BENGAL PROGRESIVE RETINAL ATROPHY

The Veterinary Genetics Laboratory at the University of California – Davis and the Lyons Feline Genetics and Comparative Medicine Laboratory at the University of Missouri announce the release of the **new genetic test for progressive retinal atrophy (PRA)**, which causes an autosomal recessive blindness in Bengal cats. The disease causes the destruction of the cells that register light (photoreceptors) in the back of the eye (the retina). The loss of the cells begins around 7 weeks of age and slowly progresses until the cat has very compromised vision by approximately 2 years of age[1]. However, blindness develops at different rates in different cats. We have examples of cats over 2 years of age chasing a laser pointer; however vision testing by an ophthalmologist indicated that the cats should be blind. Blind cats tend to have more difficulty at night, sometimes becoming more vocal and more attached to their owners. The pupils are usually more dilated for affected cats than for cats with normal vision in the same lighting conditions. Affected cats also tend to carry their whiskers in a more forward position. Once affected cats know their surroundings, they are very mobile and active. Our thanks to the breeders who came forward and helped us establish a colony so that we could define the condition and find the gene responsible for this defect.

The mutant DNA variant appears to be novel to the Bengal breed and occurred early in a popular lineage of the Bengals. We expect Bengal cats worldwide to have the condition and we have had reports of affected cats in the United Kingdom, Europe and the USA. Bengal PRA is autosomal recessive, thus two copies of the mutant DNA variant are required for the cats to be blind. The blindness can be detected either by the DNA test or by an eye exam prior breeding age. Carriers, cats with one copy of the mutation, can only be detected by the DNA test.

Because the mutation is recent in evolutionary time, an extended DNA region and several genes around the casual mutation had to be examined as many DNA variants appeared to be perfectly linked with the blindness. This is a common and well known phenomenon, called linkage disequilibrium. Other examples of linkage disequilibrium have been published for cat DNA variants that cause *Brown* coloration[2] and cat AB blood group.[3]

Several DNA variants within the region needed to be evaluated and after working with European breeders, many non-causal variants were eliminated. Initially, a gene mutation in a known vision gene was implicated, but after screening hundreds of cats, a second mutation in a different and novel vision gene was identified as being perfectly

linked with the disease. Only by continued cooperation and participation of breeders was this discovery possible!

To the best of our ability and knowledge, the new test is correct and perfectly linked with the disease. However, we are limited by the cat samples obtained for research and we will continue studies to demonstrate the function of this gene in cats. We will be publishing this research in a scientific journal, however, we are releasing the new test ahead of the publication because breeders are continuing to use carrier cats and even blind cats in their breeding programs, and thus Bengal PRA blindness is rapidly spreading.

All cats previously tested for Bengal PRA by the VGL have been retested for the new mutation and new reports will be issued to clients.

1: Ofri R, Reilly CM, Maggs DJ, Fitzgerald PG, Shilo-Benjamini Y, Good KL, Grahn RA, Splawski DD, Lyons LA. Characterization of an Early-Onset, Autosomal Recessive, Progressive Retinal Degeneration in Bengal Cats. Invest Ophthalmol Vis Sci. 2015 Aug;56(9):5299-308. doi: 10.1167/iovs.15-16585. PubMed PMID: 26258614; PubMed Central PMCID: PMC4539567.

2: Lyons LA, Foe IT, Rah HC, Grahn RA. Chocolate coated cats: TYRP1 mutations for brown color in domestic cats. Mamm Genome. 2005 May;16(5):356-66. PubMed PMID: 16104383.

3: Bighignoli B, Niini T, Grahn RA, Pedersen NC, Millon LV, Polli M, Longeri M, Lyons LA. Cytidine monophospho-N-acetylneuraminic acid hydroxylase (CMAH) mutations associated with the domestic cat AB blood group. BMC Genet. 2007 Jun 6;8:27. PubMed PMID: 17553163; PubMed Central PMCID: PMC1913925.