

## Hypertrophic Cardiomyopathy (HCM) Q & A with Dr. Kittleson

Q1. At what age in males can this disease "actually" be diagnosed? And females??

1. In Maine Coons most males have evidence of the disease by 2 years of age and females by 3 years of age but we have seen it develop as late as 7 years of age in females and have seen it develop as young as 6 months of age in kittens from mating two affected cats. No one knows in Bengals.

Q2. Is there scientific proof that this genetic disease is 100% NOT diet related??

2. There is in Maine Coons, American Shorthair, and humans where HCM is inherited as an autosomal dominant trait. There has been no evidence of a nutritional cause in any species.

Q3. Can two parents "test" negative throughout their lives and still produce some kittens that may have HCM??

3. I think so. This happens commonly in humans where an individual will have the mutation but never have evidence of the disease. In our colony I think we see 100% penetrance but in the real world I have seen and heard of cats that had the disease where both parents were "normal." This means that either one of the parents had whatever mutation but did not manifest the disease or the affected offspring had a mutation develop spontaneously in utero. The second has also been shown to happen in humans but is much less common than the first scenario.

Q4. Does the word "genetic" mean the kittens are born with it or just predisposed?? As soon as the cat reaches adult hood, should it show at that time, since the organs are fully mature??

4. Genetic means there is an abnormality in the genome. This may be the form of one base pair mutation or may take the form of multiple abnormalities. The word for a disease that is present at birth is "congenital." For HCM, for example, in Maine Coons the disease is not present at birth but develops over time and so it is not a congenital disease in the classic sense.

Q5. If a cat tests "negative" for a number of years, and then reaches say.....10-12 years of age, is there a 100% chance that it will never have HCM??

5. That's a very tough question to answer. The real answer is that I don't know. I suspect that there are a fair number of cats that have mild to moderate HCM all their life and then develop something like hyperthyroidism or high blood pressure when they are older and this exacerbates the HCM to become severe. So it's theoretically possible to have a cat with mild disease (which would be difficult to detect on an echocardiogram) that developed hyperthyroidism when it was 15 years of age and all of a sudden show up with what appears to be severe HCM. I don't think the disease shows up on its own at this sort of age but I have no way of telling that for sure.

Q6. All the studies that have been done, are they 100% set in stone or can there still be other possibilities??

6. There haven't been very many studies. Maine Coons are set in stone. I doubt there will ever be anyone else that will develop a colony to prove mode of inheritance, etc. so anything said about another breed is extrapolation. However, it is extrapolated from data that has been consistent across species and is the only known cause. However again,

even in humans around 40% of cases do not have one of the 150 or so mutations in 11 genes identified to date so there most still be something else out there that causes the disease. That something else, however, is still probably genetic.

Q7. Is this an all breed cat condition in general or is it more towards specific breeds of cats??

7. It's a strange phenomenon. HCM is very common in cats, whether they are mixed breed or purebred, yet it's a rare condition in dogs. And it seems as things progress, more and more purebreds are recognized as having the disease. However, the disease is still most commonly seen in mixed breed cats. That shouldn't be that surprising, however. If the disease is inherited as an autosomal dominant trait in all cats it would be passed on to 50% of all offspring, whether they were purebred or mixed so it would be easy to disseminate a mutation throughout a large population. Look at hypercholesterolemia in humans. It's all over the place. But the intriguing question here is why is the cat genome is so susceptible to developing mutations in the genes that are involved with HCM - or is it one mutation?

Q8. Is this condition a game of Russian roulette??

8. You're correct - echocardiographic screening is a bit of a game of Russian roulette. If not all cats with a mutation manifest the disease, then it isn't seen in all cats on an echocardiogram that can pass on the disease. That's why I always tell people that echocardiographic screening is not going to rid a breed of the disease. It should reduce the incidence (depending on how many breeders copy) and it might get rid of it in a breed or two but in most I think all we can hope for is a reduction in incidence. That's better than nothing but not as good as knowing what the mutation is and being able to screen for that mutation.

What we need in veterinary medicine is the type of labs they have in human medicine that have millions of dollars of funding and hundreds of graduate students working on the problem. In veterinary medicine we have a couple of people and a few thousand dollars. It doesn't make one optimistic. I think Bill Gates needs a purebred cat with HCM.: -)

I hope this helps.

Dr. Kittleson

Mark D. Kittleson, DVM, PhD  
Diplomate ACVIM (Cardiology)  
Professor, Dept. of Medicine and Epi.  
School of Veterinary Medicine  
UCDavis