

Mitral Valve Dysplasia (MMVD) in Dogs: A Genetic Insight

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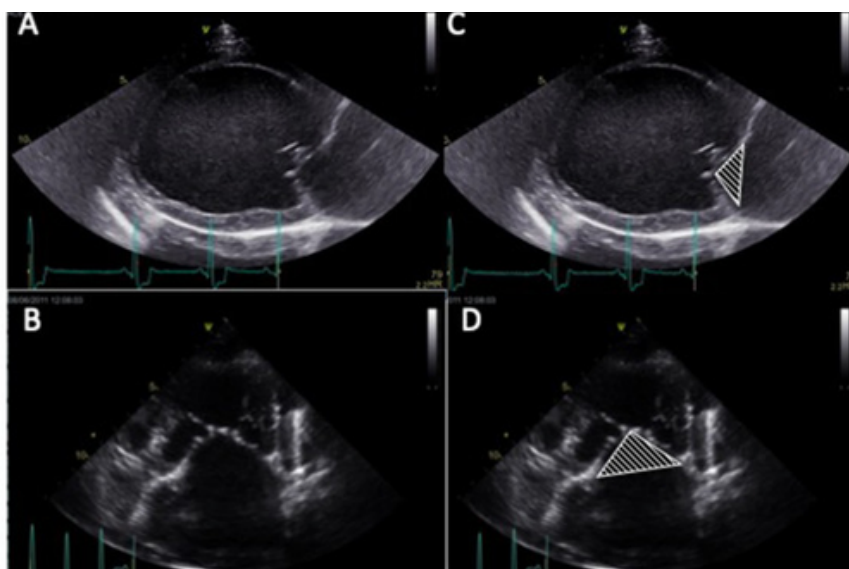


George Sofronidis
AUTHOR

Hi, I'm George Sofronidis from Orivet Genetic Pet Care! As a passionate advocate for animal health and responsible breeding, I have firsthand experience with the joys and challenges of pet ownership, thanks to my beloved pets, Charlie the Boston Terrier and Mary the Griffon. At Orivet, we are dedicated to improving the genetic health of pets, offering innovative solutions to ensure their well-being.

What is MMVD?

Myxomatous Mitral Valve Disease (MMVD) is a congenital heart condition where the mitral valve between the left atrium and left ventricle does not form properly. This can cause the valve to leak during heart contractions, leading to a heart murmur, heart enlargement, and fluid accumulation in the lungs. MMVD accounts for 75% of all cardiac issues in domestic dogs, making it the most common cardiovascular disease in these animals.



Symptoms

The symptoms of MMVD can vary widely based on the severity of the disease. Here's a breakdown:

- **Mildly Affected Dogs:** These dogs may only have a murmur and might not develop clinical symptoms for years, if at all. They might show no outward signs of heart disease and live relatively normal lives for several years.
- **Moderately Affected Dogs:** These dogs might start showing signs of exercise intolerance, coughing, and fatigue as the disease progresses. Their heart function is compromised, but they might still lead active lives with proper medical management.
- **Severely Affected Dogs:** These dogs can suffer from heart failure at a young age, with a poor prognosis. Symptoms can include severe coughing, difficulty breathing, and general lethargy. These dogs require immediate and intensive veterinary care to manage their condition.

Breeds at Risk

While MMVD can affect any breed, certain breeds are particularly predisposed to this condition. Cavalier King Charles Spaniels (CKCS), Poodles, and Dachshunds are among those with a higher risk. Notably, CKCS exhibit a high rate of early-onset MMVD, leading to more severe heart problems and a higher risk of death compared to other breeds. This breed-specific predisposition makes understanding and managing MMVD crucial for responsible breeders.

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Diagnosis & Screening

Diagnosing MMVD typically involves a combination of physical examination and imaging techniques. Here are the primary methods used:

1. **Auscultation:** A veterinarian uses a stethoscope to listen for a heart murmur, which is often the first indicator of MMVD. Murmurs are graded based on their loudness and the timing during the heart cycle.
2. **Echocardiography (Ultrasound):** This imaging technique provides a detailed look at the heart's structure and function. It can reveal abnormalities in the mitral valve and measure the size of the heart chambers.
3. **X-Ray:** Chest X-rays help document the degree of lung congestion and heart enlargement, which are common in advanced MMVD. They provide a visual confirmation of the heart's size and the presence of fluid in the lungs.
4. **Electrocardiogram (ECG):** This test records the electrical activity of the heart and can detect arrhythmias that might accompany MMVD.

