

Feline Hypertrophic Cardiomyopathy: Advice for Breeders

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What is hypertrophic cardiomyopathy?

Hypertrophic cardiomyopathy (HCM) is the most common heart disease of cats, whether they are random bred or pedigreed. It is a heart muscle disease in which the papillary muscles (the muscles in the left ventricle that anchor the mitral valve) and the walls of the left ventricle become abnormally thickened. HCM is often a progressive disease, and a proportion of affected cats develop heart failure if the muscle hypertrophy and subsequent scarring of the heart muscle significantly affects heart function. Cats with the disease may die suddenly and may develop a blood clot in the chamber above the left ventricle (i.e., the left atrium) that often then gets carried into the systemic arterial system, most commonly lodging in the terminal aorta, stopping blood flow to the rear legs.

For more information on HCM, see:

<http://members.aol.com/jchinitz/hcm/index.htm>

What causes HCM in cats?

This is currently unknown in most cats although familial (hereditary) HCM has been observed in several breeds, such as the Maine Coon and American Shorthair. Anecdotal information suggests there is familial HCM in many other breeds. Heart muscle hypertrophy in cats can be caused by other diseases, such as systemic hypertension (high blood pressure) and hyperthyroidism. HCM is a primary disease of the heart muscle. Hypertension and hyperthyroidism cause secondary thickening of the left ventricle and so are not causes of HCM (although it is possible that they may exacerbate the disease if

they become present in a cat with mild to moderate HCM). HCM is diagnosed when these other causes are ruled out.

Is HCM genetic?

In Maine Coons and American Shorthairs, HCM has been confirmed as an autosomal dominant inherited trait, as it is in humans where over 200 gene mutations in 10 genes have been found to cause the disease. The disease has variable expression; meaning some cats are severely affected, others are only mildly to moderately affected, and some cats may not have evidence of the disease yet produce affected offspring.

Recently, a mutation in the cardiac myosin binding protein C (cMyBP-C) gene causing HCM in the Maine Coon cat has been identified. Undoubtedly, other mutations responsible for HCM in cats remain to be discovered. However, since few veterinary cardiologists and geneticists have the expertise to study genes, it may be some time before the responsible gene or genes for each affected breed will be found. The mutation identified as a cause of HCM in Maine Coon cats may not be the same mutation or even on the same gene in other breeds. The genetics of HCM in each breed will require investigation of each individual breed.

Can HCM have a nutritional cause?

There is no evidence in cats, humans or other species of animals that HCM can have a nutritional cause.

How is HCM diagnosed?

HCM is diagnosed using ultrasound of the heart – an echocardiogram. Echocardiography is a good way to detect moderate to severely affected cats. However, it may not always detect mildly affected cats where changes in the heart can be minimal. Ideally, an echocardiogram to test cats for HCM should be performed by a board-certified cardiologist or radiologist.

In addition to an echocardiogram, other tests may also be useful in assessing cats with HCM. For example, a chest x-ray is necessary to detect heart failure in cats with severe HCM. An electrocardiogram is useful in cats that have abnormal heart rhythms. Blood pressure measurement and blood testing for hyperthyroidism are indicated to rule out other diseases that mimic HCM, especially mild to moderate HCM.

A genetic test is now available for the known cMyBP-C mutation causing HCM in Maine Coon cats. The test is available from the Veterinary Cardiac Genetics Lab of Dr. Kathryn Meurs at the College of Veterinary Medicine, Washington State University (<http://www.vetmed.wsu.edu/deptsvcgl/>). The test can identify which cats have the

mutation. If a cat is identified as having the mutation, the test can also determine whether the cat carries one copy of the gene (a heterozygote) or two copies of the gene (a homozygote).

Should my cats be tested for HCM and how often should they be tested?

In clinical practice, the most common patients tested for HCM with echocardiography are cats with suggestive clinical signs of heart disease, such as a heart murmur. Testing cats used in a pedigree breeding program is a more difficult endeavor. Echocardiography is not a perfect tool for diagnosis of HCM – some affected individuals will escape detection and access to good quality ultrasound services may be difficult and expensive for some breeders. At the very least, breeding cats should be auscultated (examined by a vet with a stethoscope) for heart murmurs or arrhythmias once yearly. Any cat with an abnormality should have an echocardiogram. A significant percentage of cats with HCM will not have a heart murmur, however.

Since HCM can occur at any age, a single normal echocardiogram does not guarantee a cat is free of disease. Breeding cats should probably have an echocardiogram yearly during their breeding years. Examining retired cats periodically is also advantageous as this may allow the identification of affected cats that have offspring in a breeding program.

A Maine Coon cat that tests negative for the cMyBP-C mutation is not guaranteed to be free of HCM, for it is not known if other mutations causing HCM are present in this breed. Ideally, cats that test negative for the cMyBP-C mutation should still undergo echocardiogram screening. Cats that test positive for the disease should not be bred. They will most likely develop the disease at some time during their life although it may be too mild to detect even on an echocardiogram.

