Original Article

Detecting Polymorphism of Myosin-binding Protein C3 Gene in Persian Breed Cat With and Without Hypertrophic Cardiomyopathy

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ABSTRACT

Background: In cats, hypertrophic cardiomyopathy (HCM) stands out as a prevailing heart disease. The mutations in the gene that encodes cardiac myosin-binding protein C (*MYBPC3*) have been detected in the Ragdoll and Maine Coon breeds.

Objectives: HCM is believed to be hereditary in other breeds, too.

Methods: Blood samples were collected for DNA extraction from 2 unaffected and 7 affected Persian breed cats with HCM. Besides accomplishing conventional polymerase chain reaction, DNA sequencing was performed. The sequence changes were utilized to detect single nucleotide polymorphisms in the *MYBPC3* gene and predict amino acid substitutions based on the Acc. No. XM_019812396.1 and comparisons with the literature on identified breed variants and control samples.

Results: Although many single nucleotide polymorphisms were found in the affected and unaffected Persian cats, no causative mutation for HCM was observed.

Conclusion: In this breed, HCM does not seem to be caused solely by mutations in this cardiac gene. Potential cardiac genes should be investigated to uncover other genetic reasons for this cardiac disease in the Persian cat breed.

Keywords: Hypertrophic cardiomyopathy (HCM), Myosin-binding protein C3, Persian breed, Cat, Polymerase chain reaction (PCR)

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Introduction

Hypertrophic cardiomyopathy (HCM) is defined as left ventricular myocardium hypertrophy without any existing disease or anomaly. HCM is a widely known disease in humans but is especially common in cats. HCM is often an occult disease, which can result in potentially fatal complications. In cats, it is a prevalent heart disease, impacting approximately 15% of their population (Payne et al., 2015). It is frequently related to specific breeds, including Ragdoll, Maine Coon, and British Shorthair (Meurs et al., 2007). However, most patients are non-breed domestic cats (Côté et al., 2011).

In addition, diagnosing heart abnormalities by cardiac auscultation alone is difficult. Therefore, para-clinical examinations, such as radiography and echocardiography, must confirm the diagnosis (Sadri et al., 2022).

The limited treatment options for HCM result in poor prognosis in cats. Thus, the significance of etiology knowledge for prevention measures is neglected. HCM is associated with an underlying genetic defect in humans, which is also typically accepted in cats. However, the genetic variant causing the disease has largely remained unclear in cats, and only two breed-specific variants have been identified (Schipper et al., 2019).

A genetic etiology is suspected in some breeds, including domestic shorthairs (DSH) (Côté et al., 2011). In cats, however, only two variants causing *MYBPC3* mutations have been detected; the XM_019812396.1: c.91G >C in Maine Coons (Meurs et al., 2005) and the c.2455C >T in Ragdolls (Meurs et al., 2007). In humans, HCM is caused by the latter variant ortholog (Schipper et al., 2019).

MYBPC3 (human Gene ID: 4607) comprises 33 exons that can encode protein C of 1274 amino acids binding cardiac myosin. Apparently, the protein is crucial in controlling sarcomere contractility and could also influence sarcomere architecture (Gupta et al., 2014). The widely known variants as causatives in *MYBPC3* can be either truncating or missense (Walsh et al., 2017a).

HCM, in humans, is a relatively complex disease whose pathogenesis, genetics, and modifying factors are still unclear. However, a limited knowledge of feline HCM is currently available, suggesting many similarities between species.

In cats, a more comprehensive knowledge of the genetic aspect of HCM would allow us to screen the genetics of breeding animals more comprehensively. Then, the breeders can restrict the spread of variants causing disease in the cat population, reducing the HCM incidence.

Given the popularity of the Persian cat breed among owners in Iran, we intended to conduct the first study on genetic hypertrophic cardiomyopathy in cats in Iran.

Based on these considerations, the present study aimed to determine the variants causing disease in affected Persian cats. We performed *MYBPC3* gene analysis due to its significance in human HCM.

