

## Day Blindness/Retinal Degeneration DB/RD Test

Description from the OptiGen website, written by Dr. Gustavo Aguirre

The support of the **Poodle Club of America Foundation** was critical to a successful collaboration between OptiGen and researchers at the University of Pennsylvania School of Veterinary Medicine that led to the discovery of a mutation causing day blindness and subsequent retinal degeneration in the Standard Poodle. Multiple cases of day blindness in this breed were submitted for investigation to [OptiGen's Free DNA testing/Research program](#). After OptiGen DNA testing revealed that none of the mutations previously known to cause day blindness in other breeds were responsible for the clinical signs observed in the Standard Poodle cases, the project was turned over to [Dr. Karina Guziewicz](#), Research Assistant Professor of Ophthalmology at the University of Pennsylvania School of Veterinary Medicine. Dr. Guziewicz is part of the team led by [Dr. Gustavo Aguirre](#) and that has included a consortium of researchers who, over the years, have discovered mutations that comprise the majority of OptiGen's DNA tests for inherited eye diseases in dogs. Dr. Guziewicz carried out genomic analyses on the day blind Standard Poodles that led to discovery of a mutation in one of the genes critical to retinal function. Further research aimed at developing therapeutic strategies for DB/RD in Standard Poodles is ongoing.

### **Clinical signs/Disease description:**

Day blindness, also known as achromatopsia, is characterized by a failure of cone cells in the retina to function properly. Cone cells are responsible for vision in bright light conditions while their retinal counterparts, the rod cells, function in dim light. Similar to signs observed by owners of [Alaskan Malamutes, Australian Shepherds or German Shorthaired Pointers affected with Cone Degeneration](#) or [Labrador Retrievers with Achromatopsia](#) and German Shepherds caused by other mutations, Standard Poodle puppies with the DB/RD form of day blindness manifest signs of poor vision in bright light but initially retain normal vision in low light levels. However unlike other forms of Day Blindness observed in other breeds, the DB/RD mutation causes a more complete retinal degeneration in the Standard Poodle and affected dogs eventually lose both cone and rod cell function resulting in vision loss under all lighting conditions.

### **DB/RD mutation based test:**

A DNA-based test has been developed to target the mutation responsible for Day Blindness/Retinal Degeneration (DB/RD) in the Standard Poodle. The DB/RD test can be used to determine the genetic status of a dog in respect to the mutation and to make informed decisions about mating options to prevent the occurrence of this undesirable blinding disorder in progeny.

The DNA-based DB/RD test allows genotype determination of a tested dog as being:

**NORMAL** - the dog has two copies of the normal gene and it is not going to develop Day Blindness/Retinal Degeneration caused by the DB/RD mutation.

**CARRIER** - the dog has one copy of the normal gene and one copy of the DB/RD mutation. The dog will not develop clinical signs of Day Blindness/Retinal Degeneration due to the DB/RD mutation but it will transmit one copy of the DB/RD mutation to about 50% of its progeny.

**AFFECTED** - the dog has two copies of the DB/RD mutation and is expected to develop/is already showing signs of the Day Blindness/Retinal Degeneration caused by the DB/RD mutation.

**Testing and Breeding recommendations:**

By selecting the mate with the appropriate genotype, it is possible to breed affected or carrier dogs and never produce affected progeny. In this manner the genetic diversity of the breed can be maintained.