



SCOTTISH TERRIER CLUB OF AMERICA

Cerebellar Abiotrophy and Breeding Strategies

By Vicki Campbell

Dr. Jerold Bell, of Tufts University, addresses a crowd of approximately 70 Scottish Terrier Breeders, exhibitors, and interested others at the LuLu Temple, October 2, 2004, sponsored by Scottish Terrier Club of America.

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On Saturday evening, October 2, 2004, about 70 Scottish Terrier breeders, exhibitors and ordinary Scottie lovers dragged their dog-show weary bodies into the Merrimack Room at Lulu Temple in Plymouth Meeting, PA. This was not a large room so this was a big crowd! There were many big name breeders there and some not so big, like yours truly. There was also a contingent of CA online support group members present. You could tell who they were because they were proudly sporting their brand new CA logo T-shirts.

The CA seminar was to be given by Dr. Jerold Bell of Tufts University and by Dr. Natasha Olby of NC State. We later learned that Dr. Olby had some health problems and would not be there. Dr. Bell introduced himself as a dog breeder, a veterinarian and a geneticist. I had never met Dr. Bell, but had emailed him once or twice when he first started this study three years ago. I heard he was a good speaker from a Briard breeder/veterinarian friend of mine, so I was hoping for the best. Right off I decided the guy must have a good sense of humor because he had a slide on the screen of a Mark Parisi cartoon dealing with snowmen and inconclusive DNA tests.

The program started with historical review of registration trends in Scotties in the last 20 years. In 1985 there were 5750 Scotties registered with the AKC (making us number 38 of all breeds). This ranking rose to number 36 in 1990 with 8724 dogs registered, but dropped down into the 40s during the 1990s due to many reasons including other purebred registries and pet store/puppy mill type groups. In 2003, 3559 Scottish Terriers were registered.

Dr. Bell next talked about genetic diversity in all breeds and how the "popular sire syndrome" can shove the gene pool toward a particular individual and bottleneck breeding programs. This technique can shut out the use of other dogs with fewer ribbons and trophies, but who could add to the quality of the breed.

On the subject of breeding goals, Dr. Bell said that the PRIMARY goal of a breeder should be to maintain and enhance the quality of the breed - this MUST be your goal. The SECONDARY goal of a breeder is to breed genetically healthy dogs. There are a lot of healthy mutts out there and a lot of ugly, but healthy Scotties. You must breed for the "whole" dog. A breeder has to be able to see the whole picture. *(When I heard this, I thought, this is going to ruffle a few feathers, but in the long run I thought it made sense.)* It is important not to produce affected dogs that are burdens to themselves

and to their owners. As a breeder you must decrease the carrier frequency of defective genes, but if you refuse to breed carrier dogs altogether, there is the chance that you will eventually lose the breed.

Dr. Bell talked about the quality of the AKC Canine Health Foundation. They have phenomenal resources and the highest quality of research. He feels we should all support this program. The Scottish Terrier Health Trust Fund works with the CHF on research programs that involve Scottish Terrier health issues. The Canine Genome Sequencing project results will be announced in Holland soon, according to Dr. Bell.

Health surveys are important because they help determine trends of disease in the different breeds. According to the CHF, frequently occurring disorders in all dogs include the following: epilepsy, hip dysplasia, cancer, cataracts, bloat, hypothyroidism, PRA (progressive retinal atrophy), allergies, cardiac disorders, patellar luxation and eye disease other than PRA. The STCA health survey done in 1995 was looking for the following genetic health issues: Scottie cramp, VWD (von Willebrand's disease), Cushing's, hypothyroidism, epilepsy, CMO (cranial mandibularosteopathy), liver shunt and juvenile cataracts. The 1995 survey was the first comprehensive survey done by our parent club. Dr. George A. Padgett, DVM reviewed the findings and applied the Hardy-Weinburg rule to them and produced a "guesstimate" frequency rate. The "guesstimate of frequency" indicates the percentage of Scotties that may carry this particular trait. It does not indicate the number of affected dogs. The Hardy-Weinburg rule states there will be approximately 1% affected for every 18% of carriers. Usually the Hardy-Weinberg rule is applied to single gene traits. Dr. Bell then showed many screens of things like risk assessment, and lots of numbers and ratios and percentages. There was way too much to try to write down and explain, but I did come out with one item in this segment that made sense to me. He was talking about the VWD testing and said that if one dog tests positive then the whole family should be tested. I would think this would be true for any disorder for which there are tests. Dr. Bell said the STCA (and all breed clubs) need to do health surveys more often than every ten years (the next STCA health survey comes out in 2005).

