Hypertrophic Cardiomyopathy found in Ragdoll

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 Ragdoll, a point mutation (R820W) in the MYBPC3 gene has been found as causative for the disease. Most cats heterozygous (1-copy) for this mutation do not develop clinical signs, whereas homozygous (2-copies) are at high risk of developing severe HCM signs early in the life and shorter lifespan.

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.

Disease severity

Considerable

Mode of Inheritance

Autosomal Dominant (Incomplete Penetrance)

Results of the genetic test are reported as follows:

Clear

At risk

Mutation(s) found in:

Ragdoll

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, Norgard M, Ederer M, Hendrix K, Kittleson M. A substitution mutation in the myosin binding protein C gene in ragdoll hypertrophic cardiomyopathy. Genomics. 2007;90(2):261-4.

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.
Borgeat K, Casamian-Sorrosal D, Helps C, Luis Fuentes V, Connolly DJ. Association of the myosin binding protein C3 mutation (MYBPC3 R820W) with cardiac death in a survey of 236 Ragdoll cats. J Vet Cardiol. 2014;16(2):73-80. doi: 10.1016/j.jvc.2014.03.005.