Further comments on Shaking Puppy Syndrome (SPS)
bym the Border Terrier Breed Health Co-ordinator

As a follow up to earlier comments on SPS, I understand much has been said on social media and what follows is intended to provide Border Terrier owners with further advice and guidance on progress to date.

It would appear some people are claiming to hold significant amounts of data on litters where SPS has arisen. If this is correct, this is important information which needs to be made available to those with experience in identifying emerging inherited diseases. Furthermore those experts need to be engaged in the research and funded correctly or the progress will stall or fail.

Until this data is made widely available little can be done and I once again urge anybody who has direct information to release it without further delay. It is impossible for either myself as Breed Health Co-ordinator or the breed clubs themselves, to make statements regarding SPS if the condition is not being properly reported. As BHC I have received a few snippets through third party reports of affected litters but despite asking for further information this has not been forthcoming.

What do we know at this stage?

A published Case Study from the Ohio State University, Columbus reports upon four cases of tremors in young Border Terrier puppies. The information on these pups was reported by researchers and clinicians from Missouri and the Norwegian Veterinary School in Oslo. There is much useful and detailed pathology in the paper, as this work has been entirely based on post-mortem examinations of euthanased pups.

In summary the report suggests this is the first recorded appearance of a degenerative spongiform change of myelin in white matter of the brain, principally in the cerebellum, brainstem and spinal cord and to a lesser extent in the thalamus and cerebral hemispheres. This provides the descriptive term Spongiform Leuco-Encephalo-Myelopathy (now being shortened to the acronym SLEM). Myelin provides insulation around nerve fibres and any serious defect in the formation of the myelin sheath is likely to lead to poor transmission of nerve impulses and therefore neurological symptoms.

The symptoms reported include uncontrolled tremors and incoordination of hind limb movements which was consistent with the areas of the spinal cord that were affected. The pups were around three to four weeks of age when presented for euthanasia and the tremors were first noted when the pups started to walk. The Scandinavian pups were reported as becoming progressively worse over time.

The authors note the similarities and differences between this condition and other myelin sheath degenerations in other types of dog where perhaps the most well known is the shaking puppy condition in the Weimaraner. However it would appear pathologically the sites of the brain most affected and the microscopic appearance of this myelin degeneration in Border Terriers is unique.
As a common male ancestor was found in all four pups (bearing in mind one was from a US breeder, two were presented at Uppsala in Sweden and the fourth at the Oslo Vet School) a genetic mutation is considered possible. The US pup was male and the others were female.

As this condition has some similarities with a human condition (Canavan’s disease), which is known to be an autosomal recessive gene mutation, the authors speculate how this might be a single gene autosomal recessive mutation leading to defective myelin production.

The Case Report was accepted for publication in late 2011 and therefore it is likely these pups were first presented in either 2010 or the early part of 2011.

There is much in this report that requires further work but of course the first question is the likelihood that the cases of SPS being seen in the UK are the same condition. Given the circumstances and the fact that the common ancestor was born in 1975 we can only hope that this is so. Of course other congenital myelin defects are reported in other breeds and this raises the possibility of another mutation. However the working assumption that we are dealing with the same inherited condition in the UK is valid for the time being, especially as the common ancestor from 1975 is likely to be a British dog although this is not stated.

**Further work is apparently underway**

Why has this condition taken so long to surface here in the UK? The fact that the common ancestor was born in 1975 does not in fact mean this was the first time the mutation occurred as this could have happened even further back in history. If we are fortunate enough to have a single mutation identified and it proves to be autosomal recessive, then we will have the basis for a useful gene test to assist us in dealing with this neurological condition. Until then we have to rely on more traditional methods.

Currently the hypothesis proposed suggests a single recessive gene defect. To produce affected pups requires that both parents will be carriers of the mutation. Their mating will theoretically produce 25% clinically affected pups; 50% carriers; and 25% clear. The carriers and the clear pups will be clinically unaffected. In addition both parents although carriers, will be clinically normal too.