Finally Dr Bell arrived at the topic we had all been waiting for-Cerebellar Abiotrophy, also known as Cerebellar Ataxia. He said that it was the first time since starting this project three years ago that he was speaking to a group of Scottish Terrier breeders. CA is a disorder of movement and muscle coordination - and timing. It is like a muscular dystrophy BUT it is not MD. The Purkinje cells in the cerebellum begin to die off with this disorder. These cells act as a connector between several other cells in the cerebellum to control a process called proprioception. This process allows the mind to recognize where your limbs and body are in space. Because of this process you can close your eyes and still touch your nose because you know where it is. This process is progressively lost in a CA dog. Some suffer severe loss, others only mild loss. In fact, most affected Scottish Terriers show only the mild symptoms. The severely disabled are in the minority. As we have all heard and said before, no one wants to produce a CA affected dog (or one with any other genetic disorder). It just happens and we as breeders and owners have to learn to take the stigma out of genetic disorders.

One of the signs of Cerebellar Abiotrophy is a hypermetric gait-goose-stepping. Dr. Bell showed video clips of affected Gordon Setters. These signs are much more obvious in the long-legged breeds.

Stairs are very difficult for CA dogs because stair climbing is a highly coordinated skill. Video clips showed an affected Old English Sheepdog carefully trying to place its paws on the steps so it would not fall - it was not a graceful sight. CA dogs have a smooth gait; they just don't know where their legs are going. Some CA dogs roach their backs, but not all of them do this. This is all part of the coordination problem caused by the loss of the Purkinje cells.

Dr. Bell did say a bit about the difference between Scottie Cramp and CA. Scottie Cramp is episodic - and there may be some cramping pain involved. CA is continuous and is painless (we think). It is often difficult to tell the difference between a mildly affected CA dog and one with Scottie Cramp, especially to the novice eye.

In diagnosing a CA dog, you need more than a video of the dog moving. You must have a diagnosis by a board certified veterinary neurologist in addition to the tape.

The tape must include the dog participating in running off lead, climbing stairs, chasing a toy and any other problem movements your dog may be exhibiting. It is also helpful to have an MRI of the cerebellum to check for shrinkage. Some young dogs will have clinical signs without a clinical history. Other brain or spinal disorders can cause movement problems too and these must be ruled out before a diagnosis of CA can be confirmed. Dr. Bell, Dr. de Lahunta (Cornell) and Dr. Olby will be reviewing submitted cases.

While discussing pedigree research-an important part of careful breeding-Dr. Bell said that a lot of dogs of various breeds came to the United States during the 40's because of the war. Research in genetic disorders can sometime go back to these imported dogs.

In the current CA project, 38 Scotties have been confirmed with the condition. Of these there are 32 with submitted pedigrees. This is out of 111 Scottish Terrier contacts with Dr. Bell. There are more affected Scottish Terriers than any other breed in the study.

