We greatly appreciate your efforts in sample collection. All new cases and controls are critically important for us to complete the study. We have so far identified two putative loci possibly associated with BT epilepsy and need new samples for confirmation and to identify the disease causing mutations. We have massive sequencing experiment ongoing in the associated loci to find the mutations.

Our vet neurologist, Tarja Pääkkönen (DVM, DiplECVN; née Jokinen) from the Department of Clinical Veterinary Sciences, University of Helsinki, has studied clinically 8 affected Finnish Border Terriers and 7 healthy controls. These studies included a complete physical and neurological examination, serum biochemistry, urinalysis, CSF examination, EEG recordings and MRI. The results of these studies indicated that the affected Border Terriers suffer from idiopathic epilepsy. Based on the pedigree analyses, there seems to be a strong genetic component behind the disease.

We aim to identify the genetic risk factors and we hope to be able to develop a genetic test for breeding purposes. In this research we need DNA samples from affected dogs, their close relatives and old, definitely also from healthy controls preferably over 7 years old without any history of seizures. To be able to get all the necessary information of each affected dog that is giving sample to our research, we have developed a detailed web-based epilepsy questionnaire. If the owners fill out the questionnaire properly, we are able to define the seizure type of each dog. If you do not have the questionnaire we can send you the link.

We start by mapping the epilepsy gene(s). This can be done by a case-control association study or a family based linkage study or by combination of these study methods. We have specific SNP array technologies for this purposes. Once we know the chromosome where the mutation lies, we try to confirm our findings in an independent sample set. If the associated region is large, it can be narrowed down by fine-mapping the region with additional markers and samples. Then we search good candidate genes from the located regions and search mutations from those genes. Also targeted sequencing can be used for sequencing of the whole associated region. If a mutation can be found, it is genotyped from a larger sample set to see whether it associates with the disease. Then other breeds are studied as well to see is the mutation breed specific. Next we study the function of the mutation; does it have an effect to the gene function. Ultimate goal is to find new epilepsy genes that can be tested from the human patients. In each study phase we select a set of suitable samples from our DNA bank.
Please remember that the next phase depends always of the results of the previous phase but this is the basic protocol that we use.

The samples controlled by our research group and owned by the University of Helsinki. Samples are stored in our lab (Canine Genetics Research Group; Biomedicum Helsinki, Helsinki). We will keep the samples virtually forever (or as long as we have the group the University). Samples won't be destroyed for any reasons.

We apply standard ethical practices established by the University. All the information from the dogs and the owners are stored in our secured database. The access for the database is restricted to our group members and all information is kept strictly confidential. We do not publish anything by name and anonymous animals cannot be recognized from the studies.

Hannes Lohi, professor
Group leader and PI of the epilepsy project