

## **Request for Information and Samples from Cavalier King Charles Spaniels suffering from Mitral Valve Disease and their Family Members**

### ***DISEASE DESCRIPTION***

Degenerative valve disease is an adult onset disease resulting from myxomatous degeneration of the mitral (most often) valve and chordae tendinae. These structural or compositional changes in the mitral valve reduce valve strength and lead to valve leakage (and the clinical sign of a cardiac murmur). Ultimately, significant valve leakage may lead to congestive heart failure. Cavalier King Charles spaniels as a breed are at increased risk of developing this disease and tend to develop a more rapidly progressive form at a younger age than other predisposed breeds. Therefore, it is suspected to be inherited.

### ***STUDY DESCRIPTION***

The goal of this study is to find the altered version of the gene or genes that is responsible for the development of Mitral Valve Disease (MVD) in the Cavalier King Charles Spaniels. Knowledge of this variation within the gene will allow for testing of all the CKCS as early as a few weeks old to identify potentially affected animals, carriers and normal dogs. Hence, this will allow for controlled breeding and eventually the elimination of the disease from the Cavalier King Charles Spaniel breed.

In order to achieve this goal, **we need your help** to gather **clinical information, pedigrees and blood or tissue samples** from MVD affected dogs, their siblings, parents and if possible grandparents (normal or affected). The more samples we get with accurate information the faster we will be able to find the gene. All information will be handled confidentially. **In particular, we need dogs that have been examined by a cardiologist (and a copy of the examination form) with a grade 3/6 or louder murmur noted at 5 years of age or younger. We also need samples from dogs which were cleared of a murmur or clear on echocardiography at 9 years of age or older.** Older dogs which currently have a murmur but were cardiac clear at 9 years of age can be enrolled as long as a copy of the clearance at 9 years of age (or greater) is available. If possible, we would also like a copy of the pedigree, but it is not necessary for enrollment.

Please keep in mind this is NOT a test, we will not be able to provide you with any results. The identification of the gene variation, the verification of it as the actual cause of the disease and the development of a test might take several years

If you are interested in making a donation to this research project, please send a check written to "Trustees of the University of Pennsylvania" with "Cavalier heart fund" in the memo to the same address. Donations are tax deductible.

This research is possible through the generosity of an anonymous donor and the ACKCSC Charitable Trust.

**THANK YOU VERY MUCH FOR YOUR HELP**

Dr. Paula Henthorn

Meg Sleeper, VMD

# Mitral Valve Disease Study in Cavalier King Charles Spaniels

## Contact Information

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Owner First name: \_\_\_\_\_ Last name: \_\_\_\_\_  
 Veterinarian Address: \_\_\_\_\_  
 Owner's agent City: \_\_\_\_\_ State/Province: \_\_\_\_\_ Zip: \_\_\_\_\_ Country: \_\_\_\_\_  
Home phone: \_\_\_\_\_ Business phone: \_\_\_\_\_ Fax: \_\_\_\_\_  
Email: \_\_\_\_\_

## Owner Information (if different from above)

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First name: \_\_\_\_\_ Last name: \_\_\_\_\_  
Address: \_\_\_\_\_  
City: \_\_\_\_\_ State: \_\_\_\_\_ Zip: \_\_\_\_\_ Country: \_\_\_\_\_  
Phone: \_\_\_\_\_ Fax: \_\_\_\_\_ E-Mail: \_\_\_\_\_

## Animal Information

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Official name: \_\_\_\_\_ Call name: \_\_\_\_\_  
Date of birth: \_\_\_\_\_ (mm/dd/yy) Registration #: \_\_\_\_\_  CKC  Other: \_\_\_\_\_  
Sex:  Male  Female  Neutered  Intact  
Sire's name \_\_\_\_\_ Sire's registration #: \_\_\_\_\_  
Dam's name \_\_\_\_\_ Dam' registration #: \_\_\_\_\_

## Sample Information

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Date of sample collection: \_\_\_\_\_ (mm/dd/yy) (see instructions below)  
Sample type:  EDTA blood  
Pedigree included:  Yes  No If not, please give reason why or source to obtain it: \_\_\_\_\_  
\_\_\_\_\_

Clinical Information:  affected  Relative known to be affected/carrier  
 Other: \_\_\_\_\_

If affected, please include clinical diagnosis, time of onset, name and address of veterinarian that made the diagnosis

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University of Pennsylvania School of Veterinary Medicine



### **Instructions for Sample Submission**

Please follow these instructions to obtain a DNA sample and remember to provide all the necessary information when you send your animal's sample. You need to send either a blood sample. **We must have a blood sample because the DNA quality is superior.** Because of the new Cardiac snip-chips being used for DNA extraction, we cannot accept cheek swabs for analysis. If you have further questions contact us VHUP by calling (215) 898- 8894 or ACKCSC coordinator 610-459-2098.

#### **Blood Sample Collection** (performed by a veterinary clinician or nurse)

1. Label EDTA (purple top) tube with owner's last name and animal's name (or AKC#)
2. Take a 2-5 ml blood sample.
3. A blood sample must be kept cold but not frozen.
4. Complete required submission form and mail with sample.
5. Mail tube in mailer by 2-day delivery or regular service keeping it as cool as possible (using a cold pak). Your veterinarian may have special styrofoam containers or cardboard mailers and can use a Ziplock™ bag in bubble wrap for protection.

Your participation and your animal's results will be kept *strictly confidential!*

**This research is possible through donations by an anonymous donor and the ACKCSC Charitable Trust.**

#### **Contact and Shipping Information**

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CKCS

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