

An Overview of Feline Hypertrophic Cardiomyopathy

Genetic Pet Care

Hypertrophic cardiomyopathy (HCM) is an inherited disease of cats which appears to be similar to the disease seen in humans. In humans many different mutations have been found that cause HCM and it is thought that there are over 400 separate causative mutations in people.

HCM is a disease where the heart muscle becomes severely thickened and cannot function properly. This can lead to signs of congestive heart failure, thromboembolic disease ("throwing" clots) or sudden death without previous signs. HCM can sometimes be detected at your cat's regular veterinary check-up if a murmur or abnormal heart rhythm is present, but an ultrasound of the heart (echocardiograph) is required for diagnosis and to document the severity of the disease.

Mode of Inheritance: HCM is an autosomal dominant disease, which means a cat only needs one copy of the mutation to be affected. It has variable penetrance and expression, which means the age of onset of disease and the severity of disease can vary quite a lot. It does tend to be a disease seen in adult cats (the average age of onset is 8-9 years).

THE GENETIC TEST

There are two tests (mutations) available for hypertrophic cardiomyopathy. One is for the (A31P) mutation in Maine Coon cats; the other is for the (C820T) mutation of Ragdoll cats. Both mutations occur on the cardiac myosin binding protein C gene (MYBPC3), which codes for part of the contractile protein within the heart muscle cells.

Interpreting the genetic results – Maine Coon

Normal/clear/negative: Your cat does not carry the mutation and is unaffected

Affected heterozygous/affected 1 copy: Your cat carries one copy of the mutation and is affected. Your cat may or may not develop HCM and should have regular monitoring cardiac ultrasounds. Most Maine Coons with one copy do not go on to develop HCM.

Affected/positive/homozygous two copies: Your cat carries two copies of the mutation and is affected. It is more likely that your cat will develop HCM. Your cat should have regular cardiac ultrasounds.

Interpreting the genetic results – Ragdoll

Normal/clear/negative: Your cat does not carry the mutation and is unaffected.

Affected heterozygous/affected 1 copy: Your cat carries one copy of the mutation and is affected. Your cat may or may not develop HCM and should have regular monitoring cardiac ultrasounds. Most Ragdolls with one copy do not go on to develop HCM

Affected/positive /homozygous two copies: Your cat carries two copies of the mutation and is affected. It is more likely your cat will develop a severe form of the disease. Disease is likely to occur at an early age (1-2 years) in homozygous individuals.

Note that the Ragdoll is prone to an early onset form of HCM when an individual inherits two copies of the (C820T) mutation. This form of the disease tends to almost always be severe, with an age of onset between 1-2 years of age and most cats succumb by the age of 5. Ragdolls with one copy of the mutation tend to have the adult onset of the disease, similar to that seen in the Maine Coon and others.

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THE GENETIC TEST (CONT)

Also note that a Normal/Clear result does not mean a cat will not develop HCM. There is at least one other gene mutation suspected in the Maine Coon. The genetic test is only testing for one specific mutation that is causative of HCM. It is recommended all breeding animals undergo regular cardiac ultrasound examination as well.

Breeding Recommendations for Reducing the Prevalence of HCM:

1. Genetic testing should be undertaken for all breeding animals.
2. Never breed a cat that gives a result "Affected Homozygous" - all progeny will be carriers
3. Ideally, do not breed cats with an "Affected Heterozygous" result - half of progeny will be carriers. However - if a heterozygous cat is deemed by a breeder to have exceptional qualities that will be of great benefit to the breed otherwise, a cat with such a result may be bred once to a clear/unaffected cat. All of the kittens produced should be tested, and the affected kittens should not be bred. One or more clear/unaffected kittens should then be selected to continue the line.

This approach allows the HCM gene to be removed gradually from the population, taking into account the relatively small gene pool available. In this way we are not creating other problems by removing too many cats from the gene pool too quickly.

4. Always ensure that the DNA status is fully disclosed to potential owners prior to any offer of a placement of a cat or kitten, and ensure that the potential owner is fully aware of the requirements of managing a HCM cat.

5. Where there is no genetic test available, (e.g. the Maine Coon second mutation, and other purebreed cats), all breeding cats should undergo regular cardiac ultrasound to screen for signs of HCM. Breeds which have shown and documented HCM include the Sphynx, Bengal, Siberian, Scottish Fold, Norwegian Forest Cat and Persian.

This is done with the understanding that the disease is usually adult onset, so breeding may have already taken place by the time any signs are seen, and that the penetrance and expression of HCM can vary, so a cat with a mutation for HCM may not show sufficient signs of disease to be detected. These are unfortunate current limitations until further genetic mutations are characterised for different breeds.

