

Rafaela

Breed: Maine Coon

Microchip number: 900215001277550

Birth date: 2020-12-05

Registration number: SBT120520094

Test date: 2024-07-17

ID kit: FLNGQWH

Rafaela's Profile

Pet information

Registered name

Rafaela

Sex

F

Owner reported breed

Maine Coon

Date of birth

2020-12-05

Microchip number

900215001277550

Genetic Diversity

Rafaela's Percentage of Heterozygosity

34%

Health summary

At Risk 0 conditions

Carrier 0 conditions

Clear 50 conditions

Rafaela

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Genetic Diversity

Heterozygosity

Rafaela's Percentage of Heterozygosity

34%

Rafaela's genome analysis shows an average level of genetic heterozygosity when compared with other Maine Coons.

Typical Range for Maine Coons

32% - 37%

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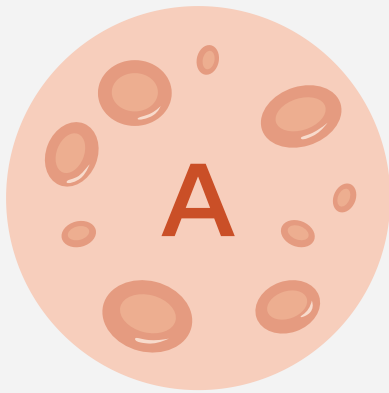
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Blood Type



Blood type

Type A (Most common)

Genotype*

A/A

Transfusion risk

⚠ Moderate

Rafaela has the most common blood type. She can be transfused with Type A blood.

Breeding risk

✅ Low

If breeding, Rafaela has a low risk of blood type incompatibility with nursing kittens.

Blood variants tested*

Variant Tested	Description	Copies
b variant 1	(Common b variant)	0
b variant 2	(Discovered in Turkish breeds)	0
b variant 3	(Discovered in Ragdolls)	0
c variant - Causes AB Blood Type	(Discovered in Ragdolls)	0

*This test identifies three known 'b' variants and one known 'c' variant in the CMAH gene when determining a cat's genetic blood type. Blood Type A is inferred in reporting when less than two genetic blood variants are detected.

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Interpreting feline blood types

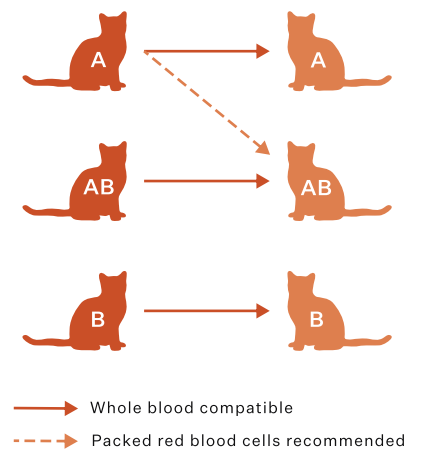
About blood type determination

The three important feline blood types of A, B, and AB are governed primarily by variants in the CMAH gene. A cat's blood type can be determined by its genotype, which consists of two gene variants – one inherited from each parent – that should be interpreted together. When determining blood type based on genotype, the A variant associated with blood type A is most dominant while the b variants associated with blood type B are most recessive. The c variant associated with blood type AB is intermediate between the A and b variants, meaning it is recessive to the A variant but dominant to b variants. Therefore, a genotype with at least one A variant will result in blood type A. For a cat to have blood type B, the genotype must consist of two b variants. Because the c variant is intermediate, a cat with blood type AB can either have a genotype consisting of two c variants or one c variant and one b variant.

About transfusion risk

Similar to humans, the different cat blood types will express different antigens on the surface of their red blood cells. This is significant because both type A and B cats are born with antibodies against other blood cell antigens. Notably, type B cats have high levels of antibodies against type A antigens. Cats with the rare blood type AB are most versatile as they express both red cell antigen types and, thus, can receive both type A and type AB blood transfusions.

Unlike humans, there is no cat blood type that can act as a universal blood donor. If a cat receives a non-compatible blood type during a transfusion, it may cause a severe, life-threatening reaction including fever, kidney failure, and widespread destruction of red blood cells. Prior to all transfusions, cats should be serologically typed and crossmatched to ensure compatibility.



