four pups with such a cross will be red. If either parent is BB, though, the combination can’t

produce red pups. If both parents are red, bb, then all their puppies will be red; there is no B available from either parent to make a black pup.

The end product of most genes is some sort of biochemical substance. In the red color, the chemical is a pigment called eumelanin. This is one of a group of pigments, the melanins, which cause color in animal skin, hair, and feathers. It is responsible for very dark brown or black color. All Border Collies have in their hair a red version of melanin, called phaeomelanin. In the black dogs, the black eumelanin covers the appearance of the red. If you've ever looked closely at your black dog in bright light after he has spent a lot of time in the sun, you will see a faint red glow to his hair. The eumelanin has been bleached by the sun, and the red color is showing through, however slightly.

Only the dominant version of the color gene results in eumelanin production; if the dog has two copies of the recessive version, he will have no eumelanin. His hair will contain only the red pigment, and anywhere that he would otherwise have been black, he will instead be red. This means he may have the same variety of white markings as any black Border Collie; he may be tri-color, with lighter brown markings in all the usual places; he may even be a red merle instead of a blue merle. His nose and toe pads, which would be black on a black dog, are red-brown.

But why are there suddenly more of them? One reason is that the red gene is present in some of our favorite breeding lines. The first recorded red Border Collie was a bitch named Wylie, grandmother of the famous Dickson's Hemp (153). The recessive gene passed through the generations to J. M. Wilson's Cap (3036) who appears in the pedigree of Wiston Cap at least 16 times! Wiston Cap carried the red gene and passed it to many of his sons and daughters. Our current Border Collies tend to have many crosses of Wiston Cap in their background; each one increases the chances of receiving that e gene. Crosses on both sides of the family, likewise, increase the chances of a double dose and the appearance of more and more red dogs.

Unlike most simple genetic traits, however, good (or bad) hips don't result from a single pair of genes. With a single pair, like the "red" genes, the probability of each genetic combination in the next generation is easy to measure. We know exactly how many red pups, statistically, to expect from any combination. With hip dysplasia, on the other hand, we have no idea what to expect in a litter of pups, even if we have x-rayed both parents.

Instinct and behavior, like hips, are affected by a large number of genes; some may be recessive like the b; some may be dominant like the B. The problem is that we don't know how to identify any of them, and we have no idea how many there are. If we had some kind of behavioral measurements on all the members of hundreds of litters and their parents and offspring, we could make a start. We aren't even close. We have no real measurements at all; our assessment of herding ability is subjective, and deals with the whole dog and his ability to get the job done.

Studies by Scott and Fuller on spaniels revealed the genes involved in the behavior known as "crouch"; the crouch itself is controlled by two major genes, with the crouch (or sit) dominant over the stand. The quiet attitude was also controlled by two genes, with the quiet behavior recessive to the more active. The whole pattern of quietly crouching, then, results from four genes altogether. The number of different genetic combinations that can be formed from 4 (2-allele) genes is 81! If the parents are heterozygous for all four of these genes, any of these genetic combinations is possible in the same litter.

What does all this mean to the Border Collie? Imagine, if that simple quiet crouch behind the sheep depends on 4 separate genes, what must be involved in the entire collection of herding behavior: eye, balance, power, biddability, etc. And what must the chances be of accidentally combining the right factors to remake a herding dog, if those combinations are ever lost?

The complexity of the genetics of behavior is probably not a surprise, but it is the basis of the entire argument that the performance dog must be bred for performance at every generation. The more genes are involved, the more different combinations are possible, the more easily they become separated and lost.

If the dogs selected for breeding for conformation are not the ones with the best herding genes, the population will inevitably drift away from the wonderful performance combinations that have been selected in the breed for so many generations.

Because the extremely complex instinct that makes up the working dog is not fixed in the population, constant selection is needed, at every generation, to maintain the best combinations. Any relaxation of this pressure will result in the increase in numbers of those dogs which have less than ideal genetic combinations. Breeding for any other purpose without also selecting for truly high quality working genes will inevitably result in the dilution of the working instinct within the breed.

