Background

Sudden acquired retinal degeneration syndrome (SARDS) is a common cause of incurable blindness in dogs. This disease is diagnosed in dogs with a rapid and complete loss of vision combined with a normal appearing ocular fundus and a flatline electroretinogram (ERG; Figure 1). In some cases, affected dogs also exhibit systemic signs like polyuria, polydipsia, polyphagia, lethargy, or weight gain. The cause of this disease is unknown, although an association with endocrinopathies or an autoimmune pathogenesis has been suggested.1

One criticism of SARDS research to date is the lack of a clear definition for the disease.1 A detailed phenotypic description will help to prevent misdiagnosis of SARDS and similar diseases, and will improve the quality of future SARDS research. In addition, discovery of genetic factors predisposing to development of SARDS in Dachshunds (one of the most commonly affected breeds)2 could contribute to a deeper understanding of the disease’s etiology, identification of at-risk animals, and potentially the development of treatments.

Aim 1: Genotypic Analysis

Because certain breeds are predisposed to SARDS,2 it is likely that there is a genetic component of the disease. We hypothesize that we will be able to identify regions of the canine genome that are associated with the presence of SARDS in a genome-wide association study (GWAS) using a new Affymetrix® canine single nucleotide polymorphism (SNP) array. This array is capable of identifying nucleotide variations in over a million different locations within the canine genome. The genes or regions of DNA containing these variations could be involved in the disease process, and would be excellent candidates for future study (Figure 2).

Aim 2: Phenotypic Analysis

Lack of consistent phenotypic characterization of SARDS makes it difficult to accurately identify cases and compare data among studies. To better characterize SARDS in vivo, we are using optical coherence tomography (OCT) to compare retinal layer thickness among 6 dogs affected with SARDS for < 1 year, 6 dogs affected with SARDS for > 1 year, and 12 healthy control dogs old enough to have demonstrated SARDS (Figure 3). Based on previous work,1 we hypothesized that SARDS-affected dogs will have decreased retinal photoreceptor layer thickness < 1 year after diagnosis, and decreased overall retinal thickness > 1 year following diagnosis. Fundic photography will be used to record visible changes in dogs with SARDS (Figure 4). To date, 8 dogs have undergone fundic photography and OCT imaging.

Thirty-six of the planned 48 samples have been sent out for GWAS analysis. Of those sent out three did not produce results, most likely due to poor DNA quantity or purity. Array data for the 33 samples that were successful are currently being analyzed. Blood samples have been collected from 12 additional dogs, and will be analyzed once the array is commercially available.

Selected References


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