

Pumpkin  
Registration: N/A  
Breed: Maine Coon

Sample ID: KTBR06800  
Test Date: 11/25/2020  
Optimal Selection - Feline - Legacy

# DNA Test Report

## Owner Info

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**First Name**

Traci

**Last Name**

Heppler

## Pet Info

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**Registered Name**

Pumpkin

**Date of Birth**

6/12/2020

**Nickname (Call Name)**

Pumpkin

**Sample ID**

KTBR06800

**Sex**

Female

**Registration**

N/A

**Country of Origin**

US

**Microchip ID**

N/A

**Owner Reported Breed**

Maine Coon

**Tattoo ID**

N/A

# DNA Test Report

## Health Conditions Known in This Breed

Genetic Condition	Gene	Risk Variant	Copies	Result
Cystinuria Type B (Variant 3)	SCL7A9	T>A	0	Clear
Factor XII Deficiency (Variant 1)	F12	Deletion	0	Clear
Factor XII Deficiency (Variant 2)	F12	Deletion	0	Clear
Hypertrophic Cardiomyopathy (A31P; Discovered in Maine Coon)	MYBPC	G>C	0	Clear
Pyruvate Kinase Deficiency	PLKR	G>A	0	Clear
Spinal Muscular Atrophy (Discovered in Maine Coon)	LIX1	Deletion	0	Clear

## Other Conditions Tested

Genetic Condition	Gene	Risk Variant	Copies	Result
Acute Intermittent Porphyria (Variant 1)	AIP	Deletion	0	Clear
Acute Intermittent Porphyria (Variant 2)	AIP	G>A	0	Clear
Acute Intermittent Porphyria (Variant 3)	HMBS	Insertion	0	Clear
Acute Intermittent Porphyria (Variant 4)	HMBS	Deletion	0	Clear
Acute Intermittent Porphyria (Variant 5)	HMBS	G>A	0	Clear
Autoimmune Lymphoproliferative Syndrome	FASL	Insertion	0	Clear
Burmese Head Defect (Discovered in Burmese)	ALX1	Deletion	0	Clear
Chediak-Higashi Syndrome (Discovered in Persian cats)	CHS	Insertion	0	Clear
Congenital Adrenal Hyperplasia	CYP11B1	G>A	0	Clear
Congenital Erythropoietic Porphyria	UROS	G>A	0	Clear
Congenital Myasthenic Syndrome (Discovered in Devon Rex and Sphynx)	COLQ	G>A	0	Clear
Cystinuria Type 1A	SCL3A1	C>T	0	Clear
Cystinuria Type B (Variant 1)	SCL7A9	C>T	0	Clear
Cystinuria Type B (Variant 2)	SCL7A9	G>A	0	Clear

# DNA Test Report

## Other Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Dihydropyrimidinase Deficiency	DPYS	G>A	0	Clear
Earfold and Osteochondrodysplasia (Discovered in the Scottish Fold)	TRPV4	G>T	0	Clear
Familial Episodic Hypokalaemic Polymyopathy (Discovered in Burmese)	WNK4	C>T	0	Clear
Glutaric Aciduria Type II	ETFDH	T>G	0	Clear
Glycogen Storage Disease (Discovered in Norwegian Forest Cat)	GBE1	Insertion	0	Clear
GM1 Gangliosidosis	GLB1	G>C	0	Clear
GM2 Gangliosidosis	GM2A	Deletion	0	Clear
GM2 Gangliosidosis, type II (Discovered in Burmese cats)	HEXB	O>O	0	Clear
GM2 Gangliosidosis, type II (Discovered in domestic shorthair cats)	HEXB	Insertion	0	Clear
GM2 Gangliosidosis, type II (Discovered in japanese domestic cats)	HEXB	C>T	0	Clear
Hemophilia B (Variant 1)	F9	C>T	0	Clear
Hemophilia B (Variant 2)	F9	G>A	0	Clear
Hyperoxaluria type II	GRHPR	G>A	0	Clear
Hypertrophic Cardiomyopathy (Discovered in Ragdoll)	MYBPC	C>T	0	Clear
Hypotrichosis (Discovered in Birman cats)	FOXN1	Deletion	0	Clear
Lipoprotein Lipase Deficiency	LPL	G>A	0	Clear
Medication Sensitivity (MDR1)	ABCB1	Deletion	0	Clear
Mucopolysaccharidosis Type I	IDUA	Deletion	0	Clear
Mucopolysaccharidosis Type VI (mild form)	ARSB	G>A	0	Clear
Mucopolysaccharidosis Type VI (severe)	ARSB	T>C	0	Clear
Mucopolysaccharidosis Type VII	GUSB	G>A	0	Clear
Mucopolysaccharidosis VII	USB	C>T	0	Clear

