The Road to Hell: Epilepsy and the Australian Shepherd

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The following is a seizure log kept by the owner of an Australian Shepherd with epilepsy.

Aug. 25  9:00 am  grand mal seizure
Sept. 9  1:45 pm  grand mal
Sept. 10 3:30 pm  grand mal
        6:10 pm  grand mal
        9:10 pm  grand mal
        10:40 pm grand mal
Sept. 11 1:25 pm  grand mal
        3:30 pm  grand mal
Nov. 2   5:20 am  grand mal
        7:20 am  grand mal, semi-conscious
        12:00 noon grand mal, hospitalized
Dec. 17  9:00 am  grand mal, Primidone increased
        1:00 pm  grand mal
        6:10 pm  grand mal
        7:20 pm  grand mal, preceded by running fit. Admitted to emergency clinic, continued seizing overnight and 3 more next day. Picked up 8am.
Mar. 18  12:15 pm grand mal
        3:15 pm  grand mal
        4:45 pm  grand mal, 1.75cc extra potassium bromide
        6:20 pm  grand mal, .5gr phenobarbitol
        7:05 pm  grand mal, 5mg Valium
        7:20 pm  grand mal
        8:20 pm  grand mal
        9:35 pm  grand mal, another 5mg Valium
        10:25 pm taken to emergency clinic, seizures continued all weekend
Mar. 21  8:00 am  picked up unconscious and on IV, taken to [regular vet]
Mar. 22  Unable to control seizures
Mar. 23  Unable to control seizures
Mar. 24  am  Seizures controlled only with toxic levels of phenobarb and potassium bromide. Released, told by two vets if seizures reoccurred there was nothing further to be done.
Picture an elderly woman losing her beloved companion to this horror. Picture a child witnessing his best buddy contorted with seizures. Picture yourself and whatever dog you hold dearest to your heart going through this hell. Is this what you want for our breed?

Slightly over a decade ago the Australian Shepherd community started seeing the first signs that epilepsy might be a concern in the breed. Those signs have expanded to billboards we continued racing in pursuit of other interests. Today, few breeders can assume their lines are free of this terrible disease. The road we took from there to here is of our own choosing, but it isn't too late to exit this road to hell. We can get this disease under control so long as we have the will to do so.

History lesson

Epilepsy is a common canine ailment. Sporadic cases occur in mongrels and every pure breed. When a related group of purebreds are diagnosed, it may indicate a serious problem for that line if not an entire breed. In the early 1990s several dogs in the United Kingdom were diagnosed with epilepsy. Aussies were relatively new outside North America. As is often the case when a breed is introduced to a new country, the first dogs in Britain were related.

Initial reaction in the UK was not constructive and foreshadowed things to come on a worldwide scale. Major players immediately adopted a defensive posture, attempting to silence those who spoke openly through threats and coercion. A core group of dedicated fanciers would not be intimidated. Faced with a small, interbred population and severe import restrictions, those who stood by the breed cooperated to limit the spread of epilepsy. They shared pedigrees of affected animals, collected samples for research, and imported new bloodlines. Meanwhile, those of us in North America remained indifferent, dismissing the UK situation as a tempest in a far away teapot: Their problem, not ours. But we were about to get a wake-up call.

In 1994 a memorial ad placed by Pat Culver, a pet owner who lost a young bitch, appeared in the September/October issue of the Aussie Times. Like most memorial ads, it had a picture, the registered name of the dog and her dates of birth and death. Unlike most memorials, it stated the cause of death and gave two generations of pedigree. The seizure log that heads this article was recorded by Culver.

The ad created a short-lived stir. Predictably, some breeders with closely related dogs made it know to Culver and others that they were furious. There was nothing wrong with their dogs and that Culver's bitch had not had anything hereditary. Other people discussed the need for action and sharing of affected pedigrees. Culver, the author, and Ann DeChant, a breeder who had multiple cases of epilepsy in a litter not closely related to Culver's bitch, tried to rally people, urging them to take the threat of epilepsy seriously. After an initial flurry of response, interest waned.
As time passed, more and more Aussies were reported with epilepsy in the United States, Canada, Australia and several European countries. Breeders, and especially stud owners, denied that dogs were affected, denied that epilepsy was the cause of the seizures, and denied that the genes could have come from their side of the pedigree. And the band played on as they continued their self-destructive waltz.

Meanwhile, epilepsy was attracting notice outside the breed community. A veterinarian who's connection to Australian Shepherds was limited to what she saw in her clinic remarked that whenever the topic of epilepsy came up in a gathering of vets Aussies were "always" one of the breeds mentioned. Several years ago, two research groups looking at canine epilepsy, VetGen and the University of Missouri, included Aussies among the breeds they would investigate. Response on the part of the Aussie breeders has been spotty, impeding their efforts to locate the gene or genes responsible.

