

OFFICIAL USE ONLY

This protocol has been approved by the MJR-VHUP Privately Owned Animal Protocol Committee and the University of Pennsylvania Institutional Animal Care and Use Committee. **POAP #269.**

**UNIVERSITY OF PENNSYLVANIA SCHOOL OF VETERINARY MEDICINE
OWNER INFORMED CONSENT FORM**

Study Title: "Genetic Basis of Cystinuria"
Principal Investigators: Drs. Paula S. Henthorn and Urs Giger
 Section of Medical Genetics
 University of Pennsylvania School of Veterinary Medicine
 3900 Delancey St., Philadelphia PA, 19104-6010
 Phone: 215-898-8894, FAX: 215-573-2162, <http://vet.upenn.edu/PennGen>

You are being invited to have your pet participate in a research study. Your participation is voluntary which means you can choose whether or not you want to participate. Before giving your consent to your pet's participation, please read the following, ask as many questions as needed to understand what your participation involves, and sign and date the statement at the end of this document.

What is the purpose of this research study? The purpose of the study is to discover the genetic basis of cystinuria. Understanding the genetic basis of this disease can lead to the development of genetic tests that could help eliminate the disease and to new treatments for this sometimes life-threatening disease. Cystinuria is an inherited disease that can cause the formation of a particular kind of urinary tract stone (cystine stones) that can block the urinary track and cause other health problems.

What am I being asked to do? You are being asked to provide blood or cells from the lining of the cheek, as well as urine, from your dog. You will consent to the investigators use of the blood, cheek brush or swab, and urine samples, and will provide a pedigree and requested medical information concerning your dog. The investigators may ask you to provide additional samples from your dog at some future date. You will be asked to provide clinical and laboratory information as well as contact information for the breeder and veterinarian of your dog.

What are the possible risks or discomforts to my pet? The study requires that up to 10 ml of blood (less than one tablespoon, but no more than 5 ml/kg body weight) be obtained from your pet. You will choose the veterinarian who will be performing this procedure, and will not hold the University of Pennsylvania responsible for any complications associated with drawing the blood. DNA will be isolated from the blood. The risk involved in drawing blood is minimal. However, your dog may experience mild redness or bruising at the collection site. Additionally, in some cases and with your permission, the hair may be clipped so that the vein can be seen more easily. Free catch urine samples, and in some cases, swabs/brushes for DNA isolation, will also be collected. You can perform the cheek swab/brush and urine collection, neither of which is associated with any risks to the animal. If you need help with the cheek swab procedure or are not comfortable handling your dog's mouth, please discontinue the procedure and contact your veterinarian.

What are the possible benefits of the study for my pet? Your pet may or may not benefit directly from this study. The information gained from your participation may provide you and your veterinarian with additional information and a better understanding of cystinuria and, ultimately, this may influence the course of treatment or genetic testing to help your dog and other animals in the future. In addition, the urine you provide will be tested for excess cystine using the nitroprusside test. The results of the nitroprusside test will be sent to you at no charge if you contributed a blood sample. A positive nitroprusside test indicates that your pet may be at increased risk for forming cystine stones. While the purpose of this study is to develop new DNA tests for cystinuria, you will not be provided with the DNA test results for cystinuria nor for any other genetic diseases.

Will I have to pay for anything? You will be responsible for any blood drawing or other fees that your veterinarian charges, and for shipping costs. However, there is no fee for participating in this study.

Who can see or use my pet's information? How will my personal information be protected? We will do our best to make sure that the personal information in your pet's medical record will be kept private. However, we cannot guarantee total privacy. Your personal information may be given out if required by law. If information from this study is published or presented at scientific meetings, you and your pet's name and any other identifiable

information will not be used. The cystine/nitroprusside test results will only be reported to the submitting person or his/her agent.

Who can I call with questions, complaints or if I'm concerned about this research? If you have questions, concerns or complaints regarding your pet's participation in this research study, you should speak first with the Principal Investigators listed on page one of this form. If a member of the research team cannot be reached or you want to talk to someone other than those working on the study, you may contact the Chair of the Privately Owned Animal Protocol (POAP) committee at the University of Pennsylvania by calling 215-898-5448 and leaving a message for Dr. Lili Duda.

When you sign this form, you are agreeing to have your pet take part in this research study. This means that you have read the consent form, your questions have been answered, and you have decided to volunteer your pet. Your signature also means that you are permitting the Matthew J. Ryan Veterinary Hospital of the University of Pennsylvania (MJR-VHUP) to use your pet's health information and DNA collected for research purposes within our institution. You are also allowing MJR-VHUP to disclose that information to outside organizations or academic personnel involved with the operations of this study. Your signature also confirms that you are over 18 years of age and the legal owner or authorized agent of this pet.

