

VETERINARY TECHNICAL DATASHEET

Cone-Rod Dystrophy 2, (crd2); mutation originally found in American Pit Bull Terrier



Mutation Found In :American Pit Bull 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

crd2 is a progressive eye disorder characterized by degeneration of photoreceptor cells and retinal thinning at a young age impacting bright light vision first followed by dim light vision. The condition progresses rapidly and the loss of vision is usually noticed in puppyhood. Retinal degeneration in crd2 generally leads to severe loss of vision by one year of age and to total blindness in early adulthood. Retinal changes are usually observable by 3 to 6 months of age.

Mode of Inheritance

- autosomal recessive

Gene Name

- IQCB1

Next Steps

Early on, affected dogs should not be exposed to bright light as it can be irritating or even painful. The diagnosis of crd2 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 Sci 54:7005-19, 2013.