Today, epilepsy rivals cataracts for the dubious honor of the Australian Shepherd's most common hereditary ailment. The show lines are riddled with it and at least two non-show lines are also affected. Given the silence that surrounds the disease, there may be more. In a recent discussion about epilepsy, one breeder remarked "I had a newcomer to the breed, a friend who owns a very nice bitch, sitting next to me during BOB, and she kept asking me about dogs she saw that she thought she would like as potential studs...I was amazed to find myself telling her quietly, 'No, he's produced epilepsy...no, he has epilepsy tight up behind him...no, he's too closely related to the other one you just asked about...'' One dog after the other, all the dogs she asked about had epilepsy issues that I knew about. The poor woman was stunned, I think. I have to admit, I'm appalled at the risks so-called reputable breeders take. But nothing much surprises me anymore. It's sad."

Claire Gustafson, another breeder that has experienced epilepsy in her dogs, remarked, "If you think it doesn't affect your lines, you just aren't talking to the right people. This isn't a disease that can 'magically' go away in three or four or five generations, and many of today's modern show pedigrees will span the breeding programs of several different people (not counting the numerous people who may indirectly be involved through ownership of siblings, half siblings, aunts, uncles, cousins, etc.) There are virtually no pedigrees left that don't present some level of risk through a carrier or affective relative--it's simply a matter of talking to enough people to find out where the risk lies. You need to form breeding strategies today as though you and every dog or bitch you breed to already have carriers in their pedigrees, because odds are, they do."

**How can something like this happen?**

Part of the problem stems from the nature of the disease itself. It has long been called "idiopathic epilepsy." Idiopathic means "of unknown cause." The term is used because there is no positive diagnostic test for epilepsy; it is diagnosed by ruling out all other reasonable causes. [see Dr. Margaret Muns' article, "Canine Idiopathic Epilepsy" for information on the diagnosis and treatment of epilepsy.] Today, virtually all veterinary authorities agree that "idiopathic" epilepsy is inherited. Perhaps the disease would better be named "familial" or "primary" epilepsy.
Another contributor to our situation is the Ostrich Syndrome, a term I coined two decades ago to describe the steadfast refusal to recognize unpleasant genetic facts.

"It's something in your water."

"You gave him a vaccination."

"She got stung or bitten by something."

"You're feeding the wrong food."

"The garden shed was open, he must have got into something."

"She hit her head."

And so on...

One of my all-time favorites is "heatstroke." Severe hyperthermia can induce seizures. However, the dog in question was working livestock in sleet. I doubt there is any working line Australian Shepherd so over-coated it can get heatstroke in that kind of weather.

There are many things that can cause seizures. Thorough veterinary follow-up is required so the dog can receive the proper treatment. While the excuses above do indicate other possible causes, most are unlikely to cause long-term repeated seizure episodes, would have additional symptoms, or they could be identified through testing.

Having run hard up against the Ostrich Syndrome, breeder Dusty Craig had this to say: "I always loved CA's term for breeders who couldn't bring themselves to admit to problems. When it comes to such a rotten problem as epilepsy, some of the biggest names in our breed bring the term new meaning. Continuing to bury problems and heads in the sand will only insure that there will be no clear lines. The Aussie will be lumped with other genetic nightmare breeds--one to stay away from because of all the problems. You laugh now, but when your bitches produce more problems in a litter than puppies and your stud dog becomes radioactive waste to other breeders, you will be wondering how this could have happened. It didn't have to. Sharing information rather than spurious witch hunts or passing unfounded rumors could have given you the knowledge you needed to make careful, informed breeding decisions that would have been for the good of the breed. What a concept, eh?"

The "idiopathic" label has played directly into the hands of those suffering from Ostrich Syndrome. Since there is no positive test, a dog with recurring seizures surely must have something else. No matter what tests have been run, there will be something Ostriches can point to that wasn't done or can't be followed up on allowing them to exonerate their dogs' genes. Something that often escapes the attention of Ostriches is the fact that several of those other diseases are also genetic.
This aspect of deniability has also played into the hands of those who are not in denial at all. The Incorrigibles consciously cover up and carry on as usual. Their behavior may be as simple as blowing-off anyone who brings a case of epilepsy to their attention or practicing some variant of "shoot, shovel, and shut up" with troublesome dogs. Incorrigibles with longevity in the breed or who have gained some degree of status have been known to abuse their positions to intimidate or coerce others into silence. This behavior is sometimes coupled with righteous condemnation of the bloodlines of those breeders who openly admit they have had a problem.

