



## 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:** Mid-Year 1

**Report Due:** 4/30/2018 **Report Received:** 10/18/2018

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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.



**Presentations:** None at this time.

**Report to Grant Sponsor from Investigator:**

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 many anecdotal reports of the disease and little, if any, medical diagnostics. Affected dogs present with microphthalmia of varying severity, other eye abnormalities, hind limb weakness, voracious eating, abnormal vocalizations including chirping when eating, urinary incontinence, swallowing disorders, respiratory disease, sterility, and short stature. To date, we have been able to collect DNA from 19 affected dogs and from 4 known producers. Males and females can be affected, although we have seen more affected females (about 70%). The frequency of this syndrome in is as high as 25% in certain lines. 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 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. It would be most helpful to receive more samples.