CA and the Scottish Terrier

By John McNabney

Cerebellar Abiotrophy (CA) is a genetic (autosomal recessive) neurological degenerative disease that affects movement. Its onset can occur as early as 3 to 6 months of age or it may not become apparent until significantly later in life. More details about this disease have been published in previous articles on CA in the Bagpiper. The above details are the salient facts for the purpose of this article.

CA exists in the Scottish Terrier. This is a fact that can not be disputed since it has been confirmed by necropsy. Apparently, almost every other characteristic of this disease is subject to dispute. This state of affairs is unfortunate as it becomes a rationale for doing nothing or provides fertile grounds for disputing actions that might be taken to reduce the frequency of occurrence.

Geneticists accepted by the AKC Canine Health Fund as experts in the field of canine genetics have determined that CA is an autosomal recessive genetic disease in Scottish Terriers. This is based on an analysis of the pedigrees of Scotties diagnosed as having CA. The presence of this disease and mode of inheritance has been accepted in many other breeds. Along with the STCA, the Old English Sheepdog, The Gordon Setter and the American Staffordshire Terrier parent breed clubs have agreed to sponsor a research project to find the DNA marker for CA under the auspices of the CHF. One of the diagnostic protocols for a clinical diagnosis of this disease is based on an analysis of a video of the subject dog performing specific movements such as climbing and descending stairs, side profile of movement at different speeds, etc.

Additionally, the owner must furnish the health history of the dog and information on the progression of the disease. This protocol was developed by Drs. de Lahunta and Bell and has proved to be accurate in identifying affected dogs confirmed by subsequent necropsy 100 % of the time. Since the purpose of this protocol is to identify dogs for inclusion in the research project to find the DNA marker for this disease, it is conservative and may, in fact, not diagnose affected dogs whose symptoms have not developed to the point of being obvious on the video, or whose movements have not been captured that would rule out other disorders. Other protocols are: in advanced states of this disease, a MRI showing a reduction of the cerebella along with an analysis of the movement by a neurologist is an indication of the presence of CA and a post-mortem necropsy provides the best evidence of CA.

To date, 43 Scotties have been diagnosed as having CA. They cross the spectrum of the Scottie community, from rescue Scotties with no known pedigree, to Scotties bred in active show breeding programs with carefully selected pedigrees. An analysis of the pedigrees shows the errant gene to be widespread throughout the Scottie gene pool. The spread of the gene and occurrence of the disease can be likened to a hockey stick. For a period of time the rate of occurrence remains low since the likelihood of two carriers being mated is statistically low. However, as the gene propagates throughout the gene pool, the likelihood of two carriers mating increases to the point where the rate of occurrence increases significantly. Even though the gene is widespread throughout the gene pool, the rate of occurrence of CA today would indicate that the gene has not reached this "tipping point" (the bend in the hockey stick) and it is possible to impact the gene distribution without taking drastic actions. However, time is not on our side in this matter. Procrastination or doing nothing does not slow the propagation of the errant gene.

Given the facts and the available science, the Board of the STCA decided to be proactive in addressing this disease. In doing so, they recognized that there is the possibility of damage being done to the Scottie fancy. However, the potential harm caused by doing nothing far outweighed the possible damage and made the actions taken prudent for an organization given responsibility for the welfare of the breed. Specifically, the Board created a Health Database Committee charged with establishing an open, voluntary CA database of affected CA Scotties and their pedigrees. Also included in the mission of the committee was responsibility for creating educational materials, programs, articles, etc. about the use of databases in breeding programs and the evaluation of risk of any proposed breeding. It is important that with the publication of this database, individuals who wish to use the information must recognize that there are many genetic conditions within the gene pool so that breeding decisions made on the basis of avoiding any dog that appears in the pedigree of an affected dog in this database is not a responsible decision. For one thing, the database does not reflect the full distribution of the errant gene. More significantly, concentrating your concern on the only condition with a database will only tend to bring other conditions to the fore, some of which could be far worse than CA. If the dog in question has the traits you need in your breeding program, evaluate the risk, but do not eliminate him from consideration.