In the event that DNA from your pet is used in the development of commercially available diagnostic markers or medical or surgical treatments, you understand and agree that any proceeds or benefits from such development are the sole and exclusive property of the University of Pennsylvania. You also understand that the University of Pennsylvania will not cover any charges that may be incurred for sample collection and shipment.

After signing, please make a copy of this consent form for your own records & enclose the original with sample shipment.

Owner/Agent (Please Print) _____ Signature of owner/agent _____ Date _____

Pet's Registered Name: _____

Call Name: _____ AKC#: _____

Chip #: _____ Date of Birth: _____ Sex (circle): **M / F** Intact / Neutered at _____ (age)

Breed: _____ Coat Color: _____

Sire's Name: _____ AKC #: _____

Dam's Name: _____ AKC #: _____

Owner's Name: _____

Address: _____

Phone (day): _____ (evening) _____

Fax: _____ E-mail: _____

Dog's Breeder's Name: _____

Address: _____

Phone (day): _____ (evening) _____

Fax: _____ E-mail: _____

Date and time of Urine Collection: _____ Current medications: _____

Hours between dog's last meal and urine collection: _____ Diet being fed to dog: _____

- Has your dog ever had any of the following:
- Yes No **Blood in the urine**
 - Yes No **Urinary tract infection**
 - Yes No **Urinary tract stone, if so please provide specific information and copy of stone analysis report**
 - Yes No **Straining while urinating**
 - Yes No **History of cystinuria in your dog or its relatives, if so please provide specific information (such as evidence of cystinuria in your dog or relative, relationship of affected relative to your dog, and pedigree)**

Please submit signed form, pedigree, and samples according to the following instructions:

SAMPLE SUBMISSION-Instructions for Veterinarians and Owners/Shipping Instructions

Genetic Basis of Cystinuria Study at the University of Pennsylvania

Thank-you for participating in this study, which requires a DNA sample (in the form of blood) from your pet/patient. (Under special circumstances, cheek brushes or swabs may be sufficient. In this case, the investigators will provide you with the brushes and instructions.) Please be aware that participation in the study does not imply that this dog is affected with cystinuria, nor does it imply that the dog is at risk of producing offspring affected with this disease. In order to investigate the genetic basis of the disease, it is necessary to obtain DNA and urine samples from unaffected as well as cystinuric dogs/cats.

The blood and urine supplied by you will be used to understand the causes of cystinuria in animals, and to develop genetic approaches to detect the carrier animals. This will allow breeders to make breeding choices that can eliminate this disease from their breed.

Blood Sample: Please provide 5 -10 cc of whole blood in purple-topped (EDTA) blood tubes if the dog weighs 2.5 lbs or more. If the animal weighs less than 2.5 lb (1 kg), please supply 5 cc/kg body weight. Tubes should be rocked gently to distribute anticoagulant, but should NOT be centrifuged. If sample will be shipped within 24 hours, just refrigerate until shipped. If sample must be held for greater than 24 hours, please freeze the sample after placing the blood tubes in sealable plastic bags (in case the glass tubes break during freezing).

Urine Specimen: Collect urine as a free catch while the dog is urinating. A new, clean disposable pie plate is a convenient collection vessel. Urine can then be transferred to a standard tube, available from a veterinarian's or physician's office. Please send between 5-10 cc of urine. Repeat urine specimens may be requested of dogs younger than 18 months of age, for follow-up of initial results, or after diet or other changes. If sample must be held for greater than 24 hours, please freeze the sample after placing the urine tubes in sealable plastic bags (in case the tubes break during freezing).

Label Sample and Include Appropriate Forms: Please label each sample with the dog's call name and the owner's last name and date. Include the completed and signed **Owner Informed Consent Form** and the dogs' **Pedigree**.

Shipping: Ship by overnight delivery if urine is included. If the shipment only contains blood, 2-day service is sufficient. Place samples in a small, insulated container, and include one or more frozen cold packs. Do not send samples after Wednesday. (Please note that samples shipped through the US Postal Service are guaranteed only to reach the University in one day, and do not always reach our laboratory in one day. Thus we prefer other services such as FedEx, UPS, DHL. If urine samples do not arrive cold, we will ask you to send another sample.) Samples cannot be delivered over the weekend. Ship to:

Dr. P. Henthorn/Cystinuria
Section of Medical Genetics, 4030 VHUP
University of Pennsylvania School of Veterinary Medicine
3900 Delancey St.
Philadelphia, PA 19104-6010
Phone no. (required by FedEx): 215-573-7545