



Genetic Testing: a screening tool for dilated cardiomyopathy (DCM) in doberman pinschers

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Dilated cardiomyopathy (DCM) is a genetic condition in doberman pinschers typically manifested in young adulthood. Although, younger patients can be affected and sometimes clinical signs don't present until older years. One of the challenges with this condition is that it can be silent with no overt signs on clinical presentation or physical exam (murmur or arrhythmia) until quite advanced (heart failure or sudden death). Therefore, screening tests are paramount in helping identify and manage this condition in a proactive way.

Genetic testing is now available for dilated cardiomyopathy (DCM) in doberman pinschers and is a good first line screening directive. But, as with the advent of all tests come questions. Here are guidelines to some frequently asked questions.

1) What genetic test is available? There are actually two. At least 2 different genetic mutations have now been identified associated with dilated cardiomyopathy in doberman pinschers, thanks to research lead by Dr. Kate Meurs, DACVIM (cardiology): NC State University Veterinary Hospital. PDK4 (NCSU DCM1) was the first to be identified with NCSU DCM2 following in its path. Each mutation has its own test. Unfortunately, dogs can carry both mutations. Therefore, it is recommended to test for both.

1) PDK4 (DCM1) affects the mitochondrial energy protein resulting in a bad "energy system". 37% of dobermans that have this genetic mutation will become ill from their disease (according to Meurs).

2) DCM2 affects the sarcomeric protein that leads to poor systolic function and sudden death. 50% of dogs with this mutation will develop disease (according to Meurs).

2) What do the test results tell us? Genetic testing for this condition in dogs is probability based and predictive in nature, rather than absolute, for manifestation of the condition. So how do we interpret the results of the test?

- A negative result for either test does not necessarily exclude the possibility that the dog may develop DCM as there are most likely more mutations responsible for manifestations of DCM than have been currently identified.
- Dogs can test positive for one or both of the mutations. A positive result for either, or both tests, does not necessarily mean that the patient will develop clinical manifestations of the condition. However, according to Dr. Meurs, it is the dogs that test positive for both mutations that are at greatest risk of becoming ill from their condition.

3) Who should we test? It is recommended to screen all dobermans with the genetic tests in an attempt to better predict which patients may be more susceptible to developing dilated cardiomyopathy.

4) How do we use this information to develop an ongoing cardiac care plan?

- Recommend genetic testing for all your doberman pinscher patients.
- If positive, additional screening tests starting at 3 years of age, to include an echocardiogram and Holter, are recommended on an annual basis.
- If they test negative, inform your clients that a negative test does not rule out the possibility of DCM as other genetic mutations that have not yet been characterized are possible. Annual screening is still advised starting at 3 years of age. Thoracic radiographs can screen for cardiomegaly and physical examination can screen for ventricular arrhythmias. But, this line of testing has its limitations and dogs with DCM can be missed.
- Echocardiography and Holter monitoring remain the gold standard for diagnosing and characterizing dilated cardiomyopathy in dobermans and should be presented as options for the client, even in the face of negative genetic testing.

5) Where is the genetic testing performed? What sample do I need?

Doberman Pinscher Dilated Cardiomyopathy (DCM) genetic testing is being conducted at the Veterinary Cardiac Genetics Laboratory at NC State University College of Veterinary Medicine. Sample submission includes either a cheek swab(s) or blood test (EDTA tube with 1-3 mls of blood) shipped at room

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temperature. Instructions along with the submission form and mailing address can be found on the NC State University website <https://cvm.ncsu.edu/genetics/doberman-pinscher-dilated-cardiomyopathy/>. Again, the recommendation is to test for both of the genetic mutations.

***Special considerations for OFA Heart Certification.** If the intent is to have the genetic test be a part of the OFA Advanced Cardiac Database Certification, the sample must be obtained and submitted by the veterinary cardiologist signing off on the form. The same holds true for Holter monitoring.

For more information on this topic, Dr. Kate Meurs of the Veterinary Cardiac Genetic Lab at North Carolina State University presents free online webinars. To view, visit the following links:

- 1) 2014 Doberman DCM Update – Webinar: <https://my mediasite.online.ncsu.edu/online/Play/fb1bfb27416f452d8e8174ef18300e741d>
- 2) 2016 Doberman DCM Update & DCM2 Gene – Webinar: <https://my mediasite.online.ncsu.edu/online/Play/d84281e7f40643bd84d96f0755f0cb9b1d>