**Border Terrier Health Report 2016.**

This is possibly the most challenging report on Border Terrier health I have written since I started out in Border Terriers some 35 years or more ago. Usually I can be fairly reassuring about the health of the breed but it is currently under greater risk than it has ever been and club members, who form the backbone of the breed, need to be aware of this.

You may think this is because of Canine Epileptoid Cramping Syndrome (CECS) or Shaking Puppy Syndrome (SPS) and in one respect this is true but not perhaps for the reasons the reader might anticipate. Both conditions exist in the breed but the prevalence is arguable. Anecdotal reporting is constant but despite this it would appear from factual reporting that annual clinical cases in the UK, of both conditions, are small in number although under-reporting is likely.

The chief risk to future health of our breed rather perversely arises from the actions of what I assume are a well meaning group within the breed who appear to have gathered reports about both conditions and are offering advice to anybody who wishes to listen. For reasons that are not entirely transparent (to me at least) these reports have not been shared with myself as breed health co-ordinator, nor has the clinical detail of the cases been made available. This informal approach relies upon social media exchanges and internet listings of assumed affected dogs along with unverified analysis of unknown pedigree information by non-professionals. Sadly it is the advice that emanates from these sources that offers a fundamental threat to the future health of our breed.

I will explain this further to justify my warning. A recommendation to avoid breeding from supposed affected lines is a flawed concept unless there is very sound verified evidence which identifies affected dogs, carriers of assumed mutations and dogs established as clear of either clinical illness or genetic mutation. Simplistic advice without this critical review cannot be relied upon and does not appear to take into account current knowledge of genetics and, most importantly, it has the capacity to greatly unnecessarily reduce the gene pool for the breed. It is this risk to the genetic diversity of our breed that causes me to write this report for at this time a catastrophic and unnecessary reduction in the gene pool risks the emergence of further, as yet unrecognised, inherited conditions in the Border Terrier.

All dogs carry mutations in their genome and where these are injurious to health, in-breeding is the most likely route which brings these genes together to cause clinical illness. I will deal with a more acceptable approach to dealing with an inherited condition shortly but first let me elaborate upon some of the historical aspects associated with the emergence of both CECS and SPS.

Perhaps the most important factor in the recent development of both conditions is an appalling lack of co-operation within the breed. Claims that either I or the breed clubs were not interested in these illnesses are at best misplaced and potentially malign. We have had a mechanism in place for reporting health concerns (the Border Terrier Breed Health Survey) which has been available on club websites for at least two decades. There have also been two Kennel Club surveys carried out during the same period of time.

There has been very little reporting of either CECS or SPS to the breed health co-ordinator despite the fact that everybody who approached me with anecdotal information was asked to provide the information but largely failed to do so. This remains the case today. Excuses that the current questionnaire does not readily cater for the unweaned puppy suffering from SPS have been noted and will be remedied but this does not excuse the fact that no hard information has been provided until recently and what we do have has been very challenging to obtain.

It is the hard clinical evidence that forms the core of any investigation into the causes of a known or emerging disease. The ability to accurately diagnose a condition is the single most effective skill in planning for a programme of research for both CECS and SPS. Once a diagnosis can be securely made then the next important step is the collection of the data centrally for analysis alongside DNA samples from normal and affected dogs for genetic research. However collecting DNA samples and reports is valueless unless they are linked to strong clinical evidence and accurate data.

Despite much effort over the years, the work to establish the causes of CECS suffer from a distinct lack of progress worldwide. This should signal to us that what has been done has largely been ineffective. As breed health co-ordinator I cannot advise the breed unless those involved engage with the system put in place. However, in the face of criticism on social media the breed clubs have agreed to the formation of a small group of experienced volunteers with the necessary scientific skills to attempt to improve the situation.

It is important to realise no group will make progress without hard facts and anybody who believes they have reliable information is openly invited to contribute by supplying the information they hold on either CECS or SPS.

**Canine Epileptoid Cramping Syndrome**

I have been involved in the consideration of CECS since it was first reported by a German veterinary surgeon, who was a personal friend and fellow breeder of border terriers. From the outset it was my view, after viewing the first video evidence, that this appeared to be a neurological disease. At this time others were pursuing theories of a form of muscle cramping or a bowel or liver condition and were attempting to link diagnosis to bile acids tests. It is now generally accepted that this condition affects the Central Nervous System (CNS) and the symptomatic involvement of other organs is as a direct result of the influence of the CNS.

The very name we use for the condition is descriptive of the symptoms. However, some cases may not show seizures, or stomach or muscle cramps, which must make some owners very confused when the diagnosis is made. Furthermore most veterinary surgeons remain largely unaware of the features of this condition which adds further doubt to the accuracy of some diagnoses.

Over time, personal enquiry has revealed that CECS has probably been in the breed and other terriers for a very long time although it was limited in its clinical distribution by careful selection of breeding stock by the breeders of the past. The fact that it was first reported in Germany, the Netherlands, Scandinavia and the USA reflects this. The breed in these countries was based largely on a few imported breed lines and, most importantly, these lines were significantly in-bred in those regions.

It is inbreeding that most successfully brings recessive and polygenic (multiple gene) conditions to the clinical foreground and the use of popular sires and repetitive line-breeding in the UK are likely to be the major contributing factors to the emergence of CECS in the UK. Nevertheless, in my view it is destructive and lacks logic to blame the breeders of the original stock or the current breeders for producing clinical cases, as the emergence has not been well reported nor has it been accurately documented and thus advice so far has lacked a strong scientific base and has largely been in the hands of a small minority of lay-persons.

