

Ragdoll Hypertrophic Cardiomyopathy (RD-HCM)

About the disease

Hypertrophic cardiomyopathy (RD-HCM) is the most common form of heart disease found in domestic cats, with affected animals at risk of developing congestive heart failure and occasionally sudden death.

HCM can be seen as a secondary complication of some other diseases, usually in older cats and most notably associated with hyperthyroidism.

HCM can also be seen in younger cats of specific breeds, including Maine Coons and Ragdolls as a familial disease.



About the test

The Molecular Diagnostic Unit can now offer genetic testing for hypertrophic cardiomyopathy in **Maine Coon** and **Ragdoll** cats. We have developed PCR-based pyrosequencing assays to quickly and accurately identify the two *MYBPC3* gene mutations currently linked with HCM in **Maine Coon (A31P)** and **Ragdoll (R820W)** cats.

These mutations cause amino acid changes within the cardiac myosin binding protein and are believed to alter the protein structure and function. It is possible to detect whether a cat has one copy of the defective gene (Heterozygous) or two copies (Homozygous).

There is evidence that the presence of the mutant gene puts cats at greater risk of developing HCM, and the risk for Homozygous cats appears to be greater than that for Heterozygous cats, but this correlation is not perfect. This may be because other, as yet unidentified, genetic mutations can also cause HCM or modify the risk.

There is some evidence that cats Homozygous for the mutant gene are more likely to develop moderate to severe disease earlier in life when compared to Heterozygous cats. Results suggest that the prevalence of the R820W mutation in the Ragdoll population is around 13%.

Reception Hours

Mon-Fri 9am - 5pm

Contact Us

T: 0117 394 0510

E: labs@langfordvets.co.uk

catgenetics@langfordvets.co.uk

langfordvets.co.uk

Langford Vets

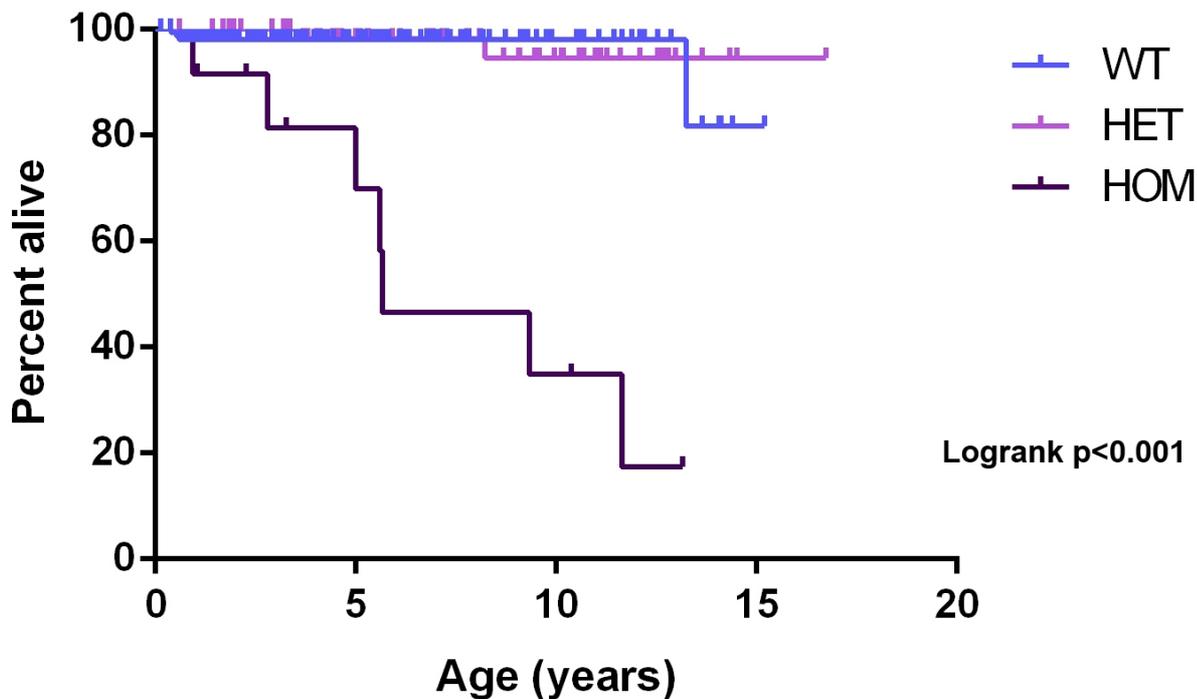


 University of
BRISTOL

Ragdoll Hypertrophic Cardiomyopathy (RD-HCM)