Materials and Methods

Sample selection

From September 2020 to March 2022, we examined the Persian breed cats presented to the Small Animal Hospital of the Veterinary Medicine Faculty, Tehran University, for cardiovascular examination or follow-up if they were previously diagnosed with HCM or had significant cardiopulmonary clinical signs. Based on the general examination, auscultation of lung and heart sounds, ECG evaluation, and Doppler blood pressure measurement, hypertrophic cardiomyopathy (HCM) was suspected. The cats were then referred to the diagnostic imaging department for chest radiography and echocardiography for evidence of HCM if left ventricular free wall thickness in diastole (LVFWd), and interventricular septum in diastole (IVSd) were >6 mm or both were detected by M-mode echocardiography (Figure 1).

Once HCM cases were identified on echocardiographic examination, other causes of left ventricular hypertrophy in cats, such as renal failure, hyperthyroidism, and elevated blood pressure, were excluded by screening renal levels (blood urea nitrogen [BUN], serum creatinine [Scr]), thyroid levels (total thyroxine [TT4]), and Doppler blood pressure. Finally, two groups of cases were selected: 7 affected cats (3 females and 4 males, 2 to 8 years old, with 5.5 years old as mean age) with HCM and 2 healthy cats (unaffected) (1 male and 1 female, aged 2-4 years old, with 3 years old as mean age) (Table 1). Blood samples were collected in EDTA tubes for all cases and kept at -18°C until DNA extraction.

Primer design

In cats, assembly of the *Felis catus* (Acc. No.: XM_019812396.1) was employed to identify where

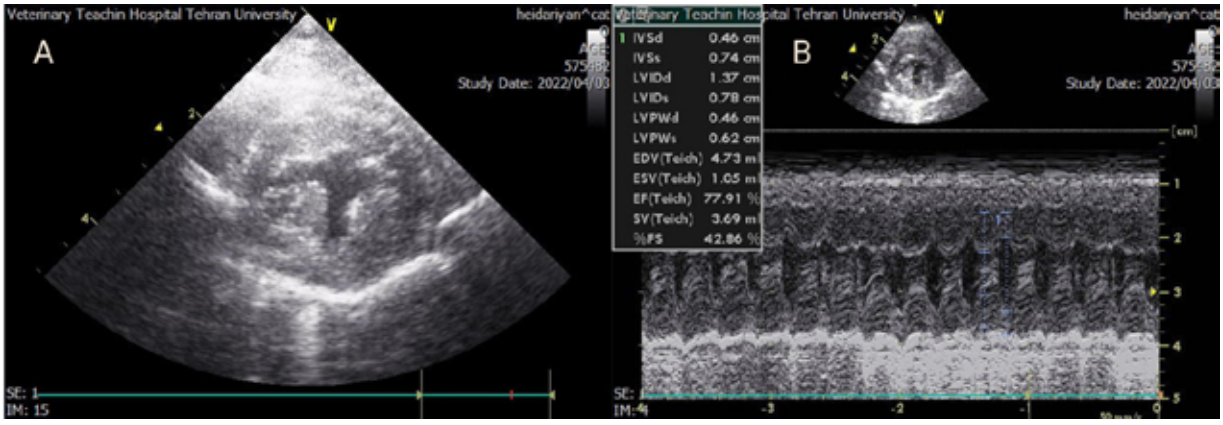

Figure 1. A) Two-dimensional echocardiographic image of a cat with HCM, B) M-mode echocardiographic measurement of LVFWd and IVSd >6 mm

the exons are located and the *MYBPC3* gene sequence. The selected primers in the present study were obtained from Schipper et al. (2019), and many primers were designed based on the Acc. No. XM_019812396.1 using the PRIMER 3 (Table 2).

DNA extraction

In all cases, the blood samples were collected to extract their DNA, utilizing a DNA extraction kit, following the manufacturer's instructions (Sinaclon, EX6071, Iran) (Ghalyanchilangeroudi et al., 2021). It was recommended to keep the extracted DNA at -20˚C.