This remains the same today despite the efforts of several well meaning individuals over several decades. The chief issue revolves around the belief that CECS is inherited by relatively simple means and furthermore it makes the huge assumption that we are able to accurately diagnose CECS. Nothing could be further from the truth and this remains the situation despite some recent progress.

This may seem strange given the plethora of video clips available on-line but to the professional eye many of these videos show different symptoms and this was confirmed by the study by Garosi and others when an attempt was made to characterise the symptoms of CECS based upon historical cases. There was some commonality in symptoms in most cases but the study served only to formalise our limited capacity to confirm a diagnosis. As a result, diagnosis is often achieved by excluding other causes and this is unlikely to provide accurate data. We will have to work with this for the time being.

Current work by an associate of Laurent Garosi (Mark Lowri), is focused upon exploring the theory that the biochemical basis for the clinical signs of CECS has something to do with the metabolism of the protein gluten. Many owners of suspected CECS cases report that dietary change was able to improve their dog’s symptoms and Mark Lowri has ascertained that in some cases it is the removal of gluten from the diet that appears to have a beneficial effect on the appearance of clinical symptoms. I will stress this information remains theoretical. Some dogs do not respond to the exclusion of gluten in their diet and this may be because of access to gluten from other sources; an inaccurate diagnosis of CECS; or the possibility that gluten is only involved in some forms of CECS.

**Shaking Puppy Syndrome (SPS)**

The situation for SPS is different to that of CECS. This condition has been characterised in the scientific press and a significant pathological change has been identified in the brain which is best described as poor myelination of the nerve pathways in specific regions of the brain, especially those associated with co-ordination of movement. Thus unlike CECS there is the potential to confirm a diagnosis albeit only at post mortem following death of the affected puppy.

Once again here is a condition that has been widely commented on in social media but apart from anecdotal reports very little hard clinical evidence has emerged since the single published report in 2012. The original scientific report emanated from the study of four young puppies in the USA and Scandinavia and based on these four pups the authors speculated this could be caused by a single recessive gene.

If this is the case then we can be very hopeful of finding the mutation and developing a diagnostic test. However studies have been going on in the USA since this report was published and as yet no tangible progress has been made and this should be a stern warning that there may be a flaw in our assumptions for this condition. This is not a criticism of the research work to date but is supportive of my view that we need to be very careful to ensure the information we are working with is as accurate as possible. Thus anecdotal evidence is helpful to an extent but is not hugely contributory to any scientific study of the condition.

For example, based upon the anecdotal evidence it has been suggested that the condition appears characteristically in 2-3 week old puppies as they started to walk and the symptoms have been described as being typical of SPS. However the most recent case report in the UK, supported by confirmatory post mortem evidence, was from a puppy that did not show symptoms of incoordination until some weeks after it was sold (i.e.10-12 weeks of age).

This alone should make us very concerned about what we are looking at, for if the original theories of this condition are correct then this puppy is exceptional and yet the pathology appears to be very similar. Does this mean we are dealing with two forms of the condition, a different condition entirely or does the manifestation of the clinical signs depend upon environmental factors or the expression of another mutated gene (or genes)?

**Moving forwards**

On social media I have been criticised for suggesting the evidence for both conditions is largely anecdotal. However this is an accurate statement and furthermore it follows that although I believe both conditions exist, the majority of the case reports I have seen lack robust supporting diagnostic evidence.

If we wish to change this situation we need to commence collecting data effectively and that is why a small group of people\*\* have been assembled by the breed clubs to work together to offer advice and recommendations to the breed in seeking solutions to aid our control of these conditions.

**The proposed direction**.

It has taken some 40-50 years to get to this point for the Border Terrier and we cannot expect to resolve these problems with simple short term breeding plans. Indeed current advice has the potential to make matters worse for the breed. It will take time, research and money to find a lasting solutions and in the interim there is a real danger of more inherited diseases emerging if the breed does not work together.

These conditions are of international concern among owners and breeders of Border Terriers. We therefore need to cooperate with our international colleagues. We need to bring together those in the science community who have an interest in assisting the breed and we need to plan for the next steps toward a solution.

Thus the current work is aimed at bringing together a core of scientists, including veterinary specialists, with an expressed interest, to form the basis of a research group in the UK.

We are reviewing progress internationally and will ensure there is communication between the different teams around the globe (principally in Europe and the USA) and the UK group.

Working with these experts we will produce a protocol for the diagnosis of each illness, the collection of data from confirmed clinical cases and arrange for independent expert analysis of pedigrees.

In parallel, there will be a published procedure for collecting and storing DNA from both clinically affected dogs, assumed carriers and dogs appearing to be from lines free of each condition with the intent to enable genetic research to identify suitable biological markers to aid breeders in the future selection of breeding pairs.

**Final comments**

Despite comments to the contrary on social media, it is in fact the Genome Project at the Animal Health Trust (AHT) that offers our best chance of identifying the genetic factors for both conditions and if successful this could lead to a genetic test for each disease. However we need to keep a perspective on the likelihood of success, for despite many years of activity we are no further forward in understanding the inheritance of CECS and the simple recessive inheritance proposed for SPS is yet to be confirmed.

Consideration is being given to arranging a meeting in the first half of 2017 to bring those interested from the breed and the potential research groups together to discuss the challenge we face and agree the way forward.

The UK Border Terrier Clubs are not alone in their concern and the fact that other countries have failed to identify the inheritance and the gene mutations associated with each illness is a cause for concern. This is strongly suggestive of a flaw in the research projects or the original theories behind the research. Thus we need to ensure we do not simply re-invent the wheel as we go forward.

Finally research costs money and eventually there will be a need to create a fund for this work and use this to seek support from other grant sources but we cannot identify these costs until we understand what the entire research project might be.

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