Genes may have more than two different alleles (there are 160 different alleles of the blood group gene, B, in cattle). The number of possible genetic combinations as we increase both the number of possible alleles and the number of genes rises abruptly. If we are dealing with, say 6 traits (see above), each with an average of only two gene loci involved, that is 12 genes; if they have only 2 or 3 possible alleles:

"With even moderately small numbers of polymorphic genes, the number of genotypes that can be produced by recombination is likely to be greater than the existing number of individuals in the species." (V. Grant, in The Origin of Adaptations).

If we are already trying to maintain some genetic equilibrium with all these possibilities, consider the additional burden of selecting at the same time for color, ear type, body size, coat quality, eye color, head shape, etc., to fit some arbitrary standard of appearance. The number of

different genetic types becomes astronomical. The number of dogs that can fit all these separate standards for all these separate genes is, as Verne Grant stated it, probably less than one in the whole breed! The number that will come close is still very small. This is particularly true within a breed like the Border Collie where there is no genetic uniformity in appearance to begin with.

5. Inbreeding

When the number of available dogs for breeding becomes very small (as, for instance, those dogs who fit both a performance standard and an appearance standard), the result will be, unavoidably, an increase in inbreeding. Furthermore, the best, and indeed the only way to fix a set of alleles within a breed is through inbreeding. Members of the same family tend to carry the same genes, so breeding among them is the quickest way to "concentrate" those alleles we want. Concentration is a poor word, although it is the one usually used here; we can never have more than two copies of the same allele–a gene pair–so we cannot concentrate any further than that. What we can do is get more and more genes to be present in identical pairs, in other words, "fixed" within the breed. This condition of having identical alleles in a gene pair is called homozygosity. And it is exactly the increase in homozygosity that is the problem with inbreeding. In addition to producing a predictable appearance in your litters of puppies–if that is somehow a valuable thing–homozygosity does other things which are downright dangerous.

Most (although not all) detrimental or even lethal genes are recessive (take CEA as an example), that is, they must be present in a homozygous pair to have their effect; increasing homozygosity leads to increasing the number of individuals with the damaging gene pair. For rather more complicated biochemical reasons, a general increase in homozygosity also leads to a decrease in general vigor and a greater sensitivity to stress–all in all, less healthy animals. Geneticists generally agree that genetic diversity within a population is always desirable. The more traits we try to fix in a breed, such as both performance and appearance, the less diversity we will have.

6. Drift

There is a mathematical formula called the Hardy-Weinberg law which predicts that in a random-breeding situation the frequencies of different alleles will remain constant. The key is in the concept of "random" breeding; every male in the population has an equal chance of mating with every female and vice versa; every mating is equally productive. Variations in these constraints cause change in the frequencies of genes, and in the number of individuals with different genetic traits.

One variation from randomness is viability. One is statistical sample size. With domestic animals, the most important is selective breeding. The fact is that among purebred dogs, there is no such thing as random breeding. It is the intention of breeders to change the frequencies of important

alleles. In the performance of this function, breeders take advantage, quite correctly, of the ability of a single male to impregnate a large number of females. In animal breeding, a relatively small fraction of all the available males produces a very large fraction of all the offspring. The choice of those male dogs determines the future of the breed. We can all point to the presence in our pedigrees of such great stud dogs as Wiston Cap, Gilchrist's Spot, Dryden Joe, Whitehope Nap, Welsh's Don, etc. In the case of the Border Collie, these central dogs have all been great herding dogs in their own right, and great sires of herding dogs as well.

The recent increase in the number of red dogs is a direct result of this system of breeding, arising from the popularity of Wiston Cap. This increase has been purely accidental, called "genetic drift," and is typical of the change in genetic makeup in a population when mating is not random. The presence of large numbers of dogs with genetic defects–CEA, hip dysplasia, epilepsy, etc.–in any breed usually results from this sort of accidental selection. A popular stud dog carries genes, usually recessive ones which are not expressed in his own life, which are passed on in increased numbers to the next generations simply because he produces a disproportionate number of pups.

Consider what will happen when the most popular stud dog is one who merely fits the appearance standard and so wins a show ring championship. He may or may not carry the particular, very complex combination of genes that makes a great herding dog. He may even be a good herding dog. If he contributes his genes to a disproportionate number of puppies, and if he is anything less than a great herding dog, his genetic contribution will lower the general level of herding ability within the breed. The extremely wide variety of appearances in the existing breed means that only a small number of our present herding dogs will be used in the breeding of showring competitors; at the very least, the genes in those great herding dogs who do not live up to the appearance standard will be sacrificed.

