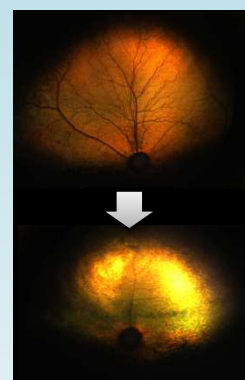


# Progressive retinal atrophy and the role of *RPGRIP1* DNA testing in English Springer Spaniels

## 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.



Blinding retinal thinning in a dog affected with PRA

### 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](http://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](mailto: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 Genetics 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 (include copy of ophthalmic examination (current & historical) or medical records describing the ocular findings):

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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. (It is safe to draw 4 ml from a 1 lb/454 g dog.)
- 3) The blood sample can be kept in the fridge up to 1 week until shipping.
- 4) Please prepare
  - Signed** submission form
  - Ophthalmic exam or medical record describing ocular findings (current & any historical)
  - 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#5-74718** in the internal reference section)

## 9. Questions?

Please contact      Email: [kmiya@upenn.edu](mailto: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.