Survival rates for Ragdoll cats with HCM

A recent study undertaken by the Royal Veterinary College and Langford Vets investigated the survival rates of Ragdoll cats with and without the hypertrophic cardiomyopathy mutation. The graph below clearly shows that Ragdoll cats Homozygous (HOM) for the mutation have a reduced life expectancy compared to Normal (WT) or Heterozygous (HET) cats. The fact that about 20% of Homozygous cats were still alive at around 12 years of age indicates the incomplete penetrance of this mutation. Over half of the Ragdolls Homozygous for the mutation did not survive 5 years, which shows that genetic testing to identify Heterozygous cats, and hence avoid breeding Homozygous kittens, is still important for Ragdoll breeders.



Kieran Borgeat, Domingo Casamian-Sorrosal, Chris Helps, Virginia Luis Fuentes, David J. Connolly. Association of the myosin binding protein C3 mutation (MYBPC3 R820W) with cardiac death in a survey of 236 Ragdoll cats. *Journal of Veterinary Cardiology* (2014) 16, 73-80.

Reception Hours

Mon-Fri 9am - 5pm

Contact Us

T: 0117 394 0510

E: labs@langfordvets.co.uk

catgenetics@langfordvets.co.uk

langfordvets.co.uk

Langford Vets

University of
BRISTOL

Ragdoll Hypertrophic Cardiomyopathy (RD-HCM)

Interpretation of results

A **Normal** result on the Ragdoll HCM genetic test means that the cat does not have the respective genetic mutation. It is possible that some cats may go on to develop cardiac disease due to other, as yet unidentified genetic mutations.

A **Heterozygous** or **Homozygous** result on the Ragdoll HCM genetic test means that the cat carries either one (Heterozygous) or two (Homozygous) mutated copies of the MYBPC3 gene and is at greater risk of developing cardiac disease. Cats that are Heterozygous for the genetic mutation are thought likely to develop HCM.



Consideration should be given to screening cats with a positive HCM genetic test result by ultrasound to determine the disease status and progression.

Each certificate we issue will specify whether the cat is Normal, Heterozygous or Heterozygous for the Ragdoll HCM mutation.

The gene test combined with ultrasound can assist breeders in deciding whether or not to use cats for breeding. Generally Heterozygous affected cats should not be used for breeding because they are certain to pass on the genetic mutation, which will put their offspring at increased risk of developing HCM.

There is a 50% probability of a Heterozygous cat passing on the genetic mutation when bred with a HCM negative cat. In such cases the offspring should be screened for the genetic mutation and those identified as negative used for future breeding. This strategy can be used as part of a breeding programme to eliminate the defective gene from the Maine Coon and Ragdoll populations.

Reception Hours

Mon-Fri 9am - 5pm

Contact Us

T: 0117 394 0510

E: labs@langfordvets.co.uk

catgenetics@langfordvets.co.uk

langfordvets.co.uk

Langford Vets



 University of
BRISTOL

Ragdoll Hypertrophic Cardiomyopathy (RD-HCM)

FAQs

Can my cat go on the ICC negative or GCCF active registers?

Yes it can. For cats to be placed on the ICC negative register or the GCCF active register the sample (mouth swab or blood sample) MUST be taken by a vet who confirms the cat's identity using its microchip number. The microchip number must be written on the submission form AND sample.

We have a dedicated submission form for this purpose, which both the owner and vet must complete. Our result certificate will state that the cat's identity was confirmed by a vet and you can use this to register the cat on the ICC negative or GCCF active register.

If you DO NOT want your cat to go on these registers then you can take a mouth swab and submit it directly to the lab.

Can I genetically test my Bengal for HCM?

There is currently no genetic test for HCM in Bengals. The only way to diagnose HCM in Bengals is by echocardiography.

Can I genetically test my British Shorthair for HCM?

There is currently no genetic test for HCM in British Shorthairs. The only way to diagnose HCM in British Shorthairs is by echocardiography.

Reception Hours

Mon-Fri 9am - 5pm

Contact Us

T: 0117 394 0510

E: labs@langfordvets.co.uk

catgenetics@langfordvets.co.uk

langfordvets.co.uk

Langford Vets 

 University of
BRISTOL