7. The Standard

The existing Border Collie is not a breed without a standard. It has a very specific standard, by which dogs without registration papers and pedigrees can be Registered on Merit if they can demonstrate their herding ability to satisfy this standard. Whatever appearance standard is designed by the AKC and its chosen Breed Club (should it eventually designate one), it will not be the same standard to which the breed currently strives; it will therefore, by definition and unavoidably not be the same breed of dogs.

Even though the initial registration will come from the existing breed, the next generation of "showdogs" will have been bred under a different set of selective rules, and will already be at least philosophically different. After three years, when the AKC closes its books and no longer

allows dogs of the original breed to be used for breeding, the AKC breed will have become a separate entity, no matter what its name!

This already happened at least once, when the "Lassie" collie was created. The working sheepdogs used to be called "collies." They became "Border Collies" to distinguish them from the developing show breed. At the time of separation, there was no real distinction; anyone can tell the two breeds apart now.

All of this is quite apart from the possibility of a standard being chosen which is simply inconsistent with the demands of the shepherding life. This may be in the written standard or in the fashions of judges who know nothing about these physical demands. This has already happened to some of the breeds (Labrador retrievers, for instance, are currently too heavy and short-legged to be of much use in the field; Siberian huskies tend to be showring winners with legs too short to run properly and with fluffy coats that cannot shed snow and ice; bearded collies look nothing like their ancestors, and have coats which obscure their vision, and collect burrs and mud). There has been some call for the USBCC to become the breed club so that we could set the standard and thereby avoid the problems of inappropriate physical traits being used. Unfortunately, although the problem will be made worse by the "wrong" standard, it is the existence of a physical appearance standard, and not its details, that is the danger. The currently proposed standard is flexible enough to appear to cover many of our dogs. In practice, however, an appearance standard, however broad it may seem, will subject the breed to all the problems listed above.

Although there is a popular belief that a dog that looks like his father (or mother) will work like his father (or mother) this is simply not necessarily true. Because of recombination of genes, it is no more likely that the pup with his father's markings is going to behave more like his father than the pup with completely different markings. If we were to set the show standard to duplicate in every detail the appearance of the latest International Supreme Champion, this would no more guarantee us a working breed than any other conformation standard. If we don't choose the pups that work like the latest Champion, we are not selecting the right genetic blend from the many possible combinations.

8. What Is A Breed?

As was stated in the USBCC Spring Newsletter:

"To a geneticist, a breed is simply this: a population of animals whose breeding is controlled and outcrossing limited, so that genetic selection can be exercised on it. . . . A population is simply a subgroup of the whole species of dog, Canis familiaris. Controlled breeding and limited outcrossing make it possible to select . . . for whatever genetic traits the organized breeders decide on. Organized breeders is almost a necessary part of the definition; one breeder cannot

produce enough dogs to truly create a breed, and a lot of breeders going in different directions will never produce any sort of directed selection."

Currently, we have several registries, here and abroad, organizing the Border Collie breed and directing its selective breeding. They all communicate with each other, their breeding goals are the same, and dogs move freely from one registry to another, so that they are effectively a single genetic population. From the moment the AKC closes its books on the breed they have derived from the existing Border Collie, they will have created a separate genetic population, on which new selective rules will apply. Whatever its origin, and whatever the standard of selection (even if it were to be a performance standard!) this new breed will inevitably begin to differ from the breed registered by the existing registries. It will not be a Border Collie.

Reproductive isolation, the genetic separation of one group of breeding animals from another, cannot help but result in two distinct "gene pools," and thus two different breeds. In natural selection–evolution–this is the path to the formation of separate species. In artificial selection, it is the path to the formation of separate breeds. Even with a very similar standard of performance, two genetically separate populations will eventually diverge simply by the effect of drift, the accidental change resulting from the use of a few sires to produce large numbers of pups. In fact, there can be no other reason for the AKC to close the books and prevent future entry of dogs registered with existing Border Collie registries but this: to create and perpetuate a separate genetic population, i.e., a separate breed of dogs.