Researchers at Cornell University have reported the finding of the genetic mutation that causes progressive retinal atrophy (PRA) in the English Mastiff. They also have discovered the gene mutation on the X chromosome that causes PRA in Siberian Huskies and Samoyeds. Previously they discovered the genetic defect causing PRA in Portuguese Water Dogs, English Cocker Spaniels, and Chesapeake Bay Retrievers.

PRA in dogs involves the loss of photoreceptors in the eye which ultimately causes blindness. The condition in dogs mimics retinitis pigmentosa (RP) in humans. At least 100,000 people in the United States currently suffer vision loss and blindness from the disease. The genetic mutation found in Huskies and Samoyeds, in particular, is very similar to the most common form of X-linked RP.

One result of these discoveries is the ability to design genetic screening tests to help dog breeders eliminate the defective gene from their lines of purebred dogs, and ensure health and soundness of the breed. In addition, knowing the specific gene mutation will make it possible to explore the possibility of gene therapy in both human patients and dogs.