Collie Eye Anomaly (CEA) is a canine hereditary ocular disorder affecting development of the choroid and sclera. The disease segregates in several dog breeds including Rough and Smooth Collies, Border Collies, Australian Shepherds, Lancashire Heelers, and Shetland Sheepdogs. The clinical phenotype varies significantly among affected dogs of all breeds. The primary CEA phenotype, choroidal hypoplasia (CH), is characterized by regional hypoplasia (underdevelopment) of the choroid, which is the highly vascularized bed of the eye that underlies the retina. This lesion usually results in an ophthalmoscopically detectable window defect in the ocular fundus located temporal to the optic nerve.

This invention has identified the mutation associated with the CEA disease. Specifically, the critical region in which the CEA gene is located is an approximately 100 kb interval on chromosome 37. Within this interval, a 7.8 kilobase deletion associated with dogs that are heterozygous for the deletion (carriers), or are homozygous for the deletion (affected with CEA), was identified.

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The OptiGen test for CEA/CH provides a powerful management tool for the breeder. This genetic test can distinguish all three genetic states – normal, carrier and affected. With this information, the breeder can plan matings that avoid producing any affected dogs by always selecting one parent that is normal. The other parent can be normal, carrier or even affected, and no affected dogs will result. This breeding recommendation is a big step forward, especially for breeds and countries where frequency of CEA/CH is much lower. Earlier advice cautioned against breeding affected dogs, their parents, their offspring or their siblings (unless eye exams before 3 months of age demonstrate the sibling is unaffected).