



Summer 2016

The DNA clinics have been very busy over the last few months with several new breeds doing a lot more testing. Our colleagues in Shelties and Rough Collies are holding DNA clinics at their shows and of course Border Collies are most welcome to attend. See our facebook page for details. Postal clinics will continue the next one closes at the end of August.

With regard to the CEA test offered by Animal Genetics that many owners in all 4 breeds have been using rather than the very expensive Optigen one. After over a year of trying to get the UK K.C. to record the results on the health records the K.C. have rejected this wishing to limit recording to the mutation identifying test supplied by Optigen. The U.K. K.C. will also not accept any tests done overseas unless it is done by Optigen or by 2 labs Optigen support.

The K.C. have however stated that and I quote "you are, of course, very welcome to record or report test results at the club level."

At the next breed council this will be discussed. The K.C. have also confirmed that any A.G. results sent to them will not be put on health records but be noted for internal reference. Make of that what you will???

All breeds that test for CEA wish to continue to lobby the K.C. to have the results recorded. I will update you in the next newsletter. I feel a campaign coming on.

Gonioscopy – sadly we are still receiving lots of affected results (and some going on to lose eyes) Several in Europe have been notified to Roslin and DNA samples taken. 2 new ones in the USA have also been reported. So although some think this has gone away – far from it – more and more are being reported so it is out there and we need to test!

Please record your results onto the database and for UK owners send your results to the K.C. – unlike the routine eye test the BVA do not send the gonioscopy ones to the K.C. owners have to do it themselves. It is easy – you can send a scanned copy to [HBS.Mailbox@thekennelclub.org](mailto:HBS.Mailbox@thekennelclub.org)

The PBHF with the support of Midlands BCC are holding an eye testing session on the 9<sup>th</sup> October at the KC Building at the show. We will be providing the BVA discounts plus discount on gonioscopy – contact me for details.

In terms of the research here is the update from Roslin

“The sequencing is well underway. The samples have been processed and are in the QC phase. Unfortunately, as we feared, some of them have problems with the bacterial DNA and the sequencing rate is quite low. They have told us the sample from the blood worked well. They haven’t yet released the results to us, because they want to complete the QC for all samples and give us the results as a single batch. They have said we may have them in the next week or so, but our experience in the past is that they will hold off until they can be absolutely certain that they have done their best with the samples.

We will be sending off another batch for the other analysis.

We are getting closer – I know it’s been a long haul, but hopefully we will have more clarity soon. “

Thank you to everyone that have provided DNA by blood or swab and those that have contributed to the genome.

The K.C. have 2 seminars coming up for the Health coordinators so please send any questions on any health matters to your club who can bring them to the attention of the Breed Council.

Regards  
Kathie Kinton

Welcome to this update –  
Glaucoma Research Update

A summary of the research so far from the leader of the research team at the Roslin Institute, Professor Kim Summers:-

***Goniodysgenesis and glaucoma: recessive vs polygenic inheritance.***

*There are a number of models to account for conditions that are clearly heritable (run in families). Firstly if the condition is dominant, affected offspring would always have at least one affected parent. Two affected parents could have affected and unaffected offspring. If the condition is recessive, two unaffected parents can have affected offspring but if both parents are affected all offspring should be affected. Finally if the condition is polygenic (ie several – many genes involved) there will be a familial clustering of affected individuals but there will not be simple patterns of inheritance. For example, with hip dysplasia, on average two affected parents may have about one third unaffected offspring, which does not match either single gene model. The implications of these models for breeders are different.*

**1. Dominant**

*This is easy to breed out as it is just a matter of avoiding breeding from any affected individual. However, we believe this is unlikely to be the case for goniodysgenesis or glaucoma, since there are cases where neither parent was affected and yet one or more of the offspring have failed the test.*

**2. Recessive**

*The problem here is that the undesirable genetic variant can “hide” for generations. We all inherit two copies of every gene and for a recessive the good version masks the presence of the undesirable version. Two individuals with a common ancestor can both be carrying the undesirable variant inherited from that ancestor and when mated their offspring have a 1 in 4 chance of inheriting this from both parents and hence being affected. This is why we would always recommend not mating closely related individuals. As the number of generations from the common ancestor increases the probability that both parents carry that variant decreases so the chance that two related individuals are both carriers decreases. However, that possibility is always there when the animals have a common ancestor, however far back. The data we have are largely consistent with a recessive model, although we know of situations where two affected parents have apparently had one or more unaffected offspring. At this stage we can't tell from the databases whether this is due to failure to report a fail, lack of testing of the litter or the diagnostic uncertainty of having the test done by different people. Therefore we are reluctant to say that goniodysgenesis is recessive, but clearly breeding relatives (even if unaffected) would not be desirable.*

