STATEMENT ON CEA IN BEARDED COLLIES FROM THE JOINT BREED LIAISON COMMITTEE

In October 2013, Elizabeth Kershaw our Breed Health Co-ordinator published a statement in the Dog Press regarding a Bearded Collie being tested for Collie Eye Anomaly (CEA). The same statement was made available to all Breed Clubs.

The Joint Breed Liaison Committee (JBLC) have been aware for some time that the OptiGen laboratory in the USA have tested a UK KC registered Bearded Collie for CEA and that the result was positive. We have taken steps with the assistance of the Animal Health Trust (AHT) to discover the prevalence of the problem within the breed.

WHAT IS CEA?

Collie Eye Anomaly (CEA) is an inherited condition causing an abnormality at the back of the eye. It is found in a number of breeds of dogs, namely Rough and Smooth Collies, Shetland Sheepdogs, Australian Shepherds, Lancashire Heelers, Nova Scotia Duck Tolling Retrievers and Border Collies, in varying frequencies.

CEA appears to affect different breeds with varying degrees of severity. In the majority of dogs CEA is mild - there is little noticeable affect on the dogs eyesight and they lead a normal life, the condition does not worsen over time. In more severely affected dogs there is a possibility that the retina may become detached. These complications usually occur in the first couple of years of life. There is as yet no evidence that Bearded Collies have been other than mildly affected.

CEA can be diagnosed by examination of the eye by a veterinary ophthalmologist. However it can only reliably be seen in puppies between the ages of six to ten weeks of age. Later development of the eye tends to hide these features. Thus puppies diagnosed with CEA during early screening may appear to be clear on examination as adults. These dogs are still affected and carry the genes for the condition. Even litter screening will not identify carriers of the condition but only affected puppies. To identify carriers and affected adults the only reliable diagnosis is by DNA testing.

The gene responsible for CEA has been identified by geneticists and a genetic test developed which can be used to determine the status of dogs tested i.e. whether they are clear, carriers or affected. This DNA test is offered commercially by OptiGen laboratories in the USA.

An affected dog will carry two copies of the abnormal CEA gene having inherited one from each parent. A dog carrying only one copy of the gene is known as a carrier and is capable of producing affected dogs if bred to another affected dog or another carrier. If bred to a dog diagnosed as clear for CEA, statistically 50% of the litter may be carriers.

Past experience from breeders in the breeds affected by CEA seem to suggest that breeding from mildly affected animals tends to produce mildly affected pups. However, this is not always the case, and matings of mildly affected dogs have been known to produce pups with the more severe form of the condition, at risk of retinal haemorrhage, detachments and possible blindness.

Further information on CEA and its inheritance can be found at www.collieeye.org.uk

CEA AND THE BEARDED COLLIE

Until recently it was thought that Bearded Collies were free from CEA, however in August 2012 the JBLC were made aware of a report that a KC registered Bearded Collie had been referred to OptiGen for a DNA test for CEA and had been confirmed as affected.

The JBLC discussed the matter at their October meeting and learnt that the dog had been referred to OptiGen following abnormalities found at a routine eye testing session. OptiGen had never previously carried out a CEA test on a Bearded Collie and they had analysed the DNA using the test designed for other CEA affected breeds. In order to determine how widespread the gene might be in the UK Bearded Collie population the JBLC decided to approach the Animal Health Trust (AHT) for guidance and they immediately offered their help to discover the prevalence of CEA in the UK KC registered breeding population of Bearded Collies.

They suggested we analyse a sample set of 50 dogs with a wide variety of pedigrees to represent the general UK Breeding population in order to get an accurate overview of the likely frequency of CEA.

One member of the JBLC was tasked with collating pedigrees and dogs were chosen for testing to represent as many diverse bloodlines as possible. It was also decided to include a sample from the original affected dog unbeknown to the AHT. In addition samples from some of its relatives were also submitted.

Although the AHT only charged a nominal fee per dog, funding was required and this was made available from the Clubs and some of the owners of the dogs tested. The kits were sent out by the AHT to the JBLC representative who distributed them accordingly.

Sampling took place over January and February 2013 and samples from 59 dogs were sent to the AHT. Because the mutation for CEA falls under patent owned by OptiGen the AHT were unable to report individual results but they were able to present their general findings.

