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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 #373.**

**MATTHEW J. RYAN VETERINARY HOSPITAL
UNIVERSITY OF PENNSYLVANIA
MEDICAL GENETICS INFORMED CONSENT FORM**

Protocol Title:	Genetic Basis of Cystinuria
Principal Investigators:	Dr. Paula S. Henthorn & Dr. Urs Giger Section of Medical Genetics University of Pennsylvania School of Veterinary Medicine VOX: 215-898-8894 FAX: 215-573-2162 E-mail: CystinuriaDNAtest@gmail.com 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 you can make your decision, you will need to know what the study is about, the possible risks and benefits of your pet being in this study, and what you will have to do in this study. The research team is going to talk to you about the research study, and they will give you this consent form to read. You may also decide to discuss it with your family or regular veterinarian. You may find some of the medical language difficult to understand. Please ask the research team about this form. If you decide to participate, you will be asked to sign this form.

What is the purpose of this research study?

Cystinuria is an inherited disease that can cause the formation of a particular kind of urinary tract stone (cystine stone) that can block the urinary tract, which can be life-threatening. Other symptoms include urinary tract infections, blood in the urine, and straining while urinating. The purpose of this study is to discover the genetic basis of this disease, and use this information to develop genetic markers, which in turn can be used to help eliminate the disease. In order to investigate the genetic basis of disease, it is necessary to obtain samples from dogs with cystinuria, dogs without cystinuria, and relatives of both affected and unaffected dogs. Participation in this study does not imply that your dog is affected with cystinuria or at risk of producing offspring affected with cystinuria.

What am I being asked to do?

If you decide to have your pet participate in this study, we request the following:

1. Your veterinarian obtains a blood sample from a vein in your pet's leg or neck. The hair over the vein may be shaved to help see the vein better. A small needle will be inserted into the vein, and up to 10 mls (the equivalent of 2 teaspoons) will be removed, based on your pet's weight.
2. You obtain 5 to 10 mls (the equivalent of 1 to 2 teaspoons) of urine collected as a "free catch" into a clean container while your dog is urinating. Repeat urine specimens may be requested if your dog is younger than 18 months, if there have been diet or other husbandry changes, or as a follow-up on the initial results.
3. In some cases, cheek swabs or brushes may be requested in lieu of, or in addition to, blood and urine samples. In these cases, the study investigators can provide the swabs/brushes and instructions on how to collect a sample. You can perform the cheek swab yourself if you are

comfortable handling your dog's mouth. Otherwise, you can have this procedure performed by your veterinarian.

4. Label and ship samples according to the attached "Sample Submission" instructions.
5. Complete a brief questionnaire about your dog's urinary health.
6. Provide your dog's pedigree information.
7. Provide contact information for the breeders of your dog and owners of dogs that are related to your dog.
8. Provide documentation of a confirmed diagnosis of cystinuria, if applicable.

What are the possible risks or discomforts to my pet?

Risks of obtaining a blood sample include some discomfort, mild redness or bruising, itchiness, and very rarely infection at the collection site. Please discourage your pet from licking at the area where the blood sample was obtained for the next day.

Free catch urine collection and cheek swabs/brushes are not associated with any risk to your pet. If you are not comfortable obtaining these samples, you should contact your veterinarian.

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.

What are the possible benefits of the study for my pet?

Your pet is not expected to get any direct benefit from being in this research study. However, such participation may provide veterinarians with additional information and a better understanding of cystinuria. This may influence future treatment and genetic testing which may help other dogs in the future.

Will I be paid for my pet being in this study?

There is no compensation for participating in this study. In the event that DNA from your dog is used in the development of commercially available diagnostic markers or medical/surgical treatments, any proceeds or benefits from such development are the sole and exclusive property of the University of Pennsylvania. Your dog will have access to any publically available tests or procedures.

Will I have to pay for anything?

You are responsible for the costs of having your pet's blood drawn, and collection of urine and/or cheek swabs/brushes if applicable. In the very unlikely event that your pet is injured during collection of the samples, you are responsible for all costs of any associated treatments. You are also responsible for the cost of shipping the samples.

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 with the Principal Investigator 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 Matthew J. Ryan Veterinary Hospital of 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 collected for research purposes within our institution. You are also allowing MJR-VHUP to disclose that information to outside organizations or people 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. You should retain a copy of this consent form.

Signatures - REQUIRED

Client/Owner/Agent's Signature: _____ *Today's Date:* _____

Client/Owner/Agent's Printed Name: _____

Owner Information

Owner's Name: _____

Address: _____

_____ *E-mail:* _____

Phone: (day) _____ (evening/cell) _____ *Fax:* _____

Dog/Sample Information

Pet's Registered Name: _____

Call Name: _____ *AKC#:* _____ *Chip #:* _____

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

Breed: _____ *Coat Color:* _____

Sire's Name: _____ *AKC #:* _____

Dam's Name: _____ *AKC #:* _____

Has your dog ever had any of the following: Yes No *Urinary tract infection*
 Yes No *Blood in the urine* Yes No *Straining while urinating*
 Yes No *Urinary tract stone (please provide specific information and copy of stone analysis report).*
 Yes No *Is your dog related to any dog that has been diagnosed with cystinuria? If yes, state the relationship to your dog and provide pedigrees showing the relationship.*

Date and time of Urine Collection: _____ *Hours between dog's last meal and urine collection:* _____

Current medications: _____ *Diet being fed to dog:* _____

Breeder Information

Breeder's Name: _____

Address: _____

Phone: _____ *E-mail:* _____

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. If you have further questions, please feel free to contact us via email at Henthorn@vet.upenn.edu or call at 215-898-9601.

Cheek swabs/brushes: Under special circumstances, cheek brushes/swabs may be sufficient. In these cases, the investigators will provide brushes/swabs and instructions.

Blood Sample Collection:

- Please obtain 5 -10 cc of whole blood in purple-topped (EDTA) blood tubes if the dog weighs 1.0 kg (2.2 lb) or more. If the dog weighs less than 1.0 kg (2.2 lb), please obtain 5 ml/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 for some reason the sample must be held for greater than 4 days, please freeze the sample after placing the blood tubes in sealable plastic bags (in case the glass tubes break during freezing).
- Please provide adequate packaging to protect the tubes. Your veterinarian may have special styrofoam containers or cardboard mailers and you can use a Ziplock™ bag in bubble wrap for protection.

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**, and any documentation **showing a confirmed diagnosis of cystinuria**, if appropriate.

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. Please ship to:

Dr. P. Henthorn/Cystinuria
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 University of Pennsylvania School of Veterinary Medicine
 3900 Delancey St.
 Philadelphia, PA 19104-6010
 Phone no. (required by FedEx): 215-573-5703