VETERINARY TECHNICAL DATASHEET

Cone-Rod Dystrophy 1, (crd1); mutation originally found in American Staffordshire Terrier



Mutation Found In: American Staffordshire Terrier

Disorder Type

Eye

Disease Severity

Moderate

Background

The onset of this disorder involves the reduction of cone cells required for day vision and continues with the reduction of rod cells necessary for night vision.

Key Signs

- Severe retinal degeneration
- Blindness

Clinical Description

crd1 is a progressive eye disorder that typically causes retinal cell degradation and thinning of the retina, often visible at 3 to 6 months of age. Typically, clinical signs of this fast-progressing disease will appear in puppyhood and vision will be severely affected by one year of age, leading to blindness in the juvenile dog.

Mode of Inheritance

autosomal recessive

Gene Name

• PDF6B

Next Steps

Early on, affected dogs should not be exposed to bright light as it can be irritating or even painful. The diagnosis of crdl can be confirmed by performing a fundic exam to evaluate the retina. Treatment is supportive.

References

Goldstein O, Mezey JG, Schweitzer PA, Boyko AR, Gao C, Bustamante CD, Jordan JA, Aguirre GD, Acland GM. IQCB1 and PDE6B mutations cause similar early onset retinal degenerations in two closely related terrier dog breeds. Invest Ophthalmol Vis Sci54:7005-19, 2013.

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