



Hypertrophic Cardiomyopathy (HCM)

The Animal DNA Laboratory is pleased to announce that as of June 2008, we will be offering DNA testing for specific mutations in the MYBPC3 gene that have been associated with HCM in Maine Coon and Ragdoll cats.

Hypertrophic Cardiomyopathy (HCM) is the most common form of heart disease in cats. Onset of clinical symptoms usually occurs in middle age however, cats as young as one have been described. Cats with severe HCM and heart failure usually only live for a few months.

The inherited form of HCM is autosomal dominant, which means that carriers or heterozygotes (ie, those having one copy of the gene) will be affected. Although all cats with the mutation will be affected, the age of clinical onset and severity can vary considerably.

It is important to note that there are many forms and causes of HCM. HCM leads to a thickening of the heart wall, particularly on the left side. This in turn, can lead to heart failure, embolism and death.

Two specific mutations at different sites on the MYBPC3 gene have been found to be associated with HCM in Maine Coon and Ragdoll cats, respectively. The MYBPC3 gene is partly responsible for the heart's ability to contract. These specific mutations result in the production of faulty proteins. The heart tries to compensate by producing more protein. This overproduction can result in thickening or hypertrophy of the heart.

The two tests that are available are able to be performed on a simple cheek swab sample. They identify specific mutations as described by Drs Meurs and Kittle-son (Published in Genomics 2007,90,261 & Human Molecular Genetics 2005, 14, 3587).

A Positive Result ?

A positive result is reported as HCM Positive Heterozygous or as HCM Positive Homozygous.

A positive test result means that the cat will develop HCM, although at what age and how severe is unknown. Some report that males may get the disease at an earlier age and may have a more severe form.

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**STOP
PRESS!
HCM TEST
IS NOW
AVAILABLE**

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A Negative (Clear) Result ?

A “clear” result means that the cat does not have the specific mutation that has been found to be associated with HCM in the breeds mentioned. As the test does not detect other causes of HCM, a “clear” result does not mean the cat will never develop HCM.

Breeding outcomes.

The table below summaries the probability of affected offspring by various matings.

Percentage of Offspring expected to be affected

WE CAN HCM
DNA TEST
MAINE COONS
AND RAGDOLLS
USING A SIMPLE
CHEEK SWAB

	Unaffected Clear	Positive Heterozygous	Positive Homozygous
Unaffected Clear	0%	50%	100%
Positive Heterozygous	50%	75%	100%
Positive Homozygous	100%	100%	100%

The HCM NA test for Maine Coons and Ragdolls will be available from June 2008.

Simply request a sample collection pack and submission form and mark either HCM-Maine Coon or HCM-Ragdoll.

Test submission forms can also be downloaded from the bottom of the ‘fee schedule’ page of our website.

DNA Question Corner—win free tests

Animal DNA Laboratory gives all readers the opportunity to ask questions relating to animal genetics. The best question each issue will win 2 free DNA tests and be published in the ADL newsletter. So if you have a question simply send it to:

dnaquestions@animalsdna.com

This quarter’s winner is Jenny Taaffe from Brunswick who asked. **“I have a breeding queen who is positive with PKD. She currently shows no signs or symptoms of the disease. After testing her last litter of five, results came back telling us that all kittens were positive for PKD. I know the sire is negative. How can this be?”**

Dear Jenny,

There are two issues here. Firstly, is the cat you presume to be the sire really the true biological sire. There are many reports demonstrating that some breeding registries contain as high as 10-15% errors that have occurred because of the assumption of a sire

in a breeding program. Only careful isolated breeding programs, or DNA parentage testing can overcome these errors. A number of animal breeding registries are now instigating some form of DNA testing or auditing to prove parentage before registration occurs.

The second point is that breeding a PKD positive cat to a PKD negative cat should, by probability, result in half the litter being positive and half being negative. However, this is not so with your litter. The word “probability” is important here. Probability here means “on average, over time.” Who is to say that your next litter of five may all be negative, bringing the breeding outcome back to half (5 out of a total of 10) being positive.

Below shows a simple table that explains the breeding outcomes of mating a positive PKD cat (one positive PKD gene and one negative PKD gene) to a negative PKD cat (two negative genes). As can be seen in the example, of the four kittens born (in orange boxes) by probability, half will have inherited the Dams positive gene and hence will be PKD positive.

		Dams Genes	
		PKD Positive	PKD Negative
Sires Genes	PKD Negative	Positive Kitten	Negative Kitten
	PKD Negative	Positive Kitten	Negative Kitten

Enquire today about how your club or association can receive 10% discount off all tests!

Economical Animal DNA Testing

Animal DNA Laboratory

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Animal DNA Laboratory offers a 10% discount to the members of Clubs and Associations that have registered with us.

Clubs, Associations and Catteries registered with Animal DNA Laboratory.

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[Asian Group Cat Society](#)

[Aurorapetz](#)

[Australian Cat Federation](#)

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[Bombay Breed Club](#)

[Basic Organization of Cat Breeders Trutnov](#)

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[Cat Authority of Victoria](#)

[Cat Owners Association of Western Australia](#)

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[New Zealand Cat Fancy Inc](#)

[Rasclub Maine Coon](#)

[Richmaure Persians](#)

[Riverina Cat Club](#)

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[Shorhair Cat Society](#)

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[Sophisticats](#)

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continuously
adding new tests
so feel free to
keep up to date
with our
progress;

www.animalsdna.com