Challenges in Breeding Programs

MMVD presents significant challenges for breeding programs due to its hereditary nature. Breeding affected parents increases the risk of passing the condition to offspring, perpetuating the cycle of heart disease within certain breeds. This highlights the importance of genetic testing and informed breeding decisions to reduce the prevalence of MMVD.

The Cry for Help

Cavalier breeders around the world frequently ask me about reliable genetic tests or screening methods to manage MMVD in their breeding programs. This congenital heart disease, characterized by the improper formation of the mitral valve, has been a significant concern due to its high prevalence in Cavaliers. Breeders are seeking ways to reduce the incidence of MMVD and improve the overall health of their breeding lines.



The Breakthrough

Thanks to pioneering research by Axelsson and his team, we have promising news. Their extensive whole-genome sequencing of 20 dog breeds, including the Cavalier, has led to a better understanding of the genetic underpinnings of MMVD. This research has identified several candidate risk alleles associated with the disease.

Genetic Markers and MMVD

Research has shown that specific genetic markers, particularly six locations within the Nebulette (NEBL) gene, are linked to MMVD in CKCS. In a recent study, 180 Australian CKCSs were tested for these risk markers. Among them, 178 dogs had their heart health assessed through echocardiograms, focusing on measurements like the left atrium to aortic root ratio (LA) and the left ventricular end diastolic diameter (LVIDdN).

A variant in the NEBL gene is associated with an increased risk of developing this early-onset form. Dogs homozygous for this risk factor are more likely to be diagnosed with MMVD early in life compared to heterozygous dogs. The study found that genetic tests could accurately predict the presence of risk variants in the NEBL gene. Three of these variants (NEBL1, NEBL2, and NEBL3) were always found together in the CKCS population studied.

What This Means for Breeders

The identification of these genetic markers is a game-changer, paving the way for the development of genetic tests that can identify dogs carrying these risk alleles. Such tests will enable breeders to make more informed decisions, reducing the incidence of MMVD in future generations of Cavaliers and other at-risk breeds.

Unfortunately, the test is not the golden egg breeders have been looking for, but it is a start and can be highly beneficial when used alongside traditional screening methods.

Orivet currently offers this test and screens for all five variants of the NEBL gene, with NEBL3 referred to as the "candidate" variant and the other four (NEBL1, NEBL2, NEBL4, and NEBL5) seen as risk and research variants. The NEBL3 variant is recognized as the 'causative' mutation and is the key variant to consider for any breeding decisions. A negative test result is referred to as 'normal' risk, a carrier/heterozygous result indicates an increased risk, and a positive/homozygous result indicates a significantly increased risk.



Words of Wisdom from Charlie & Mary

Research suggests that even though the NEBL risk alleles are common in CKCS, there are still some healthy alleles that can help reduce the severity of MMVD. Breeders can use this information to make better breeding decisions and improve the heart health of future generations of CKCS.

As Charlie and Mary would say, "Knowledge is power!" By utilizing these genetic insights, breeders can make informed choices to promote healthier lives for their pets.

Conclusion

In conclusion, while MMVD remains a significant challenge for breeders and pet owners alike, advancements in genetic research offer hope. The identification of risk alleles and the development of genetic tests are crucial steps forward in managing this disease. By integrating these genetic tests with traditional screening methods, breeders can make more informed decisions and reduce the prevalence of MMVD in at-risk breeds.

For more detailed information, please refer to our comprehensive FACT sheet, which can be found on our website. Together, we can work towards a future where our beloved pets live healthier, happier lives. To understand more about this complex test, please refer to our comprehensive FACT sheet, which can be found on our website and the references below.

[Axelsson et al. \(2021\)](#),

<https://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1009726>

[Mead et al. \(2022\)](#)

<https://pubmed.ncbi.nlm.nih.gov/36553559/>

<https://www.cavalierhealth.org/mvdprotocol.htm>

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About the Author

George Sofronidis is the Co-Founder and CEO of Orivet Genetic Pet Care, a leading genetic testing organization based in Australia. George has over 30 years of devoted experience in molecular biology. Since 2014 George has been dedicated to developing and introducing genetic testing for Canines and Felines to breeders and pet enthusiasts globally.