Conventional PCR

Polymerase chain reaction (PCR) was carried out in 25 µL as final volume by a ready-to-use 2X Master Mix, containing 0.2 mM of each dNTP, 2 mM $MgCl_2$, 2 U Taq DNA polymerase, 10X PCR buffer (Sinaclon, Iran), along with 1 µL of 10 pmol concentration of each primer (Sinaclon, Iran). The thermal conditions included initial denaturation for 5 min at 95˚C, then 35 cycles for 30 s

Table 1. Phenotypic information for HCM-Affected and non-affected Persian cats breed

HCM-affected	Age (y)	Gender	IVSd (mm)	LVFWd (mm)	Renal Panel		Thyroid Panel	BP (mm Hg)
					BUN (mg/dL)	Scr (mg/dL)	TT4 (mcg/dL)	
1	8	F	6	13	28	1.2	6.5	120
2	7	M	8.5	12	30	1.37	5.5	130
3	2	M	6.6	8	23	1.31	8	100
4	4	F	5.1	10	19.6	1.4	7.5	120
5	3	M	6	6.2	23	1.8	5.8	100
6	8	F	6	8.3	24	1.6	6.8	130
7	6	M	8.7	6.7	25	1.8	5.8	140
Non-affected								
1	4	F	3.2	5.5	32	1.2	6.2	100
2	2	M	4.3	3.8	30	1.3	5.8	100

Abbreviations: IVSd: Interventricular septum in diastole; LVFWd: Left ventricular free wall thickness in diastole; BUN: Blood urea nitrogen; Scr: Serum creatinine; TT4: Total thyroxine; BP: Blood pressure.

Table 2. Selected PCR primers for detecting polymorphism in *MYBPC3* gene (Schipper et al., 2019)

Exon	Forward	Reverse
1	5'-TCAGAAGGATGGGAAGGAGAAACCAAGA-3'	3'-GGTTGGGCAAGAGGCAGATAAGAAATCC-5'
2	5'-TCAGCCAAAGCAAAGGCGAGACA-3'	3'-CCAGGAAGGAAGGGTCAGGTATCCAA-5'
6	5'-ACCCCACATTCTGAGCCTTTCCA-3'	3'-CTCCTCCCACTCTTCCACAGTCTT-5'
7, 8, 9	5'-GACAGACAGGGAAATTGGTTTAGAGAGGT-3'	3'-CAAGGGTCATGGATGGGCAGGT-5'
13, 14	5'-TCTGGCACTCACCACTTGACCT-3'	3'-CCACGGCGATGCGTGTGA-5'

at 95˚C, for 30 s at 52˚C, for 1 min at 72˚C, and a final extension for 7 min at 72˚C. The analysis of the PCR product was performed using electrophoresis on a 1.5% agarose gel and then illustrated under UV light. The PCR results were later examined to identify the nucleotide sequences (Peighambari et al., 2022).

Partial sequencing

All PCR products with sufficient amplification of the correct fragment were taken for Sanger sequencing. The quality of the sequences was assessed individually using Chromass software, version 2.6.5. CLC viewer software, version 8 was used to examine nucleotide and amino acid sequences. The splice site and exon sequences of the gene of feline *MYBPC3* were obtained from feline contigs utilizing NCBI blast function and prior studies. The nucleotide sequences were compared to the published normal feline sequence from the feline contigs, as well as previously described mutations in Ragdoll and Maine Coon cats.

Results

In this study, positive PCR results detected the polymorphism of the *MYBPC3* gene. This research's partial *MYBPC3* gene sequences were deposited in GenBank with No: NC018732.3 (Table 3).

Sequencing of exons 1, 2, 8, and 9 revealed no single base pair change in all affected and unaffected control cats; a single nucleotide polymorphism (SNP) change was, however, spotted in exon 6 (c.866 A >G) in 3 cats and exon 13 (c.677 C >A) in 2 cats with HCM and exon 7 (c.803 C >T) in 1 affected and 1 unaffected cat (Figure 2).

Although several SNPs were detected in the gene analyzed in this study, they did not isolate with the disease or alter the amino acid.

The previously reported mutations in the *MYBPC3* gene in Ragdoll and Maine Coon cats were not detected in any of the affected or unaffected cats in this study.