The Incorrigibles' attack mode dovetails with the all too human tendency to gossip. Fear of whisper campaigns and public condemnation has kept more than a few people from openly sharing information about epilepsy. Fear is further exacerbated by those who avoid breeders that openly admit difficulties in preference for those who tell them what they want to hear.

Both Incorrigibles and Ostriches have slowed research efforts by refusing to provide samples from their dogs. The information provided to the projects is confidential; they have no need to fear exposure. If they are correct and nothing genetic is wrong with their dogs, the completed studies would show that.

Epilepsy is now so pervasive that breeders are hard pressed to find crosses without risk. The shroud of secrecy and denial makes it impossible to know with any certainty where one should go. A few stricken breeders have become so discouraged they have given up breeding or left the breed altogether. Some just shrug it off: "It's everywhere." Perhaps if we had paid more attention ten or twelve years ago, it wouldn't be.

**Taking a closer look**

Since the lack of a positive diagnostic tool is central to the deniability problem, the best case scenario would be identification of the responsible genes on the molecular genetic level. Knowing which genes were involved and how they had mutated to cause disease would provide not only positive diagnosis for sick dogs, but a way to identify those that could become ill and pinpoint carriers before they were bred. A DNA screening test could greatly reduce the risk of producing affected dogs. Carriers that were otherwise of high quality could be bred to non-carriers and then their best non-carrier offspring used to perpetuate their positive qualities. The knowledge gained would also add to understanding of the disease process and might ultimately lead to better treatment.

Perhaps the most successful genetic study of canine epilepsy in a breed of dogs to date has been that conducted by Anita Oberbauer, PhD and Thomas R. Fanula, PhD, at the University of California, Davis. Oberbauer and Fanula started their work with the Belgian Sheepdog and Belgian Tervuren, two closely related breeds so close that in much of the world they are
considered varieties of a single breed. They have found that epilepsy in these dogs is polygenic with a single locus of large effect. Dogs must have a defective version of the gene at that locus in order to have disease, though some combination of other genes will determine if, when and how badly an individual will be ill. Oberbauer is currently working to develop a DNA screening test for those breeds and has expanded her studies to include several others. When contacted about this article she stated, "I suspect that seizuring in some breeds will involve a different suite of genes than those observed in the Belgians."

Oberbauer is not alone in that opinion. Dr. Ned Patterson at the University of Minnesota, is collaborating with efforts at the University of Missouri. He has identified an apparent autosomal recessive form of epilepsy in Vizslas. He is also looking at English Springer Spaniels, in which a preponderance of the affected dogs are male, though the reasons for that are not yet clear.

Belgian Sheepdogs, Vizslas and English Springer Spaniels are not closely related to Australian Shepherds. The genetics of the disease in our breed could easily be different from any or all of these. Aussies need their own study and, as stated previously, there are two in progress, which is to our benefit. The researchers for VetGen and those at the University of Missouri are almost certainly taking different approaches to the problem. Having several highly qualified people looking at the situation from different points of view gives us a better chance of having our particular puzzle sorted out.

I have looked at pedigrees of over a hundred crosses that have produced epilepsy. I suspect that it is not a single-gene trait in our breed. It cannot be dominant because it often skips generations. There appear to be equivalent numbers of males and females affected, so it is not X-linked. It clearly runs in families, but some individuals and some families seem to produce it much more often than others, indicating it may not be a simple recessive.

Some breeders have suggested that epilepsy is dominant with incomplete penetrance. This means there is a single dominant gene but dogs with the gene don't always show the trait. Those with the Ostrich Syndrome and Incorrigibles love this mode of inheritance because it enables them to point the finger at whoever owns the other parent of an affected dog. They conveniently forget the fact that even if this were true, there is no way of telling for sure which parent it came from.

Incomplete penetrance itself is open to question. I once made a concerted effort to locate an inherited disease in dogs that was proven to be dominant with incomplete penetrance. I couldn't find one. "Dominant with incomplete penetrance" is a geneticist's way of saying "it seems like it might be dominant but it doesn't quite fit the model." In other words, there are probably other genes involved. But the only way we will ever know for sure is if the genes involved are identified.
VetGen considered studying Aussies in the mid 1990s after Ann DeChant spoke to them at a booth they had set up at an all breed show soliciting support for another project. They have been gathering DNA samples from family groups that include two affected full siblings and at least three unaffected full siblings or parents. They needed a minimum of ten complete families to proceed and spent several years trying to amass that data, thanks to lack of cooperation from breeders, owners of siblings and, most especially, stud owners. They now have 11 complete and one partial family. While this meets their minimum requirements for sufficient data to begin further investigations, the project will have better hope of success if they acquire more. They have identified a likely candidate gene in a parallel project on Collies, but cannot yet say whether they see anything similar in Aussies.