About breeding risk

During pregnancy, kittens are shielded from their mother's immune system. However, when kittens begin nursing, they receive some of their mother's antibodies in colostrum. Type B cats have high levels of antibodies against type A blood, so when blood type A or AB kittens are born to a blood type B mother, these antibodies, when absorbed by the newborn kitten, cause neonatal isoerythrolysis, a potentially fatal destruction of the kitten's red blood cells. Kittens of type B mothers with fathers of unknown or type A blood should be bottle fed or foster-nursed, and separated from their mother for the first 24 hours to avoid this reaction, unless blood typing performed immediately following birth shows the kitten to have a compatible blood type to the mother.

Although some blood types are less common and require additional planning when breeding, they represent normal genetic variation and should not be selected against when choosing breeding pairs.

Current limits of this test

This test identifies 4 variants (b variants c.269T>A, c.179G>T, c.1233delT and c variant c.346C>T) in the CMAH gene discovered in the domestic cat population and has been confirmed 99% concordant with serologic blood typing¹. Mik antigens also play a role in blood type compatibility, and are not included in this test. Cats carrying undetermined, new, or undiscovered variants in CMAH or other genes may have a different blood type compatibility than that reported by this test. Accuracy of this test at predicting blood type in wildcats or wildcat hybrid breeds has not been determined.

1. Anderson H, Davison S, Lytle KM, Honkanen L, et al. Genetic epidemiology of blood type, disease and trait variants, and genome-wide genetic diversity in over 11,000 domestic cats (2022) PLOS Genetics.

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Health conditions known in the breed

	Gene	Risk Variant	Copies	Inheritance	Result
Cystinuria Type B (Variant 3)	SCL7A9	T>A	0	AR	Clear

Information about the genetic condition

Cystinuria is a metabolic disorder characterized by the formation of cystine calculi and stones in the urinary tract. The disease is caused by defective renotubular reabsorption of amino acids (arginine, lysine, cystine, and ornithine) resulting in the formation of urinary cystine crystals, urolithiasis, and urinary tract obstruction in some cases. Clinical signs may develop as early as two months of age; however, some individuals may not show signs until middle-age. Clinical signs include nonspecific signs of feline lower urinary tract disease, such as stranguria, pollakiuria, and hematuria. Urolithiasis has potential to cause urinary obstruction. Urinary obstructions require rapid intervention to prevent subsequent development of renal failure. Urinary stones usually require surgical removal. Not all cystinuric cats will form cystine crystals or uroliths, and cats with a later onset of clinical signs may have a milder degree of cystinuria than cats developing signs at an early age. The recurrence of uroliths is very high in affected cats. In addition to the clinical signs of lower urinary tract disease, cystinuric cats may show neurologic signs such as hypersalivation, lethargy, and even seizures. Various causative mutations have been found in cats with equal frequency in males and females.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to be shown. A carrier cat with one copy of the Feline Cystinuria mutation can be safely bred with a clear cat with no copies of the Feline Cystinuria mutation. About half of the kittens will have one copy (carriers) and half will have no copies of the Feline Cystinuria mutation. Kittens in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected kittens. Please note: It is possible that disease signs similar to the ones caused by the Feline Cystinuria mutation could develop due to a different genetic or clinical cause.

	Gene	Risk Variant	Copies	Inheritance	Result
Factor XII Deficiency (Variant 1)	F12	Deletion	0	ARa	Clear

Information about the genetic condition

Blood coagulation is a complex process involving many pathways. Factor XII, a plasma protein, classically initiates the intrinsic pathway of blood coagulation; although, there are alternative, slower ways to initiate this pathway. Factor XII Deficiency, also known as Hageman Factor Deficiency or Hageman trait, is a commonly inherited blood clotting disorder in cats. Unlike other bleeding disorders, cats deficient in Factor XII are asymptomatic and do not tend to show spontaneous bleeding or abnormal bleeding after surgery or trauma. However, affected individuals can have prolonged clotting time on the activated partial thromboplastin time (aPTT) screening test. Cats who inherit 2 copies of both Factor XII Deficiency (Variant 1) and Factor XII Deficiency (Variant 2) may show even higher aPTT values. Please note that 1 copy of Factor XII Deficiency (Variant 1) and 1 copy of Factor XII Deficiency (Variant 2) will not cause Factor XII Deficiency.

