Progressive retinal atrophy and the role of RPGRIP1 DNA testing in English Springer Spaniels

Call for participation in a genetic study

Background
A mutation in the gene RPGRIP1 has previously been associated with a blinding retinal disease (progressive retinal atrophy, PRA, cord1, or crd4) in multiple dog breeds. This mutation has been found to be relatively common in the English Springer Spaniels (ESS), in contrast to the relatively low incidence of clinically affected ESS. The study aims to clarify the relation between the RPGRIP1 mutation and retinal conditions in ESS. We suspect that multiple genetic factors are involved, including RPGRIP1, and depending on the state of these genetic ‘modifiers’, the significance of the RPGRIP1 mutation in retinal health may be limited. Our goal is to tease out the individual factors and establish a reliable set of DNA tests that allows accurate prediction of the course of disease in ESS.

We are recruiting English Springer Spaniel dogs

1. Diagnosed or suspected with retinal degeneration (e.g. PRA)
2. DNA tested as ‘Affected’ for the RPGRIP1 mutation
3. Related to any ESS meeting one of the above criteria

We are also recruiting any ESS that can participate in a routine eye examination at the University of Pennsylvania Ryan Veterinary Hospital (please contact us below). Additional screening eye examination opportunities nationwide will be announced. There are retinal changes that may not lead to noticeable changes in vision, and we encourage taking advantage of the opportunity to participate in the eye exam sessions.

To participate
We ask an eye examination record by a veterinarian (preferably by a veterinary ophthalmologist), blood sample for DNA analysis and a copy of pedigree if available.
Please see the attached Study Participation Form of visit or laboratory website (www.vet.upenn.edu/miyadera-lab) for details.

Contact us
Dr. Keiko Miyadera, DVM PhD DACVO
Assistant Professor of Ophthalmology
School of Veterinary Medicine, University of Pennsylvania
Email: kmiya@upenn.edu
Tel: 215-573-4319

This study is supported by the English Springer Spaniel Field Trial Association Foundation through the American Kennel Club Canine Health Foundation.
English Springer Spaniel PRA (cord1, crd4) Research DNA Submission Form

1. Owner Information
Name: __________________________________________
Address: _________________________________________
E-mail: ____________________________ Daytime Phone: ____________________________

2. Veterinarian Information
Name: ________________________________________ (circle if applicable) DACVO / other: ______________________
Clinic Name: __________________________________________
E-mail: ____________________________ Clinic Phone: ____________________________

3. Dog Information
Call Name: _____________________ Registration #: ____________________ (circle) AKC / other: ______________________
Registered Name: __________________________________________
Birthdate (mm/dd/yy): _____________________ Sex (circle each): Male / Female and Neutered / Intact
Sire’s Reg. Name _____________________ Sire’s Reg. #: __________________
Dam’s Reg. Name _____________________ Dam’s Reg. #: __________________

4. Sample Information
Date of Blood Collection (mm/dd/yy): ____________________
Clinical description and/or drawing of phenotype or reason for submission (please include copy of ophthalmic examination or medical records describing the ocular findings):
________________________________________________________________________
________________________________________________________________________
________________________________________________________________________
________________________________________________________________________
________________________________________________________________________
Relative Known to Be Affected (Give Relationship) ________________________________________
Any Other Notes ________________________________________________________________
________________________________________________________________________
________________________________________________________________________

5. Authorization
To the best of my knowledge, the information I have supplied is accurate. I understand that the sample I have submitted will be used for further research, particularly in the effort to understand retinal conditions in English Springer Spaniels as well as other inherited diseases in dogs.

Owner’s Signature: ____________________________________________ Date (mm/dd/yy): ____________________
6. Blood Sample Collection (performed by a veterinarian or nurse):

**Sample required: 2ml x 2 EDTA tubes**

1) Label two EDTA (purple top) tube with owner’s last name, animal’s name, and date of collection.

2) Draw a 4 ml blood sample. (Do not draw more than 10 ml/kg bodyweight. It is safe to draw 4 ml from a 1 lb/454 g dog.)

3) The blood sample can be kept in the fridge until shipping within 1 week.

4) Please prepare
   - Signed submission form
   - Ophthalmic exam or medical record describing ocular findings
   - Copy of dog’s registration & 3-5 generation pedigree
   - Blood samples (2ml x 2 EDTA tubes)

5) Mail samples in by mail or courier (free shipping when using the FedEx account below) by **2-day delivery** or regular service if ice packs are included. Please place tubes in a Ziplock™ bag, then in bubble wrap for protection. Your veterinarian may have special Styrofoam containers or cardboard mailers to send blood tubes.
   * We recommend mailing samples earlier in the week (Mon-Wed) for timely receipt in the lab and to prevent samples sitting in the mail over the weekend.

7. **Ship Sample(s) to:**

   **Dr. Keiko Miyadera**
   **ESS DNA Submission**
   3900 Delancey Street
   School of Veterinary Medicine, University of Pennsylvania
   Ryan-Rm 2024, Philadelphia, PA 19104 USA

8. **FedEx Account for Free Shipping**
   FedEx account #201915872 (Please put "LEG#5-60888" in the internal reference section)

9. Questions?
   Please contact
   Email: kmiya@upenn.edu (Dr. Keiko Miyadera)
   Phone: 215-573-4319 (lab)

Thank you for your participation! Feel free to duplicate and distribute this form and instructions to others interested in enrolling for the PRA genetics study in the English Springer Spaniels at the University of Pennsylvania.

_Last updated 5-8-2019_