Table 3. The positions of the exons sequenced using the accession numbers to which the sequences compared

Exon	Map of the Target (Location)	Accession Number
1	101341929-101342043	XM_019812396.1
2	101340571-101340837	XM_019812396.1
6	101338545-101338662	XM_019812396.1
7	101337987-101338035	XM_019812396.1
8	101337779-101337808	XM_019812396.1
9	101337568-101337621	XM_019812396.1
13	101335576-101335708	XM_019812396.1

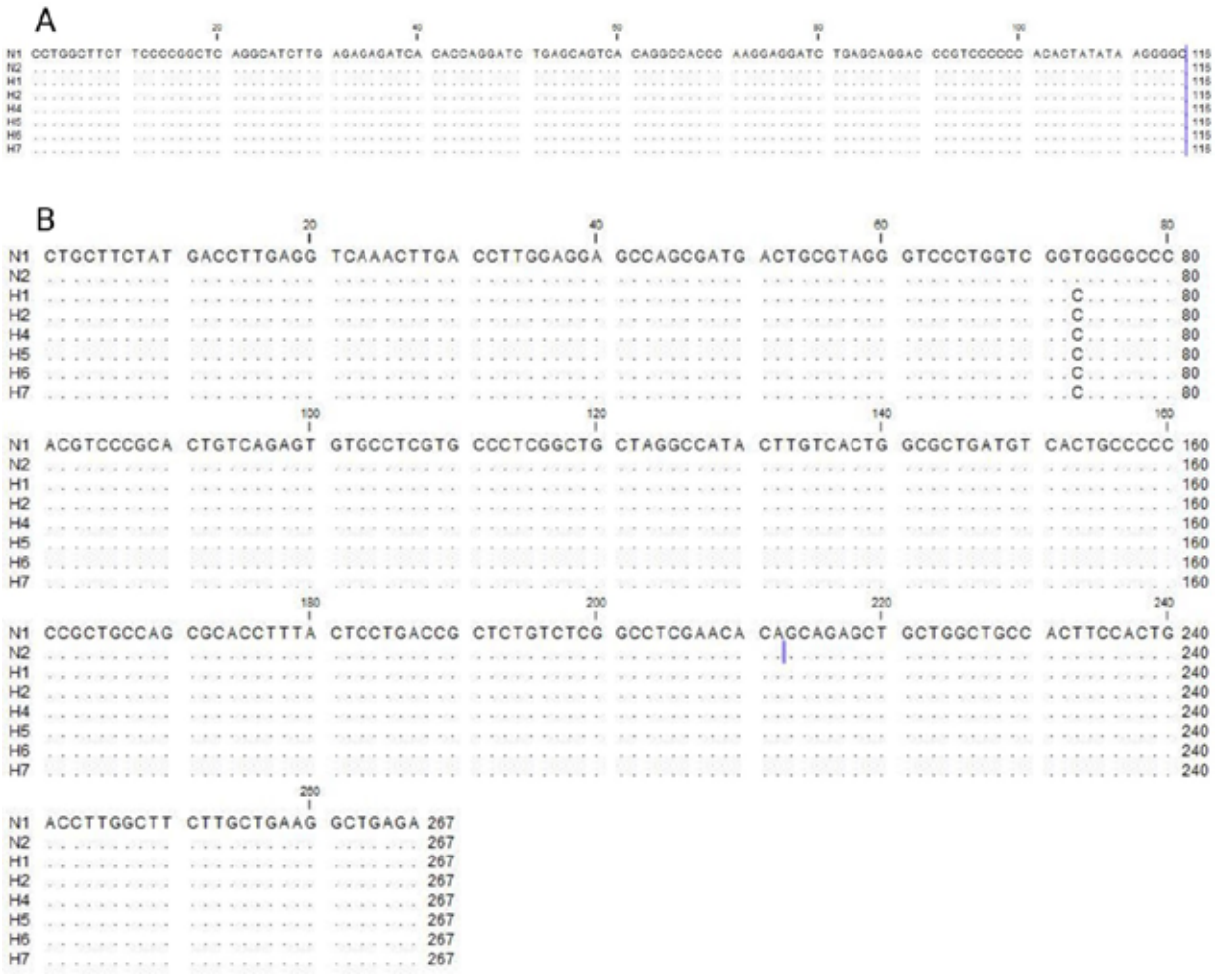

Figure 2. A) Exon 1 and B) Exon 2 sequencing and SNP detection in HCM-affected (H) and unaffected (N) Persian cats