At what age should a cat be tested for HCM?

HCM can affect cats at any age. It has been seen in kittens only a few months of age and in cats over the age of 10. In Maine Coons, most affected male cats have evidence of disease by 2 years of age, and most affected females have evidence of disease by 3 years of age although instances have been documented where the disease has not shown up until much later. Ragdolls with severe disease seem to develop it earlier in life, often at under 1 year of age. Guidelines for other breeds have not yet been developed. It is therefore hard to recommend a specific age to start testing. It may make sense to screen most breeding cats with an echocardiogram for the first time around the age of 2 years. Maine Coons may be tested for the cMyBP-C mutation as kittens.

What do I do if my cat is diagnosed with HCM?

The cat should be removed from the breeding program and all offspring should be watched closely for the development of HCM. Statistically, 50% of the cat's offspring would be expected to have the genetic mutation that causes HCM if one parent was a heterozygote. However, the most prudent approach may be not to use any of the offspring in a breeding program. The offspring of Maine Coon cats with the cMyBP-C mutation should be individually tested to determine their status.

The parents of an affected cat should also be examined with echocardiography (and tested for the cMyBP-C if a Maine Coon), as one of them likely carries a gene mutation for HCM. In some cases, identification of the affected parent may be difficult, especially if the disease is mild. In these cases, the most prudent approach may be to remove both parents from the breeding program. It is possible for a cat to develop a spontaneous mutation that causes HCM during embryonic development but this is an unlikely cause in a breed known to have the problem.

All breeders that are using cats related to an affected cat should be notified that a cat has been diagnosed with HCM. Similarly, pet owners should be notified that a relative has been diagnosed with the disease. Echocardiographic examination (and genetic testing if a Maine Coon) of cats related to the affected cat should be performed.

Will we ever eliminate HCM from my breed?

The tools we currently have to diagnose HCM (i.e., echocardiography and necropsy) are not perfect and will not allow us to totally eliminate this disease. However, echocardiographic screening will be able to reduce the incidence of HCM within a breed if enough breeders are involved.

The identification of the cMyBP-C mutation in the Maine Coon and the development of a genetic test provide breeders with a new tool to reduce the prevalence of or theoretically eliminate the mutation within this breed by not breeding affected cats. Breeders should use all the information they can gather about HCM in family lines, including pedigree analysis based on accurate identification of affected cats.

Any cat that dies suddenly or dies from HCM should have a necropsy (i.e., post mortem examination). Most cats with HCM will have a heart that weighs more than 20 grams and most cats with severe HCM will have a heart that weighs more than 30 grams. Myocardial fiber disarray, the hallmark microscopic heart muscle abnormality seen in humans with familial HCM is seen in all Maine Coon cats with HCM. Unfortunately, most veterinary pathologists are not trained to recognize this lesion.

In the long term, we will need a genetic test for HCM in each breed. A genetic test allows us to identify affected cats before they were bred and do so accurately. Since the disease is inherited as an autosomal dominant trait, once a mutation is identified, if all breeders

cooperated by testing their breeding cats for the mutation the disease could be eliminated from the breed within several generations. However, the money and resources necessary to identify the gene or genes and to develop a genetic test for each breed are scarce in veterinary medicine. Breeders and cat fanciers can help by supporting research through organizations such as the Ricky Fund established by the Winn Feline Foundation (<http://www.winnfelinehealth.org/>).

Can two normal parents produce a kitten with HCM?

Since HCM is known to be an autosomal dominant trait in the breeds where the inheritance is known, each affected cat must have one affected parent. However, there are possible situations in which an affected cat may come from two apparently normal parents.

The first possibility is that one of the parents has been misdiagnosed. This can happen due to inexperience of the ultrasonographer or poor quality equipment. It can also happen if a cat's status is decided on the basis of only one or two ultrasounds early in life. Since HCM can develop at any age, a cat that is normal on ultrasound one year could still have HCM and show signs later in life.

Since the trait has variable expression, not every affected cat will have echocardiographic evidence of HCM. It is therefore possible for a cat to test negative for HCM on ultrasound, and yet still carry a genetic mutation and pass it to offspring.

Finally, it is possible for spontaneous mutations to occur in cats from normal parents. These cats may then pass on their mutation to offspring. We do not know how often spontaneous mutations causing HCM occur in cats. Statistically, spontaneous mutations are more likely to occur in random bred cats than in pedigreed cats.

What does "HCM free cattery" mean?

There is no universally agreed upon definition of an HCM free cattery. The terminology is currently unclear, as different breeders mean different things when they use this term. Ideally, each breed should develop a specific definition and guidelines for use of this designation for catteries.