Progressive Retinal Atrophy – Type 1 Papillon (Pap_PRA1)

Affected breeds: Papillon

Progressive Retinal Atrophy (PRA) is a common cause of blindness in dogs, which results from a gradual deterioration in the light-sensitive cells in the retina at the back of the eye. A number of different causal mutations have been found in specific breeds. Specifically a mutation in the CNGB1 gene causes PRA in the Papillon. This is a late-onset PRA, with typical onset at 5-6 years – initial signs include difficulty in dim light followed by a slow disease progression.

Pap-PRA1 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 Pap-PRA1 ("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.
- To conclusively confirm Pap-PRA1 in an affected dog

This test will be reported as:

CLEAR : no evidence of the Pap-PRA1 mutation
CARRIER : carries one copy of the defect, which will be passed to 50% of offspring
AFFECTED : carries two copies of the defect, causing Pap-PRA1

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

Ahonen SJ, Arumilli M, Lohi H. A CNGB1 Frameshift Mutation in Papillon and Phalène Dogs with Progressive Retinal Atrophy. Krahe R, ed. PLoS ONE. 2013;8(8):e72122. doi:10.1371/journal.pone.0072122.