Canine Multifocal Retinopathy (CMR1)

Affected breeds: Australian Shepherd, Bulldog, Bullmastiff, Dogue de Bordeaux, French Bulldog, Mastiff and Pyrenean Mountain Dog.

A very similar condition occurs in the Coton de Tulear, but this is caused by a different mutation in the same gene known as CMR2.

Canine Multifocal Retinopathy 1 (CMR1) is an inherited eye disease in which the retina degenerates. Under ophthalmoscopic examination affected dogs will display multiple orange/brown lesions on the retina. The condition develops in young dogs – typically before 4 months of age, and may progress slowly, may appear to heal, may disappear and then return. Some lesions disappear with no trace, while some leave a wrinkled, folded patch and are often described as retinal dysplasia or retinal folds.



CMR1 is caused by a recessive genetic mutation. This means that dogs which carry the mutation ("CARRIERS") are normal but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will develop CMR1 ("AFFECTED").

This test is particularly useful for breeders:

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- o To conclusively confirm CMR1 in an affected dog

This test will be reported as:

CLEAR : no evidence of the CMR1 mutation

CARRIER: carries one copy of the defect, which will be passed to 50% of offspring

AFFECTED: carries two copies of the defect, causing CMR

The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:

CLEAR X CLEAR = 100% CLEAR

CARRIER X CLEAR = 50% CARRIER, 50% CLEAR

AFFECTED X CLEAR = 100% CARRIER

CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR

AFFECTED X CARRIER = 50% AFFECTED, 50% CARRIER

AFFECTED X AFFECTED = 100% AFFECTED

References

Guziewicz KE, Zangerl B, Lindauer SJ, Mullins RF, Sandmeyer LS, Grahn BH, Stone EM, Acland GM, Aguirre GD (2007) Bestrophin gene mutations cause canine multifocal retinopathy: a novel animal model for best disease. Invest Ophthalmol Vis Sci. May;48(5):1959-67.

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