

## **Genetic testing for Collie Eye Anomaly (CEA) in Rough Collie, Smooth Collie, Border Collie, Australian Shepherd, Lancashire Heeler, Shetland Sheepdogs and Longhaired Wippet**

### **CEA-The disease**

Collie eye anomaly (CEA, also known as Choroidal Hypoplasia (CH)) is not just a problem for collies. The disease is seen in Rough Collie, Smooth Collie, Border Collie, Australian Shepherd, Lancashire Heeler, Shetland Sheepdogs and Longhaired Wippet. CEA is a hereditary canine ocular disorder in which the pattern of chorioretinal and scleral development is variously disturbed.

The major change, which is present in dogs with CEA, is hypoplasia (underdevelopment) of the choroid, an important layer of the eye underlying the retina. A Coloboma, or hole, may form in or near the optic disc as part of the CEA extended phenotype.

The degree of these abnormalities varies between individual dogs. It can be a mild disease or cause blindness. The abnormality can be diagnosed at a very young age, but there is no treatment or cure for CEA.

CEA is not a progressive disease. The essential features, choroidal hypoplasia and coloboma develop as the eye develops. When ocular development is complete these features are also stationary.

### **CEA-The mutation and inheritance**

The mutation which has been suggested to cause CEA has recently been published by the group of Elaine A. Ostrander at the University of Pennsylvania, USA.

A 7.8 kb deletion in the NHEJ1 gene cosegregates with CEA across multiple dog breeds. This region spans a highly conserved binding domain to which several developmentally important proteins bind.

CEA is inherited as an autosomal recessive trait. Both parents must carry at least one copy of the defect gene for the offspring to be affected.

So there are three conditions a dog can be: it can be clear (genotype N/N or homozygous normal) meaning that it does not carry the mutation and will not develop CEA. Since it also cannot pass the mutation onto its offspring, it can be mated to any other dog.

A dog which has one copy of the defect gene and one copy of the normal gene is a so-called carrier or heterozygous (genotype N/CEA); while it will not be affected by CEA, it can pass the mutation onto its offspring and should therefore only be mated to clear dogs.

Dogs that develop CEA have two mutated gene copies (genotype CEA/CEA or homozygous affected); they will always pass the mutated gene onto their offspring and should also be mated only to clear dogs.

## **CEA-The DNA test**

By DNA testing, the responsible mutation can be shown directly. This method provides a very high accuracy test and can be done at any age. It offers the possibility to distinguish not only between affected and clear dogs, but also to identify clinically healthy carriers. This is an essential information for controlling the disease in the breed, as carriers are able to spread the disease gene in the population, but can not be identified by means of common laboratory diagnostic.

To ensure maximum test reliability, the test is always performed in two independent test runs per sample. This test allows a breeder to control frequency of the defect gene in the Collie breed thus preventing the production of puppies affected with Collie Eye Anomaly.

The DNA test does not provide informations about onset of clinical signs and the severity of disease symptoms.

## **Requirements**

The test is performed out of EDTA whole blood (0.5 ml) or special cytobrushes. These cytobrushes are available upon request and free of charge. Testing is performed a few times per week. The results are due about 3-5 working days after arrival of the sample in our laboratory.

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