East Anglian Collie Association

DNA testing for Collie Eye Health

Collie Eye Anomaly (CEA/CH)

The CEA clinical test tends to be somewhat subjective and, especially with blue merles, often misdiagnosed.

When a breeder produces CEA clinically clear pups from two clinically clear parents (both also tested clear as baby puppies), there is a small possibility that one or more of the offspring might be genetically clear-eyed. However, this can only be proved by having the whole litter DNA tested.

CEA/CH genetics

The DNA test for CEA can only determine the Choroidal Hypoplasia (CH) aspect of the disease. However, it is strongly suggested that those dogs DNA-tested **clear** of CEA do not suffer from other CEA-related conditions such as colobomas or detached retinas. It is believed colobomas and detached retinas may be carried on a different gene mutation which only co-exists with the gene mutation for CH, so only CEA-affected Collies can present with these more serious conditions.

However, until this is confirmed, it is imperative that breeders continue having their seven week old puppies clinically examined for CEA to eliminate the possibility of colobomas and/or detached retinas. Any CEA clinical clears could then be subsequently DNA tested.

A genetically **clear-eyed** dog (homozygous normal) receives two copies of the normal CEA/CH gene, one from each parent. Breeding two homozygous clear dogs together will therefore produce all normal-eyed or genetically clear offspring.

A genetically clear-eyed **'carrier'** (heterozygous normal) receives a normal copy of the CEA/CH gene from one parent and a mutant copy from the other parent. The normal copy of the gene is the dominant one and it masks the recessive mutant gene. Such an animal will therefore be diagnosed clear when clinically examined by ophthalmoscope. When a carrier is mated it randomly passes on copies of both the normal and mutant genes to its offspring, so some will be affected and some clear or normal-eyed. Breeding a carrier to a carrier could initially produce 25% clears, 50% carriers and 25% affected puppies in a litter, although these ratios are related to 100+ puppies!

A CEA **'affected'** dog (homozygous recessive) receives one copy of the defective recessive CEA/CH gene from each of its parents, so therefore breeding two
homozygous recessives together will produce all homozygous recessive (affected) puppies.

The Optigen laboratory in the USA was the first laboratory to introduce a DNA test for CEA some years ago and, as they claimed to have world-wide rights to the protocol, the KC will only publish CEA test results from this company despite several other laboratories offering the same test at a cheaper rate.

However, in October 2014, Animal Genetics claimed it had developed an equally robust, but different, test -

'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'.

Several Border Collie Breeders, whose dogs had been previously CEA-tested by Optigen, have had these same dogs re-tested using the Animal Genetics test, and the results were identical. This data has recently (March 2015) been submitted to the KC by the Pastoral Breeds Health Foundation, with the request that Animal Genetics be an acceptable alternative to Optigen - we await results!

N.B The Optigen CEA test currently costs in excess of £100 per dog whilst Animal Genetics charges £55.00!

Pat Hutchinson, EACA Health Coordinator, ©2015