Discussion

In the study of Schipper et al. (2019), 27 variants were found in *MYBPC3*, of which 17 were synonymous and 10 were missense. The variant c.175G >A p. (Ala59Thr) in exon 2 was found in 1 Maine Coon and 3 DSH cats, the c.220G >A p. (Ala74Thr) in exon 2 in 1 Ragdoll, 1 Maine Coon, 2 BSH, and 7 DSH cats, the c.222C >T p, (Ala74=) in exon 2 in 1 Maine Coon and 2 DSH cats, the c. 720C >T p. (Ser240=) in exon 6 in 1 DSH cat, the c. 772G >A p. (Val258Ile) in exon 7 in 7 DSH, 2 British Shorthair cats (BSH), 1 Ragdoll, and 1 DSH×Persian cats, and the c.1140G >C p.(Arg380=) in exon 13 in 1 DSH cat. These results do not align with the present research regarding the *MYPBC3* gene.

Longeri et al. (2013) reported a substitution p.(Ala74Thr) in Maine Coons with HCM, not matching the present research's variants regarding the *MYPBC3* gene.

In cats with HCM, Meurs et al. (2009) explained eight variants in MYBC3. However, the annotation is slightly unclear, and the variants could be c.220G >A or c.222C >T (exon 2), c.772G >A (exon 7), c.1032C >T, c.1956C >T, c.2607C >T, c.2765C >T, and c.3109G >A, as Schipper et al. (2019) explained.

No definitive conclusions, however, can be achieved about these variants since Meurs et al. (2009) did not explain them at the nucleotide level. The eighth variant by Meurs et al. (2009) could be related to none of the variants reported by Schipper et al. (2019).

In the study of Schipper et al. (2019), 10 missense variants, potentially pathogenic, were detected in *MYBPC3*. The amino acid substitution impacts were anticipated in silico to evaluate their pathogenicity, and the affected amino acid's conservation was evaluated. The substitution of Ala59Thr, Ala74Thr in exon 2, and V258Ile in exon 7 caused by missense variants in MYPBC3 were

benign (based on the PROVEN, PolyPhen-2: HumDiv and HumVar and ConSurf score).

There are over 1500 HCM-causing human mutations, mostly found in sarcomere genes. However, pathogenic variants in non-sarcomeric genes, such as those calcium signaling proteins or encoding Z-disk proteins, have also been identified (Walsh et al., 2017b). The *MYBPC3* gene is most usually related to HCM in humans, accounting for 40% of cases, followed by the *MYH7* gene (Carrier et al., 2015). In the *MYBPC3* gene, only two mutations have been explained in cats, although the sequence present in this gene has been largely conserved in mammals (cow, dog, human, mouse, rat) (Maron et al., 2015; Kittleson et al., 2015).

Numerous investigations have been conducted based on sequencing the *MYBPC3* gene fragment where this variant appears. The populations examined were mostly Maine Coon cats, either with or without HCM, even though some other breeds were examined (up to 3757 cats belonging to 17 breeds) as well. The p.A31P mutation has been identified in both affected and unaffected Maine Coon cats; in other breeds, only three cases have been isolated and reported: A Siberian, a Ragdoll, and a British longhair cat (Gil-Ortuño et al., 2020).

The SNPs in the cats investigated here did not meet the threshold for a causative mutation since they did not consistently affect the amino acid produced or isolated with the disease. Nevertheless, these SNPs could be proven effective in future research of familial feline HCM in various cat breeds through linkage analysis or broader relationship studies.

The absence of a mutation in the *MYBC3* gene region does not rule out its potential role in this disease in the Persian cat since a causative mutation may occur in this gene's promoter or untranslated areas. The cats studied could still have family feline HCM caused by a mutation in one of the other HCM genes in humans, or they could have a mutation in a gene not yet linked to HCM. Finally, these cats' sickness may have a distinct etiology.