In May 2013 following their full analysis, the AHT sent us a report on their findings:

- 59 dogs were tested, representing 47 different sires and 57 different dams, of which 46 had unique parentage within the sample studied as deduced from their accompanying pedigrees.
- The study included the known clinically affected dog, its parents, a paternal grand dam, a dog that shares the same paternal grandparents as the affected dog and a dog that shares the same maternal grand sire as the affected dog.
- The AHT results identified a single dog to be affected for the mutation and 5 other dogs were found to be carriers.
- The AHT have advised that assuming this was a random subset of the population and that mating occurs randomly with respect to the mutation, statistics can then be used to determine an approximate percentage of clear, carrier and affected dogs in the population. From this they have estimated that 88.49% of Bearded Collies are clear, 11.16% are carriers and 0.35% are affected, i.e. 3-4 CEA affected dogs per 1000
- Because the affected dog and some of its relatives, were also included in the study, the mutation frequency in reality is likely to be lower than this figure.

WHAT DOES THIS TELL US?

Because of the way that CEA is inherited we know that both of the parents of the affected dog must be carriers and therefore account for 2 of the heterozygous tests found. They in turn must have inherited the gene from one of their parents and so on. Statistically a full sibling to the sire of the affected dog would have a 50:50 chance of being clear or a carrier. Therefore it would seem reasonable to assume that the other heterozygous dogs shared common ancestors with the affected dog.

It would appear on the current evidence of its low frequency within the population that the mutation is likely to be fairly isolated and **may** only link back to one particular dog. Should new evidence come to light then this would have to be re-evaluated.

WHAT SHOULD WE DO NEXT?

Following the AHT study we have since been informed of the identity of the dog diagnosed as affected by OptiGen which has not been bred from, nor will be. The mother of this dog (who must be a carrier) has no siblings. She only produced one other puppy (female), which has not been bred from. Both these bitches have since been spayed. The affected dog's parentage has not been verified but his sire has to be a carrier. He is therefore able to pass on the mutation to his offspring and any of his full siblings have a 50:50 chance statistically, of being carriers themselves. Even if carriers are mated to clear dogs there is still a 50:50 chance of producing carrier pups. This is not a problem **IF** the status of the dogs is known so that two carriers are **NOT** inadvertently mated together.

Knowing this we are now in a better position to:

1. Attempt to identify any other carrier or affected dogs related to the known cases so far.
2 Promote wider DNA testing and awareness within the breed to identify any other carriers or affected dogs.
3. Inform the KC of the situation for them to take any necessary steps

PROPOSED ACTIONS

That the JBLC contact all the owners of immediate relatives to the affected Bearded Collie, explain the situation to them and recommend genetic testing for CEA.

The priorities are:

- Confirm through OptiGen that the parents of the affected dog are carriers and inform all owners of those Bearded Collies sharing the same sire as the affected dog that their dog may be a carrier for CEA and should be tested IF it has been bred from or there is a possibility it will be bred from.
- Owners of dogs that are siblings to the affected dogs sire should be told that their dog may be carriers and tested IF it has been bred from or there is a possibility it will be bred from.
- Those that have pups out of siblings of the affected dogs sire should be told that their dog may be carriers and tested IF it has been bred from or there is a possibility it will be bred from unless these siblings have already been tested and found to be clear.

In addition we also recommend the following actions:

- Although the incidence of CEA is low in the general Bearded Collie population the JBLC strongly recommend eye testing for all Bearded Collies. This may pick up some abnormalities that warrant further testing, however as previously explained a clear result does not necessarily mean that the dog is free from CEA.
- Members with any particular concerns should consider DNA testing through OptiGen – particularly if they know of dogs going blind in their line at perhaps a slightly earlier age than one would expect - and not just put it down to 'old age'!
- The JBLC should inform the KC about the affected dog so that they put Bearded Collies on the CEA affected list of breeds.
- The JBLC should approach the KC to see if funding is available to assist further genetic testing.

HOW TO TEST

CEA testing is offered by OptiGen (www.optigen.com) at a cost of $180 plus the cost of shipping. There are other labs that offer CEA testing but ultimately the samples all get sent to OptiGen as they own the patent for the test. IDEXX laboratories (www.idexx.co.uk) offer CEA testing at £120 plus VAT, price correct till 31st Dec. Blood samples are preferred so your vet's fee for this will have to be added to your costs. The JBLC are looking at other options to reduce the cost of the test by participating in a 20/20 clinic (for every 20 dogs sampled, 20% discount per test) but this may not take place till February at the earliest.

Any person wishing to discuss this further should contact their Breed Club Joint Breed Liaison representative for further advice

Paula Brooks - Chairman

Alan Hards - Secretary

or

Elizabeth Kershaw - Breed Health Representative

Margaret Buckley - Chair JBLC

Yvonne Fox - Secretary JBLC