Collie eye causing various degrees of blindness

Collie EyeAnomaly / Choroidal Hypoplasia (CEA) is a recessive disease which impairs the properformation of a layer of cells below the retina of the eye causing variousdegrees of blindness and is associated with a 7. 8-kilo base pair (bp) deletion in the canine NHEJ1gene (Parker and others 2007). There is no treatment or cure.

Just over 2% ofBorder Collies will present with chorodial hypoplasia, about a half of apercent will have colombomae, and less than 1 in 1, 000 will have retinaldetachment. Collie Eye Anomaly (CEA) was first described inrough collies in 1953 B. This is a canine genetic ocular disorder whichmanifests as regional hypoplasia of the choroid, layer of the eye that suppliesblood and nutrients to the retina A. Ophthalmoscopically detectable defect inthe ocular fundus located temporal to the optical nerve with possiblecolobomatous lesions which can be present as pits within optic nerve head or inthe adjacent fundus. Some affected individuals retain normal visual functionthroughout the life, but severely affected dogs with colobomas can developdetachments that lead to blindness.

Examination of theretina of the eye as early as 5-10 weeks of age can diagnose CEA. After 12weeks of age, it is hard to examine, because the lesion can be covered overwith pigment. Examination of these dogs can only confirm affected dogs, but notcarriers since this condition is inherited as recessive trait. Carriers andhomozygotes can both be confirmed by polymerase chain reaction (PCR) baseddiagnostic test, which can be the most definitive method for detecting andmonitoring inherited diseases like CEA. CEA has becomewidely spread in many breeds and has been a problem for a long time, partlybecause of the poor acknowledgement of clinical

significance of CEA and parlybecause some breeders considered that controlling breeding was unnecessarybecause of the low incidence of blindness related to CEA C. Recently, avery high frequency of the CEA mutation has been reported in the Hokkaido inu, a traditional Japanese breed, which is the first identification of a breed withCEA predisposition that is not classified as collies or sighthounds (Mizukamiand others 2012a).

A series of genetic and linkage mappingstudies were reported, in naturally occuring and experimentally derived CEApedigrees which localize the primary CEA trait to a 3. 9-cM interval on CFA37, defined by markers FH4306 and AHTh174 (linkage mapping of the primary disease infolder). The mutation cosegragating with CEA in Border Collies has beenidentified as a homozygous intronic deletion of 7799 base pairs in the NHEJ1 gene on chromosome 37 D.