Dr. Bell discussed the CA research project, which is a grant proposal to the AKC Canine Health Foundation made by Drs. Natasha Olby (NCSU) and Matthew Breen (NCSU). Dr. Breen is a top researcher who worked on both the canine genome project and the human genome project. In collaboration on this project are Drs. Dennis O'Brien (UMO) and Jerold Bell (Tufts). They are going to be doing molecular genetic research to study Cerebellar Abiotrophy in American Staffordshire's, Gordon Setters, Old English Sheepdogs and Scottish Terriers. All four breeds will require the same clinical and photographic presentation. The researchers will be looking for a simple autosomal recessive mode of inheritance and hoping to identify the defective gene for CA. The grant review has been done and apparently has a good reading. The process is to continue into January. All four breed clubs must support this research. Am Staff and the OES clubs already have DNA on affected families collected and stored. The research must be completed in two years. The Scottish Terrier part of the study must include diagnoses and confirmation of such, collection and storage of family DNA, and the finding of the markers or genes identified in the other breeds in this study. An informational website must be set up for each breed. And the research group must contact the owners of the already diagnosed dogs every three months.

According to Dr. Bell, CA is not the most prevalent or important hereditary condition affecting our breed, but it is being diagnosed all over the world. It has a simple autosomal (pertaining to a chromosome that is not a sex chromosome) mode of inheritance and a wide pedigree base. Any Scottish Terrier line can produce an affected pup at this time.

The last "formal" part of the talk was on managing genetic disease. Dr. Bell believes that no dogs should be put down or neutered because of genetic disease. The management recommended will vary due to many factors. In the case of "dominant" genes, you can replace the affected dog with a normal sibling in your breeding program. You do not want to knowingly breed affected dogs. For "recessive" genes where there are tests for the disorder, you can breed the carrier dog to a normal mate. Eventually you will breed the gene out of your breeding program. You can replace the carrier parents with genetically normal offspring. You want to select against breeding carriers, without losing the object of breeding quality dogs.

CHIC-the Canine Health Information Center-was highly recommended by Dr. Bell. It is an open health database for all breeds. National breed clubs must determine three diagnoses to be eligible. Usage of this service carries no stigma-or it should not. In fact, it will help lower the breeding risks for many disorders.

Ah, the best part of the evening was, of course, the dogs! First up was Fiona, who was almost 11 and mildly affected. You mostly saw her back legs scrambling to figure out where her front half was going. She spent her evening visiting with the admiring crowd and cleaning the floors of Lulu Temple.

Everyone wanted to pet her. Next was Mickey who is 5 and has his CDX in obedience. He is also mildly affected. He was a wild man tearing around the place doing his sits and fronts and all the cute stuff he has trained for all these years. His owner always thought he was a bit clumsy, but decided it was more than clumsiness when Mickey ran through the agility set up at his first training class-like a bull in a china shop. Both Fiona and Mickey showed that Scottie personality definitely could outshine any disability they might have. But the dog that brought everyone to tears was Holly, who is eight and is severely affected. Her dad put her down on the rug and her four legs went flat out to the sides. She maneuvered around by scooting on her tummy. Her dad said that sometimes if a strange dog or person is passing by, she somehow could manage to get up on her legs and get to where she can tell them off. She has a little harness that lets her owner get her up on legs-more or less. Her first signs started showing up when she was about two and taking handling classes. She certainly has the true "diehard" spirit of a Scottie and great Scottie attitude. But I am sure I was not the only one who had a knot in their throat watching her and seeing how devastating this disorder can be.

The seminar was to end at 9:30 P.M., but when I left at almost 10:00 P.M., Dr. Bell was still being bombarded with questions. We can only hope that this is the beginning of communication between breeders and owners and we are on our way to finding a way to breed CA out of the Scottish Terrier.

[NOTE: The following contact and research information is no longer current.]

If you would like to read more on Cerebellar Abiotrophy or managing genetic disorders, there are articles in the *Bagpiper*, 2004, #3, also in the *AKC Gazette*, October 2003. For information on the subject matter, you might want to contact Dr. Bell at jerold.bell@tufts.edu. If you suspect you have a CA affected dog, please use the following email address: Geneticvet@aol.com.