



## RESEARCH PROGRESS REPORT SUMMARY

**Grant 02403-MOU:** Microphthalmia and Delayed Growth Syndrome in the Portuguese Water Dog

**Principal Investigator:** Margret Casal, DVM, PhD

**Research Institution:** University of Pennsylvania

**Grant Amount:** \$12,960.00

**Start Date:** 11/1/2017      **End Date:** 10/31/2019

**Progress Report:** End-Year 1

**Report Due:** 10/31/2018      **Report Received:** 11/26/2018

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### **Original Project Description:**

Microphthalmia and delayed growth syndrome (aka "puppy eye syndrome") has been reported by Portuguese Water Dog breeders dating as far back as 1986. However, there is no information in the scientific literature and the majority of data concerning this syndrome has been obtained from records of breeders, which have anecdotal reports of the disease and little, if any, medical diagnostics. Affected dogs present with microphthalmia of varying severity, other eye abnormalities, short stature and other findings. To date, the investigators have been able to collect DNA from 24 affected dogs. Males and females can be affected, although females predominate (about 70%). Preliminary pedigree studies suggest an autosomal recessive inheritance. Human literature reports numerous syndromes associated with microphthalmia, and many genes have been identified as having a causative role. The goals of this investigation are to better characterize the clinical syndrome seen in Portuguese Water Dogs, confirm a suspected mode of inheritance, obtain additional samples for investigation into the genetic mutation, and develop a mutation based, genetic test for breeders to eliminate this syndrome from the Portuguese Water Dog breed.

Funding for the research is provided through the collaborative efforts and generosity of the Portuguese Water Dog Foundation, Inc., and the Portuguese Water Dog Club of America. The AKC Canine Health Foundation supports the funding of this effort and will oversee grant administration and scientific progress reports.

**Publications:** None at this time; one publication in progress.



**Presentations:** None at this time.

**Report to Grant Sponsor from Investigator:**

As a direct result of a recent call for more samples, we now have complete eye exams from 28 affected dogs but blood work from only 11 dogs. Most of the 28 affected puppies were stunted in growth, and blood work in the 11 affected puppies indicated anemia, low hematocrit, and decreased platelet numbers. The most common eye abnormalities included microphthalmia, persistent pupillary membranes and cataracts. The recent call for samples from producer parents and affected puppies has resulted in almost doubling the sample numbers we had to date! The current DNA samples will be submitted for a genome wide association study and we anticipate having preliminary results by January 2019. We are still accepting samples and any medical records that might be available from affected puppies, even if there is no DNA sample associated with the medical record. We are particularly interested in samples from affected puppies and producer parents. We are also very interested in obtaining samples from bitches and dogs that have been bred to known producers but have never produced any affected puppies themselves. Ideally, these (presumptive “clear”) breeding bitches or dogs would have produced over 16 normal puppies and no affected puppies when bred to known producers (carriers). These samples will help in the identification of the gene causing Puppy Eye Syndrome.