

## VETERINARY TECHNICAL DATASHEET

Canine Multifocal Retinopathy 1, (CMR1); mutation originally found in Mastiff-related breeds



Mutation Found In :Bulldog (American), American Pit Bull Terrier, Australian Shepherd, Boerboel, Bulldog (Standard), Bullmastiff, Cane Corso, Dogue de Bordeaux, French Bulldog, Great Pyrenees, Mastiff, Miniature American Shepherd, Presa Canario

### Disorder Type

- Eye

### Disease Severity

- Mild/moderate

### Background

Canine multifocal retinopathy 1 (CMR1) is an inherited eye disease found in multiple breeds. It is characterized by several localized, round, bullous alterations of variable size and location in the retina at the back of the eye that cause retinal decay.

### Key Signs

- Retinal degeneration

### Clinical Description

Typically, the first ocular fundus changes in CMR1 can be diagnosed by the age of four months. In many cases, the lesions may appear to heal or even go away, sometimes leaving no evidence or only a wrinkle at the site of the healed lesion. In almost all cases, lesions from CMR1 do not progress significantly over time, so there is generally no reduction in eyesight though more serious cases could exhibit vision impairment. Very seldom is the patient completely blinded.

### Mode of Inheritance

- autosomal recessive

### Gene Name

- BEST1

### Next Steps

Monitor fundus changes for evidence of healing and monitor patient for any signs of visual impairment.

## References

Guziewicz KE, Slavik J, Lindauer SJ, Aguirre GD, Zangerl B. Molecular Consequences of BEST1 Gene Mutations in Canine Multifocal Retinopathy Predict Functional Implications for Human Bestrophinopathies. Invest Ophthalmol Vis Sci 52:4497-505, 2011.

Guziewicz KE, Zangerl B, Lindauer SJ, Mullins RF, Sandmeyer LS, Grahn BH, Stone EM, Acland GM, Aguirre GD. Bestrophin gene mutations cause canine multifocal retinopathy: a novel animal model for best disease. Invest Ophthalmol Vis Sci 48:1959-67, 2007.