

## Progressive Retinal Atrophy (*prcd*-PRA)

### Affected breeds:

American Cocker Spaniel, Australian Cattle Dog, Australian Shepherd, Bolognese, Chesapeake Bay Retriever, Chihuahua, Chinese Crested, Cocker Spaniel, Cocker Spaniel crosses, German Spitz, Giant Schnauzer, Golden Retriever, Labrador crosses, Labrador Retriever, Miniature Poodle, Nova Scotia Duck Tolling Retriever, Poodle crosses, Portuguese Water Dog, Schipperke, Spanish Water Dog, Standard Poodle, Toy Poodle, Yorkshire Terrier

The genetic disorder *prcd*-PRA causes cells in the retina at the back of the eye to degenerate and die, even though the cells seem to develop normally early in life. The "rod" cells operate in low light levels and are the first to lose normal function. Night blindness results. Then the "cone" cells gradually lose their normal function in full light situations. Most affected dogs will eventually be blind. Typically, the clinical disease is recognized first in early adolescence or early adulthood. Since age at onset of disease varies among breeds. Diagnosis of retinal disease can be difficult. Conditions that seem to be *prcd*-PRA might instead be another disease and might not be inherited. OptiGen's genetic test assists in making the diagnosis. It's important to remember that not all retinal disease is PRA and not all PRA is the *prcd* form of PRA. Annual eye exams by a veterinary ophthalmologist will build a history of eye health that will help to diagnose disease. Unfortunately, at this time there is no treatment or cure for PRA.



*prcd*-PRA 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 *prcd*-PRA ("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 Progressive Retinal Atrophy caused by *prcd*-PRA

### This test will be reported as:

**CLEAR** : no evidence of the *prcd*-PRA mutation

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

**AFFECTED** : carries two copies of the defect, and will have *prcd*-PRA

### 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

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

### References

Zangerl B, Goldstein O, Philp AR, Lindauer SJ, Pearce-Kelling SE, Mullins RF, Graphodatsky AS, Ripoll D, Felix JS, Stone EM, Acland GM, Aguirre GD. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006;88:551–563. doi: 10.1016/j.ygeno.2006.07.007.