This is consistent with the Ohio Case Report as littermates of the affected pups were clinically normal as were the parents. It would be very helpful to know how many litters are known for certain to have been affected in the UK along with their pedigrees and how many pups in each were clinically affected.

The distribution of carriers from the original source of the mutation would not have been known at that time, but further pedigree analysis might reveal how extensive the spread of the mutation may be within our breed in the UK. It is quite possible that the original mutation pre-dates the identified source of the problem in the Ohio report but even if this proves incorrect there is a very real chance of being able to control further spread of the mutation if pedigree analysis can be completed quite soon.

**Avoid mistakes**

Clinically affected animals will have arisen as a result of in-breeding as this would bring together male and female carriers several generations onwards from the original source of the
mutation. Specific advice can be generated for a breeder who is unfortunate enough to have a dog or bitch that proves to be a carrier and this does not necessarily mean that either or both must be removed from the gene pool. Sadly this a mistake many have made in other breeds in the past thus creating a significant reduction in the available gene pool and a greater risk from in-breeding as a result.

One aspect of this condition which can be viewed as encouraging is the onset of clinical signs of SLEM at around three weeks of age. This allows any action to be entirely in the breeder’s hands and avoids new puppy owners buying pups that develop the problem. Healthy litter-mates can be sold with breeding endorsements which can be removed in cases where a suitable breeding strategy can be identified.

However, before any of this can be achieved breeders need to be able to recognise the symptoms. The onset of tremors in pups as they commence to walk is a primary feature. Tremors predominantly affect the hindquarters but also the head and fore-quarters too. The tremors cease when the puppy sleeps.

Eliminate other conditions

There are other conditions that may cause pups to shake or suffer tremors and these will need to be recognised to eliminate other causes. Infection or inflammation of the cerebellum, congenital defects in the development of the cerebellum and neonatal cerebellar ataxia are all possible alternative causes but these will all have a variety of differentiating clinical signs.

Finally we need to record the litters and collect the pedigree data. This is relatively easy to achieve and the Health Co-ordinator is the logical focal point for data collection.

We require
- the litter information including pedigree,
- the number of pups affected,
- a confirmation of the clinical diagnosis preferably by a veterinary surgeon and
- if available, the pathology reports.

It is not too late to provide information about past litters as described above.

Keep calm and promote co-operation

The common sense approach is to remain calm and not seek to apportion blame. I doubt anybody intended to breed a litter with affected pups, and as both parents would be healthy there would be little evidence to help a breeder anticipate trouble. If the information is supplied we can arrange to assist the breed in future selection of breeding pairs to try and control the spread of this condition.

I would also recommend we resist speaking in riddles about any perceived historical sires that could be the source. I have had at least three suggested to me but these all post date the 1975 source mentioned in the Ohio report. So they may be carriers but they are probably not the root of the problem.

What is needed is more co-operation and informed, constructive advice and can we please cease the ‘disgusted of Stoke Poges’ type of on-line media formats.
How we find a solution to the challenges of social media is certainly not an issue for your Breed Health Co-ordinator, but it would be good if, as breed supporters, we could cease the cloak and dagger approach and deal in defensible facts that are made widely available.

In the interim the AKC Canine Health Foundation has granted some money to Missouri Vet School to conduct a search for the gene or genes involved and I will provide further updates on this work when information becomes available.

Finally I understand there has been some criticism that the Border Terrier breed clubs have supported the ‘give a dog a genome’ project, describing this as a historical venture. Well the source of the SLEM problem is historical and unravelling these historical factors will aid us in finding a potential solution. However the AHT genome project is about the future not the past, and readers will be interested to know that sequencing the entire genome of an affected dog is part of the genetic programme at Missouri. So the genome project is likely to be a significant part of the solution for breeding borders without SLEM as a risk.

If you wish to contact me I can be reached on 01628 782787 or stevedean@tyrianborder.com. I have been collecting confidential information for a very long time and I can assure you that I will keep personal details entirely confidential and will not seek to embarrass or offend anybody.

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