



Recent Advancements in Understanding Hypertrophic Cardiomyopathy Genetics in Sphynx Cats

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Overview:

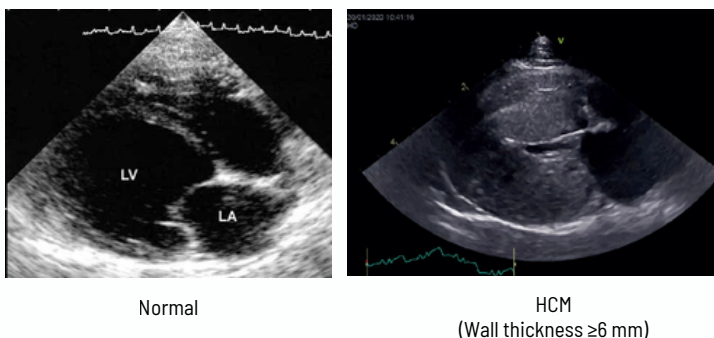
Hypertrophic Cardiomyopathy (HCM) is the most common heart disease in cats, caused by thickening of the heart muscle, especially the left ventricle. This reduces cardiac efficiency and can lead to heart failure, thromboembolism, or sudden death. Sphynx cats have a **high genetic predisposition** to HCM, often developing severe disease at a younger age.

Pathology and clinical features

- **Prevalence in Sphynx:** 21-40% , with males being more affected
- **Clinical features:**
 - Many cats remain asymptomatic until advanced stages
 - Symptoms (when present): tachycardia, labored breathing, lethargy, fainting, or sudden collapse
- **Pathological finding:** microvascular dysfunction causing myocardial ischemia or infarction (heart attack), contributing to early death.

Diagnosis

Echocardiography is the gold standard and only reliable diagnostic tool.



Genetic understanding:

ALMS1 variants

- Initially believed to be a key genetic cause of HCM (autosomal dominant) in Sphynx cats and commercialized as a test
- However, a 2024 New Zealand study found no statistical association between this variant and actual HCM diagnosis, despite its high frequency (~70% in the breed)
- It is now classified as a variant of uncertain significance - possibly a minor risk factor but not a reliable predictor

Other gene variants (MYBPC3, TNNT2, MYH7)

- The 2024 global HCM working group found no proven causal link between these variants and HCM in Sphynx, Devon Rex, or British Shorthair
- MYBPC3 variants remain relevant only for Maine Coon (A31P) and Ragdoll (R820W).

"HCM in Sphynx cats likely arises from a complex multifactorial interplay of several genetic and environmental factors. The disease shows incomplete penetrance, meaning not all cats with a mutation will develop it."

Recommendations



Prioritize annual echocardiographic screening by board-certified veterinary cardiologist. Should begin at 1-2 years of age and continue throughout the cat's life



Responsible breeding by balancing effort to reduce HCM prevalence with crucial goal of maintaining genetic diversity. This process involves meticulous pedigree tracking, transparent health screening results, and thoughtful breeding decisions.



DO NOT rely solely on genetic test for Sphynx HCM as current evidence doesn't support their predictive use. Genetic testing only for HCM in Maine Coon and Ragdoll



Focus on environmental management to support heart health as HCM is multifactorial in nature e.g. maintaining a healthy weight, providing high-quality diet, and maintaining stress.