RESEARCH PROGRESS REPORT SUMMARY

Grant 02524-E: 2018 Clinician-Scientist Fellowship - University of Pennsylvania

Principal Investigator: Margret Casal, DVM, PhD

Research Institution: University of Pennsylvania

Grant Amount: $12,000

Start Date: 1/1/2018 **End Date:** 6/30/2020

Progress Report: End-Year 2

Report Due: 12/31/2019 **Report Received:** 1/8/2020

(The content of this report is not confidential and may be used in communications with your organization.)

Publications: None at this time.

Presentations: None at this time.

Internal lab presentations and presentations to the students in the Genetics class at the University of Pennsylvania.

Over the course of this year Dr. Mariah Gentry has presented our data in internal lab meetings and to veterinary students in the genetics course (VMED 604) at the Veterinary School of the University of Pennsylvania.

Report to Grant Sponsor from Investigator:

The aim of this research project has been to investigate the inheritance of renal dysplasia in the Cairn Terrier breed and the degree to which genetics influences the severity of disease - whether it is mild, moderate, or severe. We have examined several pedigrees which, together, detail the ancestry of approximately 1500 Cairn Terriers; performed ultrasounds of the kidneys of 450 dogs; and collected DNA from about 300 dogs. The affected samples made up about 10% of all DNA samples, suggesting that there are about 42% carriers in the population. Two thirds of all affected dogs could be directly linked to a single common ancestor. However, this is not an uncommon finding in a purebred dog population. It does point out though, that until we have a DNA test available, one should aim for having an ultrasound of the kidneys performed by a board-certified veterinary imaging specialist before making breeding decisions. We have performed the first and second run of our genome wide

association study and are currently re-analyzing the data, as several of the ultrasound images had to be re-analyzed due to misclassification. The review of these images was just completed last week. We have sent DNA samples from two affected dogs for a whole genome sequence to be able to accurately pinpoint the difference between dogs affected with renal dysplasia and "clear" dogs. We expect to receive the results within the next few months.

This grant allowed Dr. Mariah Gentry to 1. become proficient in working up a genetic disorder; 2. hone her molecular skills at the bench; 3. perform a GWAS; 4. work with breeders in a productive fashion; and 5. learn how to write scientific papers.

Veterinary students have not been involved with the molecular side of the project but on average about 25% of the senior students have been involved in the ultrasounds and learning how to talk to breeders. This has made a huge impact on the way students perceive breeders.