Progressive Retinal Atrophy (rcd4)

Affected breeds: English Setter, Gordon Setter, Irish Red and White Setter, Irish Setter, Miniature Poodle, Standard Poodle, Tibetan Terrier

Progressive Retinal Atrophy (PRA-rcd4) 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, PRA-rcd4 is one of several mutations in these breeds causing PRA.

PRA-rcd4 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 PRA-rcd4 ("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 PRA-rcd4 in an affected dog

This test will be reported as:

CLEAR : no evidence of the PRA-rcd4 mutation
CARRIER : carries one copy of the defect, which will be passed to 50% of offspring
AFFECTED : carries two copies of the defect, causing PRA-rcd4

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

Downs, L.M., Bell, J.S., Freeman, J., Hartley, C., Hayward, L.J. and Mellersh, C.S. (2013), Late-onset progressive retinal atrophy in the Gordon and Irish Setter breeds is associated with a frameshift mutation in C2orf71. Anim Genet, 44: 169-177.