Progressive Retinal Atrophy (PRA3)

Affected breeds: Tibetan Spaniel, Tibetan Terrier

Progressive Retinal Atrophy (PRA3) results in the degeneration of the photoreceptor cells of the retina, resulting in vision loss and eventually complete blindness. As with many other dog breeds, PRA3 is one of several mutations in both the Tibetan Spaniel and Tibetan Terrier which causes PRA.

PRA3 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 PRA3 ("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 PRA3 in an affected dog

This test will be reported as:

CLEAR : no evidence of the PRA3 mutation
CARRIER : carries one copy of the defect, which will be passed to 50% of offspring
AFFECTED : carries two copies of the defect, causing PRA3

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

AFFECTED X AFFECTED = 100% AFFECTED
AFFECTED X CARRIER = 50% AFFECTED, 50% CARRIER
CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR
AFFECTED X CLEAR = 100% CARRIER
CARRIER X CLEAR = 50% CARRIER, 50% CLEAR
CLEAR X CLEAR = 100% CLEAR

References

Downs LM, Mellersh CS. An Intronic SINE insertion in FAM161A that causes exon-skipping is associated with progressive retinal atrophy in Tibetan Spaniels and Tibetan Terriers. PLoS One. 2014;9(4):e93990.