The University of Missouri is conducting an on-line seizure survey for Aussies and several other breeds. [see sidebar "Epilepsy Research and Resources"] They are taking a multi-breed approach, determining inheritance patterns within breeds and identifying similarities and differences between them. While they want DNA samples on currently affected dogs and their relatives, they also collect information on epileptic dogs living and deceased through the survey. Currently they have DNA samples from 71 Aussies, twelve of which are affected. This number is insufficient for them to make any conclusions about our breed.

**What to do?**

What has happened to date cannot be undone, but that doesn't mean we should accept the status quo as a normal, albeit unpleasant, aspect of the breed. There are things we can do.

The most obvious is supporting VetGen and the University of Missouri with information and samples. If they don't get enough data, we will never have a screening test and our dogs will continue to suffer.

Until such time as a test is available, breeders need to avoid producing more affected dogs. Because this disease can inflict terrible suffering on both the dog and the people who love it, I feel it is unwise to continue breeding any first step relative of a dog with epilepsy. This would mean parents, siblings and offspring. There are numerous Aussies in the US and Canada. If we have the will we could carry on without using the most problematic animals. For breeders overseas, however, it can be more difficult.

Lisa Pugh, a British woman who has been slogging in the trenches with epilepsy since it first surfaced in the UK remarked "Breeder's need to be open and honest about health issues, as silence and intransigence affects the breed worldwide. Quarantine for dogs to the UK from the USA and Canada has been replaced by pet passport, which could expand our gene pool or reintroduce health problems that UK breeders have worked so hard to eradicate."
In a number of countries outside North America, eliminating all dogs with problem pedigrees may not be possible if the breed is to continue. Expense, import restrictions and quarantines have made expanding overseas populations a slow process. Tightening already restricted gene pools may solve one problem at the expense of bringing others to the fore.

No matter where you are in the world, you should avoid breeding close kin of an affected dog to mates that also have affected near relatives. Avoid crosses that have known producers or affected dogs on both sides of the pedigree. While these measures will not provide a 100% guarantee that you won't produce an affected dog, they will reduce the risk.

But we can't do any of this unless we start talking to one another. Stop treating epilepsy as though you were personally infected with a sexually transmitted disease. Take it by the scruff, haul it out of the closet and put it on a sit/stay in broad daylight. If everyone openly shared information on which crosses produced epilepsy, the most risky matings could be avoided. We need to temper this with the understanding that this problem is so widespread that we cannot reject every animal with a family history. Breeders with low-risk bloodlines need to be willing to share those genes.

Some are attempting to "proof" their lines by test mating. There are several reasons this is an unwise approach to controlling epilepsy. We do not know the mode of inheritance. If epilepsy is polygenic it isn't possible to clear animals through test mating. Even with single gene traits, unless the mode is known there is no way to evaluate the litter results to clear your dog. Epilepsy usually doesn't start until a dog is at least a couple years old and often shows up later. Neither the dog's breeding life nor the breeder's patience are long enough to await definite status on all pups from such a test litter. Finally, given the severe impact this disease has on the health of affected individuals and the emotions of those who love them, it is unethical to deliberately risk producing epileptic dogs in the name of "proofing" breeding stock.

People are starting to take positive action against epilepsy. Kathy Usher shared her dog Rascal's story in Janet White's article in the January/February issue of the *Journal*. The issue you are now reading contains two articles covering different aspects of the disease. People are talking at shows and on the breed discussion lists. When contacted about this article, Usher stated, "People have opened up to me that wouldn't have touched this subject a year ago. We are now having open conversations ringside about dogs we have bred, owned or descendants of our breedings producing idiopathic epilepsy."

A group of concerned individuals got together at the Australian Shepherd Club of America National Specialty in November of last year. This has lead to a grass roots movement of breeders who are sharing informative pedigrees, educating others about the disease, and encouraging
participation in the research projects.

Kristin Rush, who recently had dogs affected by epilepsy, sums up the situation: "I believe that the future of our breed will be significantly impacted by the way today's breeders react to this rising problem and the decisions they make regarding their breeding plans. Breeders can choose to ignore the problem and hope it goes away, which I seriously doubt it will, or they can step up to the plate, recognize there is a problem, take responsibility for it when it does happen, and ultimately safeguard the health and well-being of tomorrow's Aussies."

Epilepsy doesn't have to destroy the Australian Shepherd. It doesn't have to shred the hearts of people who love their dogs. We can turn off of this road to hell, if we only have the will to do so.