# DNA Test Report

## Other Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Myotonia Congenita	CLCN1	G>T	0	Clear
Polycystic Kidney Disease (PKD)	PKD1	C>A	0	Clear
Progressive Retinal Atrophy (Discovered in Bengal cats)	KIF3B	G>A	0	Clear
Progressive Retinal Atrophy (Discovered in Persian cats)	PRA	C>T	0	Clear
Progressive Retinal Atrophy (rdAc-PRA)	CEP290	T>G	0	Clear
Sphingomyelinosis (Variant 1)	NPC1	G>C	0	Clear
Sphingomyelinosis (Variant 2)	NPC2	G>A	0	Clear
Vitamin D-Dependent Rickets	CYP27B1	G>T	0	Clear

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## Coat Color

Genetic Trait	Gene	Variant	Copies	Result
Charcoal (Discovered in Bengal cats)	ASIP	A <sup>Pb</sup>	0	No effect
Solid color (Non-Agouti)	ASIP	a	1	<b>Agouti (banded) hair</b>
Partial and Full White	FERV1	W or w <sup>s</sup>	1	<b>Partial or Full White likely</b>
Gloving (Birman cat - White Gloves)	KIT	w <sup>g</sup>	1	<b>No effect</b>
Amber (Discovered in Norwegian Forest Cat)	MC1R	e	0	No effect
Russet (Discovered in Burmese cats)	MC1R	e <sup>r</sup>	0	No effect
Dilution	MLPH	d	0	No effect
Albinism (Discovered in Oriental breeds)	TYR	c <sup>a</sup>	0	No effect
Colorpoint (Discovered in Burmese cats)	TYR	c <sup>b</sup>	0	No effect
Colorpoint (Discovered in Siamese cats)	TYR	c <sup>s</sup>	0	No effect
Mocha (Discovered in Burmese cats)	TYR	c <sup>m</sup>	0	No effect
Chocolate	TYRP	b	0	No effect
Cinnamon	TYRP	b <sup>l</sup>	0	No effect

## Coat Type

Genetic Trait	Gene	Variant	Copies	Result
Long hair (Mutation M1, discovered in Ragdolls)	FGF5	M1	0	No effect
Long hair (Mutation M2, discovered in Norwegian Forest cats)	FGF5	M2	0	No effect
Long hair (Mutation M3, discovered in Ragdolls and Maine Coon)	FGF5	M3	1	<b>Long coat carrier</b>
Long hair (Mutation M4, common)	FGF5	M4	1	<b>Long coat carrier</b>
Lykoi coat (Mutation Ca)	HR	hr <sup>Ca</sup>	0	No effect

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## Coat Type (continued)

Genetic Trait	Gene	Variant	Copies	Result
Lykoi coat (Mutation VA)	HR	hr <sup>VA</sup>	0	No effect
Hairlessness (Discovered in Sphynx cats)	KRT71	re <sup>hr</sup>	0	No effect
Rexing (Discovered in Devon Rex)	KRT71	re <sup>dr</sup>	0	No effect
Rexing (Discovered in Cornish Rex and German Rex)	LPAR6	r	0	No effect
Glitter	Pending	gl	0	No effect

## Body Features

Genetic Trait	Gene	Variant	Copies	Result
Short tail (Discovered in Japanese Bobtail cats)	HES7	jb	0	No effect
Polydactyly (Mutation HW)	LIMBR1	HW	0	No effect
Polydactyly (Mutation UK1)	LIMBR1	UK1	0	No effect
Polydactyly (Mutation UK2)	LIMBR1	UK2	0	No effect
Short tail (Discovered in Manx - variant 1)	T	C1199del	0	No effect
Short tail (Discovered in Manx - variant 2)	T	T988del	0	No effect