VETERINARY CARDIOLOGY SPECIALISTS

Genetic Testing: utility in veterinary medicine for cats

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Genetic testing is available for hypertrophic cardiomyopathy (HCM) in ragdoll and maine coon cats. The question is how can we incorporate genetic testing into practice and how can the results influence the cardiac care plan for our patients, including breeding recommendations?

Hypertrophic Cardiomyopathy (HCM) genetic testing

HCM has been associated with mutations of the cardiac myosin-binding protein C (MYBPC3) in ragdoll and maine coon cats. Two separate genetic tests are available (one specific for the ragdoll and one specific for the maine coon mutation). This line of testing is not valid in other breeds at this time. However, current studies are underway in sphynx cats, lead by Dr. Kate Meurs from North Carolina State. For more information on this study click here: https://cvm.ncsu.edu/genetics/sphynx-cat-hypertrophic-cardiomyopathy-study/

Genetic testing in ragdolls and maine coon cats

- The test is probability based and predictive for the manifestation of HCM
- The result will either be negative, homozygous or heterozygous

What is the predictive value of the test?

- A negative test does not mean that they won't develop the condition as there is more than one genetic mutation that can result in HCM
- If heterozygous: there is an ~ 30% chance that the cat will develop HCM
- if homozygous:
    - Ragdolls will most likely manifest the disease by 2 years of age
    - Maine Coons: 60% will develop the condition by < 4 years of age and almost all > 4 - 6 years of age will be affected

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How do we use this information to develop a cardiac care plan?

- Recommend testing for all your ragdoll and maine coon patients
- If they test negative, inform your clients that a negative test does not rule out the possibility of HCM, but it is possible that they may never be affected. Monitor NT-proBNP levels annually, before anesthesia and if concerns with respiratory distress. NT-proBNP is a cardiac biomarker elevated with cardiac stretch or stress. If NT-proBNP levels are elevated recommend echocardiogram +/- thoracic radiographs.
- If heterozygous or homozygous positive, recommend baseline echocardiogram and NT-proBNP level. If HCM is identified, the cardiologist will work with you to create a specific cardiac care plan for the individual patient. If HCM is not phenotypically present, NT-proBNP levels can be monitored every 6 months, before anesthesia and if concerns with respiratory distress develop. Echocardiography is the gold standard for diagnosing and characterizing HCM in cats. Therefore, performing echocardiograms if the NT-proBNP level is elevated as well as on an annual basis for routine screening is appropriate.

How can ragdoll and maine coon breeders benefit from genetic testing for HCM?

The genetic test for HCM in ragdolls and maine coon cats can assist breeders in their breeding programs. It is not recommended to breed a homozygous positive cat. Breedings between 2 heterozygous cats are expected to produce 25% high risk (homozygous), 50% moderate risk (heterozygous) and 25% negative kittens. Breeding a heterozygous to a negative cat will statistically reduce risk with expected results of 50% heterozygous and 50% negative kittens. Heterozygous cats are still at an ~ 30% risk of developing HCM. An echocardiogram is the gold standard for diagnosing HCM and is recommended prior to breeding.

Where is the genetic testing performed?

Cheek swabs are submitted to UC Davis: Veterinary Genetics Lab. For more information click on the following links:

-Ragdoll HCM: https://www.vgl.ucdavis.edu/services/cat/HCM.php
-Maine Coon HCM: https://www.vgl.ucdavis.edu/services/cat/MaineCoonHCM.php