Breeder recommendation

This condition is autosomal recessive, asymptomatic, meaning that cats with two copies of the variant will show the variant-associated condition but will not suffer disease due to this genetic cause. Current understanding is that a cat with one or two copies of the Factor XII Deficiency variant can be safely bred with a cat with zero, one or two copies of the variant. Please note: It is possible that clinical signs similar to the ones caused by the Factor XII Deficiency mutation could develop due to a different genetic or clinical cause.

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Health conditions known in the breed

	Gene	Risk Variant	Copies	Inheritance	Result
Factor XII Deficiency (Variant 2)	F12	Deletion	0	ARa	Clear

Information about the genetic condition

Blood coagulation is a complex process involving many pathways. Factor XII, a plasma protein, classically initiates the intrinsic pathway of blood coagulation; although, there are alternative, slower ways to initiate this pathway. Factor XII Deficiency, also known as Hageman Factor Deficiency or Hageman trait, is a commonly inherited blood clotting disorder in cats. Unlike other bleeding disorders, cats deficient in Factor XII are asymptomatic and do not tend to show spontaneous bleeding or abnormal bleeding after surgery or trauma. However, affected individuals can have prolonged clotting time on the activated partial thromboplastin time (aPTT) screening test. Cats who inherit 2 copies of both Factor XII Deficiency (Variant 1) and Factor XII Deficiency (Variant 2) may show even higher aPTT values. Please note that 1 copy of Factor XII Deficiency (Variant 1) and 1 copy of Factor XII Deficiency (Variant 2) will not cause Factor XII Deficiency.

Breeder recommendation

This condition is autosomal recessive, asymptomatic, meaning that cats with two copies of the variant will show the variant-associated condition but will not suffer disease due to this genetic cause. Current understanding is that a cat with one or two copies of the Factor XII Deficiency variant can be safely bred with a cat with zero, one or two copies of the variant. Please note: It is possible that clinical signs similar to the ones caused by the Factor XII Deficiency mutation could develop due to a different genetic or clinical cause.

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	Gene	Risk Variant	Copies	Inheritance	Result
Hypertrophic Cardiomyopathy (Discovered in the Maine Coon)	MYBPC	G>C	0	AR	Clear

Information about the genetic condition

Hypertrophic Cardiomyopathy (HCM) is the most common cardiac disease in cats worldwide. The disorder is characterized by an increase in heart wall thickness of the left ventricle and the interventricular septum. This causes turbulence in blood flow and increased venous pressure in the left atrium and lungs, which may or may not coincide with a cardiac murmur upon auscultation. The clinical signs most commonly noted are respiratory signs associated with congestive heart failure such as tachypnea, exercise intolerance and panting, difficulty breathing, and (rarely) coughing. Thromboembolism may also occur, and affected cats have an increased risk of sudden cardiac death. Clinical and echocardiographic signs typically appear after the cat has reached breeding age, as the most common age of diagnosis for HCM is five to seven years.

In the Maine Coon, a point mutation (A31P) in the MYBPC3 gene has been found to be causative for the disease with a mode of inheritance that is best described as autosomal recessive (Boeykens, et al. 2024). Research indicates cats with two copies of the mutation (homozygotes) have significantly increased risk of developing HCM while no increased risk has been found in cats with only one copy of the mutation (heterozygotes). HCM has also been diagnosed in Maine Coons negative for A31P, thus indicating that this variant is not the sole cause of HCM in Maine Coons.

Breeder recommendation

This genetic variant is considered to most closely follow an autosomal recessive mode of inheritance meaning that usually two copies of the variant are needed for disease signs to occur. Due to the high frequency of the HCM variant within the Maine Coon breed, cats with one copy of the disease variant may be considered for breeding with cats tested clear for the HCM variant in order to maintain genetic diversity within the breed. Approximately half of the kittens in a litter from this breeding will have no copies of the HCM variant and half will have one copy of the HCM variant. Kittens in a litter expected to contain carriers should be tested before breeding. Carrier to carrier matings are not advised as the resulting litter may contain kittens with two copies of the HCM variant. Please note: It is possible that disease signs similar to the ones caused by this HCM variant could develop due to a different genetic or clinical cause.