**3. Polygenic**



## Glaucoma Research Update

Following on from Kathie Kinton's recent report to the Border Collie Breed Council what follows is a summary of the research so far from the leader of the research team at the Roslin Institute, Professor Kim Summers, and well worth reading in full before reading the rest of this posting.

### ***Goniodysgenesis and glaucoma: recessive vs polygenic inheritance.***

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We have also applied for the new CEA marker test to be accepted by the UK K.C. Before requesting this the PBHF with the Breed Council sent in a number of swabs from dogs/bitches previously tested by Opitgen to ensure that the marker test did give the same results A.G. were unaware of the names of the dogs or the results. All

The test can be done by the PBHF from anyway in the world by swab or blood.

A.G. have provided the information below on the test – we await our KC decision on inclusion on the records – those of you reading this from overseas that do not have your results recorded by your KC might want to consider this as a option.

Animal Genetics has developed a panel of six DNA markers called a "haplotype" to test for CEA. Animal Genetics is interested in developing a more comprehensive test to better distinguish those individual animals that may develop a more severe form of CEA from those that do not. Although we do not incorporate the genetic mutation Optigen claims to have an exclusive right to use into our panel, our test results using this haplotype of six DNA markers provides the same result.

The Collie Eye Anomaly test at Animal Genetics is a haplotype-based association test that has 100% linkage to the actual mutation. Because Collie Eye Anomaly is a large deletion, using multiple markers on either side of the deletion allows for such a close association. This test has been validated across all breeds known to be affected by CEA by our own process, as well as individual breed clubs and owners. Additionally, the markers also could provide more information related to the age of onset and severity of Collie Eye Anomaly with additional research. Animal Genetics is committed to the health of dogs, and is continuing to use all information provided about affected dogs to look for link between certain haplotype and the progression of the disease, as well as additional markers that could provide more useful information

The PBHF have agreed a discounted price until the end of 2015 of £44.00 per dog for the CEA test.

Following on from the last update I am pleased to report that the peer review and publication updates have now been sent in by Roslin for the pre-disposition to glaucoma and we are expecting another update in July and hope that research paper can then be released publicly.

Here is the latest email from Roslin – following the updates in the last BorderWorld  
Hi Kathie,

Just a quick update to let you know that we recently (this week) re-submitted our manuscript on Goniodysgenesis in the Border Collie to the journal.

We're hoping the journal doesn't take too long to decide whether or not they will publish our research, but before they can make that decision our paper will need to go out for peer-review, which is a process that can sometime take a while. A few months at least.

Hoping you are well and the recent summary Kim sent to you was helpful to the breeders and your members

Roslin

Recently I have been asked about the DNA test for Grey Collie Syndrome – and can confirm that A.G. can do the test and we can do it by swab at any of our clinics.

The next clinics are at BCC of Wales Ch. Show and at the East Anglian Rough Collie show – so July and August.

Our colleagues in Rough Collies are working on concerns they have regarding liver conditions and wondered if any of the Border Collie owners have experience this and if so want to assist with the research.

Here is the comment from our colleagues in Rough Collies – you can contact them via the PBHF contact button and I can pass on our see the PBHF facebook page.

We need your help. We would like to thank all those who have sent records of any collie that that been lost to Unexplained Liver problems. From the information gathered so far, we have had one of the top UK Vets, based at the RCVC Cambridge, investigate. The preliminary findings on the limited information we have has shown that the cause may NOT be wholly Genetic and that other factors may play a part in the problem. Anyone who is interested in helping, could they please supply the following information (in confidence) to the Un-Explained Liver Problems is site by Messenger on the PBHF page 1) MDR1 status. 2) Wormers used. 3) Antibiotics prescribed 4) Environment (Town of Country) 5) Diet/Feeding. 6) Nutritional Supplements, etc

I have not heard to anything in Border Collies but if you know different maybe you can help.

That's all for this update feel free to contact me if I can assist in anyway or you need further information on the PBHF and what we are doing.

Regards  
Kathie