

# IS THE COLLIE-EYE ANOMALY (CEA) GENE TEST USEFUL FOR NOVA SCOTIA DUCK TOLLING RETRIEVERS?

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The gene test for Collie-Eye anomaly (commonly called CEA) was one of the first genetic tests developed for dogs. Collie-eye anomaly is a descriptive term for a range of disorders affecting the canine eye. The test has been recommended for many dog breeds, including the toller. The test considers a deletion in a non-protein coding portion of a gene. These kinds of mutations are notoriously difficult to prove as leading to functional change.

At the minor end of the abnormality spectrum associated with CEA is choroidal hypoplasia. Choroidal hypoplasia refers to abnormalities in the development or pigmentation of the choroid. The choroid is a bed of blood vessels beneath the retina which is the disc that receives light signals at the back of the eye. This is the most common abnormality found in Collie eyes. It is the least harmful and least severe form of CEA. ([www.colliehealth.org](http://www.colliehealth.org)).

At the severe end of the disease spectrum is a disorder coloboma of the optic nerve head (ONH) which can result in blindness on occasion. The optic nerve transmits light signals from the retina to the brain to be interpreted as visual signals (sight). Damage to the optic nerve can cause blindness. The term coloboma describes the incomplete formation of this nerve and the optic disc in the back of the eye.

Recent research conducted by the Bannasch laboratory at the University of California, Davis, collected DNA samples from seven tollers that had the ONH severe form of CEA. Using the same gene mapping strategies that we use to make other genetic tests (such as for JADD and cleft-palate), the research compared the DNA of the seven severely affected tollers with the DNA of normal tollers. Clearly, the condition ONH exists in tollers as affected dogs were identified in our breed. However, there was no correlation between the CEA test result and the appearance of ONH in the tollers analysed. This suggests that the DNA test is NOT helpful in determining tollers that might suffer blindness from CEA.

The conclusion of the analysis was that ophthalmic examination of puppies was a more efficient method for selecting away from the severe outcomes associated with CEA than the use of the currently available genetic test. The researchers did not test dogs with the milder forms of CEA that do not cause blindness. The researchers noted that Choroidal hypoplasia by itself has not been proven to result in blindness or clinically detectable visual deficits; therefore, there is little justification to select against it per se unless it is associated with ONH coloboma. The lead researcher in this study is a breeder of NSDTR.

The full paper can be found here: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5766432>

For those interested to read the paper the terms are used (but not explained):

OS - left eye, OD - right eye, OU - both eyes