

# DNA Testing and Genetic Disease

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## DNA TESTING & GENETIC DISEASE



CANINE GENETIC TESTING OFFERS A NEW PROMISE

Ongoing advances in canine medicine, particularly in the area of mutated gene identification, mean that some genetic diseases can now be reduced or eliminated as a result of a powerful tool known as DNA testing. These tests allow for the tagging of dogs that are carriers for certain genetic medical conditions.

Conscientious breeders, the health foundations that breed clubs have established, and researchers have all labored to realize the progress that is currently being made.

### UNDERSTAND THE ROLE OF DNA

A basic understanding of the gene and the role played by DNA (deoxyribonucleic acid) is necessary in order to grasp the challenge and the key to success in the elimination of gene-related disorders. The information or "the plan" that enables cells to make the specialized proteins that control the growth and division of cells (and for determining the difference between, for example, retinal (of the eye) cells and kidney cells is stored in the genes. Genes are made of a complex molecule, DNA, which is crimped into each cell, resulting in a structure known as a chromosome. The stored plans enable the cell to make the proteins that work together to cause unique behavior and physical characteristics in each dog.



### GENE DYSFUNCTION

Genes may sometimes undergo change, and the information contained in the gene becomes altered. Some changes, or mutations, are of little or no consequence, but others result in dysfunction. For example, a mutated retinal cell lacks a protein

critical to sight and leads to progressive retinal atrophy (PRA) and loss of vision in affected dogs. Once a gene has experienced a mutation, the damage is irreversible, and the animal will pass on the mutant gene to at least some of its offspring. Mutations can be either dominant or recessive. A dominant mutation always results in the disorder. An animal with a recessive gene, however, may appear normal but is, nevertheless, a carrier. If that animal breeds with another carrier, the disease appears in some of the offspring. That is the making of an inherited disease.

## GENETIC TESTING

If an abnormal gene can be detected in an animal, carriers of the inherited disease can be identified. This is called genetic testing. Breakthroughs in genetic testing include the work partially funded by the Collie Health Foundation in conjunction with Cornell University. After twenty years of research, they identified the gene and mutation that causes PRA in collies. A DNA test is available to distinguish between affected dogs, carriers, and those that are genetically clear. In fact, DNA testing for PRA and other genetic diseases is available for increasing numbers of breeds. The outlook for the individual breeds of dogs, breeders, and dog owners is positive, promising, and exciting as increasing numbers of genetic mysteries become unlocked by science, research, and technology.