

AHT DNA Cheek Swab Kits

March 2017

Starting with our Championship show on 11th June 2017 DNA cheek swab kits will be available at all of our club events, which may, we hope, help in the Animal Health Trust's ("AHT's") research into health in the breed.

We will be requesting packs of swabs from the AHT. In each pack there are 5 swabs to be used for an individual dog. There are full instructions in each pack as to how these swabs are to be used and what to do when sending them in to the AHT in the envelope provided, along with a form to complete with details of any illness from which the dog is suffering, or to confirm the dog is "Free of known inherited disease to your knowledge". The instructions also ask for a 3 to 5 generation pedigree of the dog whose sample is enclosed (where possible) and all relevant information regarding inherited disease i.e. copies of eye exams, hip scores, dates of diagnosis, key notes from the clinical history.

The leaflet confirms that information provided to the AHT will remain confidential and will not be shared with third parties.

The swabs are free - the only cost being the cost to you of postage back to the AHT.

The following is a document provided to us by the AHT this last week, from which you will see that the AHT are also interested in obtaining cheek swabs from apparently healthy Border Terriers of 10 years and over, required to assist in the research. If this applies to your Border Terrier, his or her swabs can greatly assist in the research, so please ask for a pack at the events (these will be available from the Secretary):

The AHT document:

"Border terrier research at the AHT – March 2017"

Spongiform leukoencephalomyelopathy, otherwise known as Shaking Puppy Syndrome (SPS) and Canine Epileptoid Cramping Syndrome (CECS) are two health conditions which are evidently a concern for many within the Border terrier breeding community, and the canine genetics group at the AHT is happy to assist potential future research by supplying swab kits for sampling to build up an archive from a wide selection of clinically affected cases and unaffected controls.

The Border terrier breed has also signed up to our Give a Dog a Genome project, and for this we will be sequencing a dog affected by SPS and also sharing sequence data with researchers in the US to benefit the study.

To date we hold DNA samples from nearly 400 Border Terriers, of which almost 90% were submitted in the past year alone. Among them are samples from 9 SPS cases and 53 that are potentially affected by CECS, although the majority of the latter are owner reported and have

not be verified by a veterinarian.

Due to the lack of a definitive diagnosis for CECS selection of appropriate samples for use in any genetic study will be challenging.

Owners are encouraged to sample dogs that are potentially affected with CECs and to also submit video evidence of episodes to the AHT, alongside their DNA sample, for potential scrutiny by a veterinary neurologist familiar with CECS. The genetic approach for CECS is likely to be in the form of a genome-wide association study (GWAS), which compares genomic markers from as large a group as possible of the most robustly diagnosed cases against a similar number of older unaffected control dogs.

Ideally for a GWAS we would use a minimum of 48 of the most robust affected cases and 48 controls comprising of the oldest dogs unaffected dogs available (10+ years preferably).

We would like to stress that no specific testing is taking place at this time. We are simply storing samples and recording the accompanying clinical and pedigree information where provided.

Work on SPS is due to start during 2017 and our intention is to also investigate CECS from a genetic perspective at some point in the future, but we will only be able to do so once samples from sufficient dogs with a robust diagnosis of the condition have been collected.

Hopefully we'll eventually be able to establish the genetic causes for each condition and provide diagnostic tools to assist in breeding programmes; however we cannot guarantee that this will be achieved within a particular time frame.

Should a DNA test be developed in the future for either CECS or SPS then any relevant results generated during the research would be reported directly to the owners of dogs that directly assisted the research. This is likely to be affected dogs and unaffected dogs over the age of 10 yo.

Donations to cover the cost of supplying swabs are certainly appreciated and additional monies we've received from concerned parties within the breed have been ring-fenced and will go a long way toward future research into both SPS& CECS in the Border terrier.

Anyone wishing to contribute samples that could potentially be of benefit towards genetic research in the breed should contact the canine genetics group at the AHT, providing a postal address and indicate the number of swab kits required. E-mail: canine.genetics@aht.org.uk or Tel: 01638 555624."