

VETERINARY TECHNICAL DATASHEET

Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shepherd Dog



Mutation Found In :German Shepherd Dog

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

- CNGA3

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

Tanaka N, Dutrow EV, Miyadera K, Delemotte L, MacDermaid CM, Reinstein SL, Crumley WR, Dixon CJ, Casal ML, Klein ML, Aguirre GD, Tanaka JC, Guziewicz KE. Canine CNGA3 Gene Mutations Provide Novel Insights into Human Achromatopsia-Associated Channelopathies and Treatment. PLoS One 10:e0138943, 2015.