Hypertrophic Cardiomyopathy

In brief

Hypertrophic cardiomyopathy is the most common cardiac disease in cats worldwide. It is characterized by the increase in wall thickness of the left ventricle and the intraventricular septum, eventually leading to congestive heart failure. In the Maine Coon, a point mutation (A31P) in the MYBPC3 gene has been found causative for the disease. The relative risk for developing HCM is around 1.8 in heterozygous cats and 18 in homozygous cats compared to Maine Coons negative for A31P. HCM has also been found in about 5.4% of cats negative for A31P, thus this mutation is not sole cause of HCM in Maine Coons.

Clinical overview

Hypertrophic cardiomyopathy is a cardiac disease that is characterized by increased wall thickness and mass of the left ventricle and/or the intraventricular septum. This causes turbulence in blood flow and increased venous pressure in the left atrium and lungs. The clinical signs most commonly noted with the disease are respiratory signs associated with congestive heart failure such as tachypnea (increased breathing speed), exercise intolerance and panting, difficulty breathing, and coughing. Thromboembolism may also occur and affected cats have an increased risk of sudden cardiac death.
In Maine Coon cats and the majority of other cat breeds, clinical and echocardiographic signs appear after the cat has reached breeding age. The most common age of diagnosis is 5-7 years. In Ragdoll cats however, the disease has an early onset with an average age of diagnosis being only 15 months. Both in Maine Coons and Ragdolls, cats homozygous for the breed-specific causative mutation have earlier onset of the disease and increased severity of the disease than those with only one copy.

Disease severity

Considerable

Clinical signs

- respiratory signs associated with congestive heart failure
- thromboembolism
- increased left ventricle wall thickness

Mode of Inheritance

Autosomal Dominant (Incomplete Penetrance)

Results of the genetic test are reported as follows:

Clear

At risk

Mutation(s) found in:

Maine Coon

Maine Coon Polydactyl

References

Online Database

Online Mendelian Inheritance in Animals, OMIA (http://omia.angis.org.au/). Faculty of Veterinary Science, University of Sydney.

Scientific Articles

Meurs K, Sanchez X, David R, Bowles N, Towbin J, Reiser P, Kittleson J, Munro M , Dryburgh K, MacDonald K, Kittleson M. A cardiac myosin binding protein C mutation in the Maine Coon cat with familial hypertrophic cardiomyopathy. Human Molecular Genetics, 2005, Vol. 14, No. 23 3587–3593. doi: 10.1093/hmg/ddi386.

Fries R, Heaney AM, Meurs KM. Prevalence of the Myosin-binding Protein C Mutation in Maine Coon Cats. J. Vet. Internal Med. 2008; 22:893-896.

Mary J, Chetboul V, Sampedrano CC, Abitbol M, Gouni V, Thehiou-Sechi E, Tissier R, Queney G, Pouchelon JL, Thomas A. Prevalence of the MYBPC3-A31P mutation in a large European feline population and association with hypertrophic cardiomyopathy in the Maine Coon breed. J. Vet. Cardiology 2010;12:155-161.

Wess G, Schinner C, Weber K, Kuchenhoff H, Hartmann K. Association of A31P and A74T Polymorphisms in the Myosin Binding Protein C3 Gene and Hypertrophic Cardiomyopathy in Maine Coon and Other Breed Cats. J. Vet. Internal Med. 2010;24:527-532.

Godiksen MT, Grandstrom S, Koch J, Christiansen M. Hypertrophic cardiomyopathy in young Maine Coon cats caused by the p.A31P cMyBP-C mutation – the clinical significance of having the mutation. Acta Veterinaria Scandinavica 2011;53:7-18.

Longeri M, Ferrari P, Knafelz P, Mezzelani A, Marabotti A, Milanesi L, Pertica G, Polli M, Brambilla PG, Kittleson M, Lyons LA, Porciello F. Myosin-Binding Protein C DNA Variants in Domestic Cats (A31P, A74T, R820W) and their Association with Hypertrophic Cardiomyopathy. J Vet Intern Med 2013;27:275–285.

Granström S, Godiksen MT, Christiansen M, Pipper CB, Martinussen T, Møgelvang R, Søgaard P, Willesen JL, Koch J. Genotype-phenotype correlation between the cardiac myosin binding protein C mutation A31P and hypertrophic cardiomyopathy in a cohort of Maine Coon cats: a longitudinal study. J Vet Cardiol. 2015;17 Suppl 1:S268-81. doi: 10.1016/j.jvc.2015.10.005.