

# *Myotonia Congenita* Test NOW AVAILABLE

## What is *Myotonia Congenita*?

*Myotonia congenita* is an inherited muscular disease found in Miniature Schnauzers. It is characterized by the delay of skeletal muscle relaxation following the cessation of a voluntary activity. The delay in skeletal muscle relaxation is not accompanied by pain or cramping.

Affected animals have significant excessive growth of the muscles (muscle hypertrophy). They also have a stiff, stilted gait that improves with exercise. Difficulty in swallowing, excessive salivation and an abnormal bark are often present. All affected Miniature Schnauzers examined so far at the University of Pennsylvania also have overshot upper jaws (maxilla) and abnormal dentition, but that is not typical of myotonia in other breeds of dogs and in other species.

## The Research at the University of Pennsylvania

Dr. Charles Vite, Dr. Urs Giger, and several of their colleagues at the University of Pennsylvania and Vanderbilt University, have done extensive work on the disorder. They have made remarkable progress in a very short time. They have developed a treatment, identified a mutated gene responsible for causing the disorder, and developed a DNA test to detect the mutated gene.

## Causation and Inheritance

Dr. Vite reports that the researchers documented that the disorder is inherited as an autosomal recessive trait. A single mutation in the gene encoding the chloride channel for skeletal muscles appears to be responsible for the clinical signs seen. Affected dogs are homozygous for this mutation, *i.e.*, they inherited the mutated gene from both parents. Carrier dogs are heterozygous for the mutation, *i.e.*, they inherited the mutated gene from one parent and a normal one from the other parent. The research on mode of inheritance is based on five litters examined at the University of Pennsylvania and

two additional litters reported to the researchers which they were unable to examine.

## Incidence

**Based on what we presently know, all Miniature Schnauzers are at risk of carrying the *myotonia congenita* gene.** Last summer, we reported that, so far as we were aware, all affected animals were descended from Ch. Blythewood Shooting Sparks, but that he had not himself directly produced any affecteds. That statement is no longer correct. We have recently learned of a myotonic litter that does *not* have Ch. Blythewood Shooting Sparks in the pedigree. (We mentioned Shooting Sparks with the kind permission of Joan Huber. In accordance with long-standing AMSC policy, we do not name any dogs or owners who have not expressly given us permission to do so.)

We do not know how many litters there have been to date with affected puppies. We have heard estimates of 20 to 30 litters.

## The DNA Test

Dr. Urs Giger, head of the Josephine Deubler Genetic Disease Testing Laboratory of the University of Pennsylvania School of Veterinary Medicine, has advised us that the DNA test for *myotonia congenita* in Miniature Schnauzers is now available. He and Dr. Vite have advised us that the test is definitive because a specific defect has been identified in the DNA sequence of affected and carrier animals. The lab will be accepting blood samples beginning immediately. The cost for testing is \$75 U.S. per sample. Master Card and Visa credit cards are accepted.

## Instructions For Submitting Samples

1. The blood must be drawn by your veterinarian. Your veterinarian will need to draw 1 to 2 milliliters of blood and store it in a EDTA tube (lavender colored top).

2. Complete and enclose a copy of the attached Submission Form for each sample. Be sure the name on the tube matches the name on the Submission Form. **Please label each tube with your name and address and the name of the animal whose blood is contained in the tube.** The tube(s) must be inserted in a small safe container to prevent breakage and shipped to Penn. (Most vets have styrofoam tube containers.)

Penn recommends Second Day delivery - Express Mail, \$2.95. Express mail is cheaper, but if you want assurance that your samples were received, send them by FedEx, UPS, or some other method of shipping that allows tracking through the shipper. **Do not call the Deubler Laboratory to see if samples have been received.**

3. **If you are testing litters, please label each tube carefully with a name or code for each animal that you can recognize.** Keep in mind, though, that the certificate of results will be show the name or identification you provide. Thus, if you test animals who have not yet been registered, you may need to test them again if you later need a certificate showing the registered name (e.g., if you offer the animal at stud and someone asks for proof that he has been tested as clear).

If you have questions about submitting samples, please call or email Vera Potiker, [Kilvercrest@aol.com](mailto:Kilvercrest@aol.com); (909) 780-6284, rather than bothering the lab personnel.

## Who Should Have Their Dogs Tested?

There is no way of predicting, without testing, which animals are carriers; nor can we even say that animals from cer-

tain bloodlines are less likely to be carriers than those from other bloodlines.

The Health Committee recommends testing all Miniature Schnauzers that you intend to breed. (Each animal need be tested only once, not every time it is bred.) Dr. Giger also recommends testing any animals with suspicious clinical signs in order to confirm a clinical diagnosis of myotonia congenita. It is not necessary to test animals that are healthy and will not be bred again or at all.

All offspring arising from the mating of animals that have both tested clear should be clear. Nonetheless, the Deubler Laboratory recommends testing every breeding animal - even if both its parents tested clear - until there has been more real-world experience with the DNA test.

So far, the incidence of *myotonia congenita* has been fairly low. Let's act before it becomes more prevalent and much harder to eliminate from the breed. The \$75 cost will be some of the cheapest insurance you can find to avoid future heartbreak.

The AMSC cannot require that members test - but individual members can decline to buy or breed to untested animals. The Health Committee recommends that you request a copy of the test certificate for any stud dog you are planning to use, and that you request a copy of the test certificates for any animal you are planning to buy for show or breeding purposes.

