



THE WINN FELINE FOUNDATION

For the Health and Well-Being of All Cats

1805 Atlantic Ave., PO Box 1005, Manasquan, NJ 08736-0805
Voice 732-528-9797, ext 31 Fax 732-528-7391 www.WinnFelineHealth.org

An Update on the Genetics of Feline Hypertrophic Cardiomyopathy

Kathryn Meurs, DVM, PhD, DACVIM (Cardiology), Professor
Washington State University, College of Veterinary Medicine, Pullman WA

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Summary prepared by Janet Wolf, Tusenkatts Birmans

Dr. Kathryn Meurs, DVM, Diplomate ACVIM (Cardiology), Washington State University presented a timely and informative talk on Hypertrophic Cardiomyopathy this year. She first described the disease and its diagnosis and then discussed the genetics behind this disease. Her update on the breed-specific research she has been conducting was of great interest to many of the symposium attendees.

Hypertrophic cardiomyopathy (HCM) is a primary heart muscle disease and is the most common heart disease in cats. It usually results in a thickening of the heart muscle, generally in the left ventricle and often leads to an increase in blood pressure in the upper chambers of the heart and causes a backward flow of blood into the lungs. The symptoms cat owners notice include difficulty in breathing (open-mouth breathing) or heart failure. Sometimes blood clots result, often moving to the back legs and causing paralysis (saddle thrombosis). Typified by adult onset, prognosis and progress of this disease are variable but it is.

HCM is often inherited or susceptibility to the disease is “genetically programmed” to develop in a specific individual. It is, however, not always inherited. At this time, we believe there are inherited forms of HCM in Maine Coon, Devon Rex, Ragdoll, American Shorthair, and British Shorthair cats. To determine inheritability and the genes that are implicated, the researchers have looked at HCM in humans, where 11 different sarcomeric genes and over 200 mutations have been identified. This provides direction for the researchers looking at feline HCM. At this time, they have identified one mutation in the contractile protein in Maine Coon cats. After comparing cats with known HCM from other breeds (Ragdolls, British Shorthairs, and Norwegian Forest Cats), the researchers know that the mutation identified for Maine Coon cats is not present in these cats. They also suspect that there may be modifier genes that affect the severity, age of onset, etc. of the disease. While identifying the one mutation in Maine Coons is a significant breakthrough, it is not the complete answer, as this mutation is not observed in all affected Maine Coons or the other breeds they have looked at with a history of HCM.

Identifying the affected gene/s is a slow and laborious process. Dr. Meurs and her research team worked from 1995 to 2005 before identifying the first gene mutation in the Maine Coon cat!

Screening for HCM is an important first step. Typically, the first sign of a cat with HCM is a heart murmur or gallop discovered during a routine physical examination by a veterinarian. Murmurs, however, are not specific for HCM in cats -- 30% are of undetermined origin and some cats with murmurs never develop HCM. So, how do we screen for HCM:

- Physical exam with a stethoscope (ausculted).
- If a murmur or gallop is heard, follow-up with an echocardiogram/ultrasound of the heart.
- Note: all ultrasounds are not of the same quality and it is important to pick your ultrasonographer carefully. Look for someone with a lot of experience in this area.
- Echocardiography/ultrasound must be repeated annually as it is a disease typified by adult onset. In Maine Coons, a DNA-based test is now available for the one identified gene mutation.

The researchers looking at HCM in Maine Coon cats have determined that 33% of the submissions for testing have been positive for the genetic mutation. So, what is the next step for breeders? Removal of 33% of the gene pool over a short period of time is likely to result in the loss of good traits at the same time and will decrease the genetic diversity within the breed. Since most cases are heterozygous for the trait, Dr. Meurs recommends carefully evaluating any Maine Coon that tests positive. Assess the quality of the cat and, if it has traits that you want to perpetuate, breed it to an unaffected cat. Test all the kittens, carefully evaluate them, and try to keep the kittens that test negative or only those cats of exceptional quality that test positive. It will take several generations of breeding to work through this challenge and produce kittens that test negative and have the traits you wish to perpetuate.

What should you do if you test the cat and it is homozygous for this gene mutation? Dr. Meurs recommends that you test the cat annually by echocardiogram and/or have it evaluated annually by a feline cardiologist as it is an adult onset disease. Do not use this cat for breeding as all offspring will carry the gene mutation.

Dr. Meurs concluded by saying she believes that additional genetic mutations will be identified in both the Maine Coon cats and in other pedigreed cats. It is a long process and there are many more questions to be answered.

Note: The Winn Feline Foundation has funded many of the studies undertaken by Dr. Meurs and Dr. Mark Kittelson into HCM in felines. We are pleased to have been able to share Dr. Meurs expertise with the cat fancy and will continue to work with breeders and others to identify the causes of HCM. If you would like to support our efforts to learn more about HCM, donations can be made to the Ricky Fund or to our general funds for further research.