

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.

THANK YOU VERY MUCH FOR YOUR HELP

Mitral Valve Disease Study in Cavalier King Charles Spaniels

Contact Information

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)

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

Animal Information

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

Date of sample collection: _____ (mm/dd/yy) (see instructions below)
 Sample type: EDTA blood Three cheek swabs
 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

Please attach all important information:

- Pedigree
- Clinical diagnosis, if applicable
- Samples (Please label samples with the animal's call name and owner's last name).

All information will be kept strictly confidential.

Ship to:
 Dr. Paula Henthorn
 CKCS
 Veterinary Hospital – Room 4033
 University of Pennsylvania
 3900 Delancey Str
 Philadelphia, PA 19104-6010
 Email: cmichel@vet.upenn.edu
 Phone: 215-898-8894
 Fax: 215-573-2162



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 or cheek swabs but not both. **We would prefer a blood sample because the DNA quality is superior.** To receive swab kits, please email cmichel@vet.upenn.edu and include information regarding number of animals to be tested, name and address. If you have further questions contact us via this email or call (215) 898- 8894.

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.

Cheek Swab Sample Collection

1. To avoid contamination with food, do not feed the animal at least three hours before you collect the sample.
2. Wash your hands before you collect the sample to prevent contamination.
3. Label the packages that contain the cheek swab brushes with the owner's last name and the animal's name and/or AKC number.
4. Open the end of the swab package that shows the word, "peel," printed on it. Be careful not to touch the brush end as you remove the swab.
5. You may also wish to ask a second person to gently restrain the animal's head while you collect the DNA sample.
6. Insert brush end between the animal's gums and inside of the cheek. Roll the brush on the surface of the inside of the cheek for 15-20 seconds. Make sure that the brush is in contact with the cheek and not just with the saliva.
7. Return brush to its original package, allow it to dry, and then tape the opened end shut.
8. Repeat steps 3-5 for the second brush using opposite side of mouth. Two to three swab brushes are needed for each animal tested.
9. If you plan to test another animal, remember to wash your hands before you start with the next dog.
10. Secure brushes from each animal tested in a separate Ziplock™ bag.
11. The samples can be mailed by regular or express mail to address below.

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

Contact and Shipping Information

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CKCS

Rm 4033

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