The Health Committee also suggests you caution prospective puppy buyers to ask about test certificates of the parents. If enough puppy buyers become knowledgeable and ask questions, maybe they can encourage the puppy mills and backyard breeders to get their stock tested too.

### Breeding Considerations

Based on the present understanding of the mode of inheritance:

1. Breeding an affected to an affected will produce affected offspring who are all affected.
2. Breeding a carrier to an affected or to another carrier yields an unacceptably high risk of producing affected offspring.
3. Breeding an affected or a carrier to a clear does not risk producing affected offspring, but does carry significant risk of spreading the mutated gene to further generations. All offspring that are to be used for breeding would have to be tested to be sure that they do not carry the mutated gene.

The only question of practical importance is whether carriers should be bred to clears. If a carrier has desirable traits that the breeder wishes to preserve in future generations - and which are not available from a clear animal - the carrier should be bred only to a known clear animal. All offspring considered for breeding should be then tested as some of them will likely be carriers.

Penn will **not** release test results for specific animals to anyone other than the owners or submitting veterinarians; but Dr. Giger has advised us that they will provide statistical results on the frequency of carriers and affecteds (once a sufficient number of animals have been tested) to the AMSC. We will report on those results as they become available, and may make additional recommendations as warranted based on the advice of the experts.

### Contributions Requested For the Deubler Laboratory

Dr. Giger has asked the AMSC and/or Miniature Schnauzer breeders to contribute funds to the Deubler Laboratory to defray at least some of the expenses it has incurred in finding the mutated gene that causes *myotonia congenita* and in developing the DNA test to identify it. Dr. Giger told us that the Deubler Laboratory incurred extra expense to complete the work on the test as quickly as possible because the AMSC urged them to do so.

More importantly, the Deubler Laboratory will not be making a profit on the DNA tests as they are only charging \$75 per test.

Dr. Giger's request is more than reasonable. The University of Pennsylvania did all of the work with no financial help from Miniature Schnauzer breeders. Most of the time when we fund research, we are asked to do so in the hope that something useful will result. Here, Penn has actually delivered the goods before asking us to pay anything. Also, Penn is pricing the test at considerably less than the \$250 to \$400 a test several of the other University-affiliated labs, such as Cornell's and Michigan's, have charged for similar DNA tests for other disorders.

In order to make the contributions tax-deductible for those paying U.S. income tax, please make your checks payable to the **'University of Pennsylvania'**. Please mark the memo section of the check **"Deubler Laboratory - Miniature Schnauzers"** and please mail the checks to **Jerry Kyser, 310 Brookridge Drive, Camden, AR 71701-3106**, so that we can keep track of the donations and make sure that Miniature Schnauzer breeders get credit for the contributions. All donations will be acknowledged in Amscope. Penn will provide the receipts.

### Information on the AMSC Web Site

To facilitate access to this information, we are also putting the foregoing information and the Submission Form on the AMSC web site. Feel free to refer non-AMSC members who may be interested in testing their stock to the AMSC site, <http://clubs.akc.org/amsc/index.html>. You may also print additional Submission Forms from the web site.

**MYOTONIA CONGENITA SUBMISSION FORM**

**Please print legibly**

Owner Name \_\_\_\_\_  
 Breeder Street \_\_\_\_\_  
 Veterinarian City \_\_\_\_\_ State \_\_\_\_\_ Zip \_\_\_\_\_  
Phone \_\_\_\_\_ Fax \_\_\_\_\_  
Email \_\_\_\_\_

Date of sample collection \_\_\_\_\_ Date of Birth \_\_\_\_\_  
Animal's Registered Name \_\_\_\_\_

Breed: **Miniature Schnauzer**

AKC Reg. # \_\_\_\_\_ or Foreign Reg. # & Country \_\_\_\_\_  
Sex:  Female  Male  
Neutered:  Yes  No  
Sire \_\_\_\_\_ Reg.#\* \_\_\_\_\_  
Dam \_\_\_\_\_ Reg.#\* \_\_\_\_\_

\* Indicate Country if not AKC

Reason for Testing (select all that apply)

General Genetic Screening  Showing  
 Suspicious Clinical Signs  Breeding  
 Relative known to be affected (please state who) \_\_\_\_\_  
 Other (explain) \_\_\_\_\_

Is Ch. Blythewood Shooting Sparks in the animal's pedigree? Yes \_\_\_ No \_\_\_

Test to be conducted: **Myotonia Congenita screening**

**\$75 U.S.**, payable by check, or Visa or Master Card credit card

Visa \_\_\_ MasterCard \_\_\_ Card No. \_\_\_\_\_ Exp. Date \_\_\_/\_\_\_

Print name as it appears on card: \_\_\_\_\_ Signature \_\_\_\_\_

Please send the blood sample, this Submission Form, and check (unless paying by credit card) to **"Trustees, Univ. of Penns./Giger"** by two-day delivery mail to:

Dr. Urs Giger/Myotonia Congenita  
Veterinary Hospital Room 4006  
University of Pennsylvania  
3850 Spruce Street  
Philadelphia, PA 19104-6010

Phone: (215) 898-3375

Fax: (215) 573-2162

Email: [penngen@vet.upenn.edu](mailto:penngen@vet.upenn.edu)

Questions? Please contact Vera Potiker, [Kilvercrest@aol.com](mailto:Kilvercrest@aol.com); (909) 780-6284, rather than calling lab personnel.