Collie Eye Anomaly (CEA)/ Choroidal Hypoplasia (CH)

- **performed by Optigen**

Affected breeds:
Australian Shepherd
Bearded Collie
Border Collie
Lancashire Heeler
Nova Scotia Duck Tolling Retriever
Rough Collie
Shetland Sheepdog
Smooth Collie
Whippet

Collies share Collie Eye Anomaly (CEA) with several other breeds – it's not just a problem for collies. CEA is more technically known as Choroidal Hypoplasia (CH). It is a recessively inherited eye disorder that causes abnormal development of the choroid - an important layer of tissue under the retina of the eye. This disease is seen most frequently in U.S. collies, but also worldwide in Rough and Smooth Collies, Border Collies, Australian Shepherds, Lancashire Heelers, and Shetland Sheepdogs. Since the choroid layer does not develop normally from the start, the primary abnormality can be diagnosed at a very young age. Regrettably, there is no treatment or cure for CEA.
Additional extensive information about the CEA test can be found on the Optigen website at **http://www.optigen.com/opt9_test_cea_ch.html**

CEA is caused by a recessive genetic mutation. This means that dogs which carry the mutation ("CARRIERS") are normal but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will develop CEA ("AFFECTED").

This test is particularly useful for breeders:

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- To conclusively confirm Progressive Retinal Atrophy caused by CEA

This test will be reported as:
CLEAR : no evidence of the CEA mutation
CARRIER : carries one copy of the defect, which will be passed to 50% of offspring
AFFECTED : carries two copies of the defect, and will have CEA

The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:
CLEAR X CLEAR = 100% CLEAR
CARRIER X CLEAR = 50% CARRIER, 50% CLEAR
CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR

References
Lowe JK, Kukekova AV, Kirkness EF, Langlois MC, Aguirre GD, Acland GM, Ostrander EA. Linkage Mapping of the Primary Disease Locus for Collie Eye Anomaly. Genomics 2003 Jul;82(1):86-95.