VETERINARY TECHNICAL DATASHEET

Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shorthaired Pointer

Mutation Found In :German Shorthaired Pointer

Disorder Type

- Eye

Disease Severity

- Moderate

Background

Cone degeneration (CD), also called "day-blindness" is an inherited eye disorder causing light-sensitivity (photophobia) and an inability to see in bright light. It is very similar to a genetic condition called achromatopsia that causes day-blindness and color-blindness in humans.

Key Signs

- Day-blindness
- Photophobia

Clinical Description

Clinical signs of CD occur at the age of 8 to 12 weeks. Affected pups show signs of day-blindness and photophobia due to the degeneration of cone cells in the retina. Cone cells are gradually lost and may be completely absent in an adult dog affected with CD. The degeneration does not affect rod cells, therefore vision in dim light remains normal; cone degeneration does not result in complete blindness.

Mode of Inheritance

- autosomal recessive

Gene Name

- CNGB3

Next Steps

Affected dogs may find exposure to bright light irritating or even painful, so exposure should be limited where possible. However, their vision at night and in dim lighting should remain intact.

References

Sidjanin DJ, Lowe JK, McElwee JL, Milne BS, Phippen TM, Sargan DR, Aguirre GD, Acland GM, Ostrander EA. Canine CNGB3 mutations establish cone degeneration as orthologous to the human achromatopsia locus ACHM3. Hum Mol Genet 11(16):1823-33, 2002.

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