

Optigen Press Release

June 1, 2005

Ithaca, NY: OptiGen, LLC, proudly announces identification of the gene causing canine *prcd*-PRA (progressive rod-cone degeneration form of Progressive Retinal Atrophy). The cause of this inherited blinding disease, occurring in at least a dozen purebreeds, is one specific mutation in the coding sequence of the gene. OptiGen now offers a direct gene mutation test (mutation test) to detect the *prcd*-PRA status of any dog among affected breeds.

This success results from years of extensive investigation by Dr. Gus Aguirre and Dr. Greg Acland and their research staff at Cornell University. Essential cooperation from multiple breed clubs and more recent participation by OptiGen completed the team. The research data defining this mutation are being prepared for publication in a scientific journal. Commenting on future work, Dr. Aguirre says: "Going forward, the biological action of the *prcd* gene in the retina will be studied, with goals of understanding the basis of this disease and searching for treatments or even, eventually, cures."

The new OptiGen *prcd* mutation test accurately detects the presence or absence of the mutant *prcd* disease gene copy. It replaces earlier OptiGen DNA-marker-based tests (marker tests) that detected changes in coding sequences of genes located near to and inherited with (linked to) the *prcd* gene. OptiGen henceforth defines result status based on the mutation test as "Normal/Clear" or "Carrier" or "Affected." Designation of Patterns A(A1), B(B1), and C(C1) for previous marker tests are retired.

While the basic research of Aguirre and Acland spans decades, OptiGen's *prcd*-PRA test evolved over the last 7 years, with the first marker test offered to Portuguese Water Dogs in 1998. Initially it detected false positive alleles. Subsequently, improved versions of the marker test analyzed coding sequence changes in more tightly linked genes and greatly reduced the frequency of false alleles. This improved marker test accuracy and gave an excellent estimate of *prcd*-PRA frequency within affected breeds.

A crucial set of information for proof of the *prcd* mutation was generated by OptiGen, according to Dr. Jeanette Felix, President. Between March 1, 2004 and May 31, 2005, OptiGen had added the potential mutation test to the standard marker test for all new samples from *prcd*-affected breeds in order to validate the identity of the mutation in large populations. Analysis now shows that the results are consistent between the marker test and the potential mutation test.

The *prcd* mutation test further improved accuracy over the marker test for Labrador Retrievers and Miniature and Toy Poodles. Only these groups will receive updated test reports by mail during June. For all other breeds, there was no difference in the outcome between marker and mutation tests. Given this match, OptiGen will not retest other breeds originally tested before March 1, 2004. All test reports that gave Pattern status can be interpreted as: Pattern (A)A1 = Normal/Clear; Pattern (B)B1 = Carrier; Pattern (C)C1 = Affected.

Statistics based on the new mutation test show that the frequency of *prcd*-PRA varies substantially among breeds, ranging between 4% to 20% affecteds and 20% to 50% carriers. Dr. Acland emphasizes: "Genetic testing used in informed breeding programs clearly is essential for preventing new cases of vision loss. I expect more genetic tests will be developed as canine genome research accelerates. Breeders with experience using the *prcd*-PRA test will be eager to make use of all new genetic information."

The *prcd*-PRA mutation test can be obtained following the same procedures as before - see www.optigen.com. As of June 1, 2005, the fee for the *prcd*-PRA test is reduced 25%, down to \$195. Opportunities for discounts for litters, through online ordering and 20/20 Clinics continue.

The Morris Animal Foundation/The Seeing Eye, Inc. is a major sponsor of research by Drs. Acland and Aguirre. Grants from the NEI/NIH, the Foundation Fighting Blindness and The Van Sloan Fund and contributions from

many breed clubs, organizations and individuals are gratefully acknowledged. Many club health committees and breeders gave welcome encouragement as well as invaluable samples and information on their breed lines.

OptiGen, LLC, is a private company established to provide canine genetic tests to breeders and owners of purebred dogs, their veterinarians and other veterinary specialists. OptiGen holds an exclusive international license to the Cornell University technology for *prcd*-PRA mutation testing, and exclusive and non-exclusive licenses to multiple other technologies developed at Cornell and elsewhere.

www.optigen.com

genetest@optigen.com