

## **Collie Eye Anomaly (CEA)**

BeaCon and Beardies of the World recently sponsored genetic testing for Collie Eye Anomaly (CEA) in 175 Bearded Collies, a further 4 dogs were tested privately by their owners. CEA is a congenital, inherited condition affecting both eyes and seen in several breeds – not all of them collies. During development the choroid, the blood vessel rich layer of the eye between the retina with its light responsive rod and cone cells and the sclera (white of the eye) fails to develop properly, and a pale hole (coloboma) can be seen. Depending upon how badly the eyes are affected the dog may have only minor visual defects or be completely blind. In some cases retinal and scleral colobomas are also apparent on eye examination. There is no cure.

CEA is inherited as an autosomal recessive with close to 100% penetrance. The mutation is located on chromosome 37. For a dog to have symptoms of the disease it must inherit two copies of the gene one from each parent. Dogs that carry a single copy of the gene are carriers for CEA and will pass this gene on to approximately 50% of their offspring. Recently a dog was reported in the UK which suffered from CEA. He was not badly affected by the condition and was not completely blind. Many owners and breeders in the UK started testing their Beardies.

We have received a report from the company that did the testing for us - Paw Print Genetics. 139 test kits have been returned. Of those 139 dogs, none are affected with CEA (two copies of the gene) but there were 16 carriers (with a single copy of the gene). That means 11.5% of the dogs sampled are carriers of CEA and 88.5% are clear of the illness. In Rough and Smooth Collies incidence of the carrier CEA gene can reach 95% with 80-85% of dogs affected. In other breeds, as with Beardies, incidence is much lower, and it would be wise if we can keep it that way.

### **What does this mean for breeders?**

- If the sire and dam are clear of CEA all their offspring will also be clear.
- If either the sire or dam is a carrier and the other parent is clear none of the offspring will be affected but about 50% will carry the mutated gene.
- If both parents are carriers about 25% of the offspring will be affected, about 50% will be carriers and about 25% will be clear.
- If one parent is affected and the other a carrier ~ 50% of the litter would be affected and 50% carriers
- If one parent is affected and the other parent is clear, all the offspring will be carriers.

### **Recommendations**

- Given the prevalence of CEA in Beardies, test all potential breeding animals unless both of their own parents have tested CEA clear – in which case their offspring will be clear
- Do not breed a carrier to a carrier
- Be extremely cautious about breeding carrier to a clear. While it is true that none of the offspring would be affected, approximately half would be carriers. Over time this will increase the incidence of carriers in the population and that makes it more difficult to find non-carriers to breed to.
- Where possible use a clear relative of a carrier dog or bitch with a phenotype (appearance, structure, etc.) similar to the carrier.
- Test all dogs from parents of unknown genetic CEA status.
- Regain dogs for breeding that are clear – all else being equal.

**Given the incidence of carriers detected in the evaluations to date, it is likely that there are undetected affected Beardies in North America.**

**Ophthalmic Diagnosis of CEA.** Before the genetic test for CEA became available the only way to identify dogs with CEA was to examine the retina and choroid of young puppies for hypoplasia (colobomas). These can be seen readily in very young animals, but in many dogs by three to seven months the coloboma may become too heavily pigmented to diagnose the condition. For this reason, puppies must be checked before 7 weeks of age. However, dogs with dilute coats have a choroid that also lacks pigment and CEA might be misdiagnosed in these animals.

It is only possible to diagnose an affected puppy when the exam is done before 7 weeks. **It is not possible to diagnose a carrier at any age with a visual exam.** Ophthalmic exam is also susceptible to false positives and negatives

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