To summarize, felines are the natural model of HCM that mimics the genotype and phenotype of humans. The cat is the ideal species to investigate the efficacy of new genetic and preventive therapies due to its life expectancy, typically about 16 years, and the disease's typical early onset between 3 and 5 years old (Gil-Ortuño et al., 2020; Sleeper et al., 2009). These opportunities should motivate researchers to use cutting-edge genetic sequencing techniques to examine HCM in cats. Collaboration between doctors and veterinarians could benefit both species, humans, and cats, affected by this dreadful disease.

Conclusion

In particular breeds (Ragdoll and Main Coon), there are only two genetic variants related to feline HCM (*p.R820W* and *p.A31P*), both in the *MYBPC3* gene. The detected variants, in high frequency, in unaffected cats support the idea of their minor to moderate effect on phenotype and the probable interplay with other environmental or genetic factors. The genetic causes of HCM in other breeds are unknown. Since the disease is similar in humans and cats, felines are an ideal model for developing new corrective and preventive therapies for human and feline HCM. Investigating the genetic origin of feline HCM should use new genetic testing methods.

Ethical Considerations

Compliance with ethical guidelines

All procedures were conducted according to the animal care guideline of the Research Committee of the Faculty of Veterinary Medicine, University of Tehran.

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Authors' contributions

All authors equally contributed to preparing this article.

Conflict of interest

The authors declared no conflict of interest.

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مقاله پژوهشی

تشخیص وقوع پلی مورفیسم ژن پروتئین میوزین بایندینگ 3C (3CPBYM) در گربه‌های نژاد پرشین با و بدون کاردیومیوپاتی هایپرتروفیک

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چکیده

زمینه مطالعه: کاردیومیوپاتی هایپرتروفیک (HCM) یک بیماری قلبی شایع در گربه‌ها است. جهش‌های ایجادکننده بیماری در ژن میوزین بایندینگ C3 در گربه‌های نژاد مین کون و رگدال شناسایی شده است. به نظر می‌رسد بیماری کاردیومیوپاتی هایپرتروفیک در سایر نژادها نیز زمینه ارثی داشته باشد.

هدف: این مطالعه باهدف شناسایی واریانت‌های ژنتیکی عامل بیماری کاردیومیوپاتی هایپرتروفیک در ژن میوزین بایندینگ C3 در گربه‌های نژاد پرشین در ایران انجام شد.

روش کار: نمونه خون کامل برای استخراج DNA از دو گروه گربه نژاد پرشین مبتلابه کاردیومیوپاتی هایپرتروفیک و سالم جمع‌آوری شد. واکنش زنجیره‌ای پلیمراز استاندارد و همچنین توالی‌یابی بر روی قسمتی از ژن میوزین بایندینگ C3 نمونه‌های گروه‌های بیمار و سالم انجام شد. تغییرات توالی برای تشخیص پلی مورفیسم ژن و پیش‌بینی جایگزینی آمینواسید براساس شماره شناسه (XM_019812396.1) و مقایسه نتایج با جهش‌های شناخته‌شده قبلی در مقالات و نمونه‌های کنترل مورد بررسی قرارگرفت.

نتایج: اگرچه تعدادی جهش تک نقطه‌ای در هر دو گروه سالم و درگیر بیماری یافت شد، اما هیچ جهش ایجادکننده کاردیومیوپاتی هایپرتروفیک گزارش نشد.

نتیجه‌گیری نهایی: در این نژاد، بیماری کاردیومیوپاتی هایپرتروفیک به نظر نمی‌رسد به تنهایی توسط جهش‌ها در این ژن ایجاد شود. ژن‌های قلبی دیگر برای شناسایی عوامل ژنتیکی بیماری کاردیومیوپاتی هایپرتروفیک باید مورد بررسی قرار گیرند.

کلیدواژه‌ها: کاردیومیوپاتی هایپرتروفیک، پروتئین میوزین بایندینگ C3، نژاد پرشین، گربه، واکنش زنجیره ای پلیمراز

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