

Cone Degeneration (CD)

Affected breeds: Alaskan Malamute, Australian Shepherd

Cone Degeneration causes day blindness due to the degeneration of the cone-shaped light receptors in the eye which respond to bright daylight. An affected dog will begin to show signs of problems with vision at 8-12 weeks of age. These pups become day-blind and exposure to bright light is irritating or even painful. An affected dog's night vision is unaffected by CD.



Cone Degeneration 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 Cone Degeneration ("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 Cone Degeneration in an affected dog

This test will be reported as:

CLEAR : no evidence of the Cone Degeneration 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 develop Cone Degeneration

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

AFFECTED X CLEAR = 100% CARRIER

CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR

AFFECTED X CARRIER = 50% AFFECTED, 50% CARRIER

AFFECTED X AFFECTED = 100% AFFECTED

References

Yeh CY, Goldstein O, Kukekova AV, Holley D, Knollinger AM, Huson HJ, Pearce-Kelling SE, Acland GM, Komaromy AM (2013) Genomic deletion of *CNGB3* is identical by descent in multiple canine breeds and causes achromatopsia. BMC Genetics 14:27