

Genetic testing for Hypertrophic Cardiomyopathy (HCM) in Maine Coon and Maine Coon related cats

HCM – the Disease

Hypertrophic cardiomyopathy (HCM) is a clinically heterogeneous myocardial disease and is the most common cardiac disease identified in domestic cats. HCM is characterised by an increased left ventricular mass due to an increase in wall thickness of the heart, with papillary muscle hypertrophy and systolic anterior motion of the Mitral valves. Subsequently, hypertrophy of the left heart chamber results in cardiac weakness and ultimately in heart failure. Death by HCM can occur via three mechanisms: (i) sudden cardiac death with arrhythmia and ventricular fibrillation, (ii) heart failure with tachycardia, increased respiration, shortness of breath, pulmonary oedema and pleural effusion or (iii) thrombus formation. Thrombi can form either in the left atrium due to abnormal blood circulation or in the heart chamber itself due to severe hypertrophy and cardiac weakness. Atrial thrombi can break free and reach the arterial blood circuit, thereby often causing blood congestion at the branching of pelvic and crural arteries with paralysis of the hind legs. Echocardiographic examination has so far been the only diagnostic tool for this disease. However, it can only identify affected cats with some years of age, when they already present first symptoms of HCM.

HCM – trait of inheritance and mutation

HCM is inherited as a single autosomal dominant condition. Heterozygous animals show all clinical signs of disease and can not live normal lives. They are able to propagate mutations throughout the population. Generally, 50% of a HCM positive cats' offspring will inherit HCM. Homozygously affected animals for HCM show more severe clinical symptoms and will pass the defect gene onto all of their offspring.

Recently, a mutation in the MYBPC gene which is suggested to cause HCM in cats was found by Dr. Kathryn Meurs (Washington State University, USA). This mutation was found in most HCM affected cats but not in cats which were tested free by means of echocardiographic techniques. In our laboratory, we were also able to identify this mutation in European cats with HCM.

HCM - the DNA test

By DNA testing the mutation can be shown directly. The testing is carried out by state of the art laboratory methods and therefore provides a very high accuracy. In general DNA tests can be done at any age.

The test can be applied to Maine Coon and Maine Coon related cats, which were cross bred to Maine Coons. With this test we can diagnose the reported mutation, but by no means we can report on the presence/absence of the disease (especially in breeds where the correlation of HCM disease and the cited mutation is not proven). The results that are transmitted contain the information on presence/absence of the G to C mutation in the MYBPC gene exon 3 in the sample of the cat examined. We want to point out that there is still a small possibility of other mutations causing HCM which are not identified so far.

HCM – test requirements

The test is performed out of EDTA whole blood (0.5-1 ml) or special cytobrushes. These cytobrushes are available upon request. Testing is performed a few times per week. The results are due about 1 week after arrival of the sample in our laboratory.

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