As the Health Database Committee progresses with additional databases, the decision making will become more complex. However, there will also be procedures published for evaluating risk from multiple sources.

For breeders, we are moving into a new era. With the availability of more information comes the responsibility for using it wisely. Properly used, the databases can slow down the progression of errant genes throughout the gene pool. Knee-jerk reaction to the databases can lead to a dangerous shrinking of the gene pool and the probability that new and perhaps far more serious genetic conditions will emerge.

Questions have been raised about the validity of the diagnostic protocol. Once the diagnosis is accepted by the fancy, most of the other issues disappear. While I will address some of those issues below, there remain others that should be answered. For readers with questions that are not addressed, I would encourage you to forward them to me and I will try to get accurate answers back to you. Significant, unresolved issues will be discussed in future articles.

1. *The protocol has not been published in a peer reviewed journal*. The clinical diagnosis protocol was developed by two researchers who have been studying Cerebellar Abiotrophy for a combined 65 years. It is based on experience, and not something that can be taught in an article. Since there is no intent for this protocol to be used by anyone other than the team that is presently using it, there is no need to publish it.

2. *There are many other conditions that affect movement in Scotties. How can we be sure that these conditions are not producing the movement defects being diagnosed as CA?* Having lived with a CA affected Scottie; I can assure you that the movement of a CA Scottie is unique to that condition. Close your eyes and touch the end of your nose with your finger. The ability to do this is the result of spatial feedback the brain receives whereby it knows the location of each limb. CA diminishes this feedback so that the affected Scottie does not know the location of the limbs, including the head. Recognition of onset of CA can be in puppy hood or as late as 6-7 years of age. Being degenerative, the condition worsens as the Scottie ages. It can become so severe the Scottie is incapable of independent movement. We have had Scottie Cramp and arthritis affected Scotties and can categorically state that those movement defects are distinctly different. Those movement problems caused by skeletal defects (Leggs Perthes, Luxating Patella, Hip Dysplasia, etc.) involve pain which is never present in CA, even in its most extreme manifestation.

3. *Without pathological confirmation, how can any visual diagnosis be considered reliable?* In the four affected breeds, to date, every dog diagnosed by Dr. Bell, Dr. de Lahunta, or Dr. Olby as having CA that has subsequently died and had a necropsy performed has been confirmed as having CA. While this record is no guarantee that a mistake will not be made, it is a better record than most generally accepted diagnostic protocols.

4. *Different people can view the same video and see different defects.* By limiting the diagnosis to one team of experienced diagnosticians and requiring unanimous agreement, consistency can be maintained.

5. *What about the study where the CA team was shown videos of Cramp Scotties and the Cramp team was shown videos of CA Scotties and each team concluded that the Scotties were affected with the condition they were addressing*. It never happened! This story has reached the proportion of urban myth, but at best is based on a misunderstanding of what was said in the parking lot of the 2004 rotating specialty.

6. *How can you be certain that your Scottie had CA?* Fiona was diagnosed as afflicted with CA by Dr. Bell at the age of 7. Her abnormal movement had not become apparent until she was over 5 years old. Her condition worsened throughout the rest of her life, but even at the age of 11 ½, when she died of other conditions, she was able to move on her own, albeit with difficulty. CA was confirmed by necropsy.

There will be no magic bullets when it comes to dealing with genetic conditions. There will be some pain as breeders find themselves affected by published databases. Proper use of the databases and registries can provide some guidance for breeders as they try to navigate their way through what has been described as the minefield of genetic defects. Full disclosure will help others avoid the landmines. Fear of consequences that leads to suppression of adverse results can only impede our way towards eliminating errant genes from the Scottie gene pool. Research devoted to finding genetic markers for these errant genes will eventually lead to their elimination, but we can not delay addressing the problem by taking action that will at least slow the spread of these errant genes now.