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	Gene	Risk Variant	Copies	Inheritance	Result
MDR1 Medication Sensitivity	ABCB1	Deletion	0	AR	Clear

Information about the genetic condition

Cats with this variant are asymptomatic until exposed to a medication that uses the drug transport pump rendered defective by the mutation in the MDR1 (also called ABCB1) gene. Drugs known to use this P-glycoprotein pump are macrocyclic lactones including eprinomectin-containing products labeled for use in cats (antiparasitic drugs), loperamide (antidiarrheal), erythromycin (antibiotic), acepromazine (tranquilizer), butorphanol (opioid), certain drugs used in cancer treatment (vincristine, vinblastine, doxorubicin), and possibly others still to be determined. When these medications are administered, they accumulate in the brain which results in adverse reactions. Typical symptoms involve generalized neurologic dysfunction which may include mydriasis, dyspnea, tremors, hyperreactivity, ataxia or paresis. In more severe cases cats may experience seizures, coma and death. However, with appropriate supportive care by a veterinarian, most affected cats may be able to fully recover.

Breeder recommendation

Further research is needed to determine if cats with one copy of the variant may have altered drug responses. At this time, breeding cats with one or two copies of the MDR1 Medication Sensitivity variant should be approached with caution. If a cat with one copy of the MDR1 Medication Sensitivity variant is bred with a clear cat with no copies of the MDR1 Medication Sensitivity variant, on average half of the kittens will have one copy and half will have no copies of the MDR1 Medication Sensitivity variant. If a cat with two copies of the MDR1 Medication Sensitivity variant is bred with a clear cat with no copies of the MDR1 Medication Sensitivity variant, the resulting kittens will all have one copy of the MDR1 Medication Sensitivity variant. If litters are expected to contain kittens with the MDR1 Medication Sensitivity variant, the kittens should be DNA tested as they may show signs of sensitivity to some common medications. Carrier to carrier matings are not advised as the resulting litter may contain kittens with two copies of the MDR1 Medication Sensitivity variant, which is known to cause medication sensitivity. Please note: It is possible that clinical signs similar to the ones caused by the MDR1 Medication Sensitivity variant could develop due to a different genetic or clinical cause.

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	Gene	Risk Variant	Copies	Inheritance	Result
Polycystic Kidney Disease (PKD)	PKD1	C>A	0	AD	Clear

Information about the genetic condition

Polycystic Kidney Disease (PKD), also named autosomal dominant PKD, is characterized by variously sized, fluid-filled cysts in the renal cortex and medulla with hepatic and pancreatic cysts also possible. The cysts develop from birth and enlarge with age. The cysts destroy the renal parenchyma and disturb renal function, eventually causing renal failure. Affected cats present with signs of renal insufficiency such as weight loss, decreased appetite, increased drinking and urination, poor body condition, and vomiting. Biochemical labwork and ultrasonography examination are helpful tools in identifying the severity of disease within an affected individual. An autosomal dominant point mutation in the PKD1 gene has been identified as the most common genetic mutation for the disease. No homozygous cats have been identified, suggesting the mutation is a homozygous lethal mutation in utero. PKD is very common in Persian and Persian-related cats, affecting approximately 38% of Persian cats worldwide. While there is no known sex linkage to the inheritance of the mutation, research has shown male cats have a higher prevalence of the mutation.

Breeder recommendation

This disease is autosomal dominant meaning that one copy of the mutation is needed for disease signs to occur. Use of cats with one or two copies of the disease mutation is not recommended, as there is a risk that the resulting litter will contain affected kittens. For example if a cat with one copy of the PKD mutation is bred with a clear cat with no copies of the PKD mutation, about half of the kittens will have one copy and half will have no copies of the PKD mutation. Please note: It is possible that disease signs similar to the ones caused by the PKD mutation could develop due to a different genetic or clinical cause.

	Gene	Risk Variant	Copies	Inheritance	Result
Pyruvate Kinase Deficiency	PKLR	G>A	0	AR	Clear

Information about the genetic condition

Pyruvate Kinase (PK) Deficiency presents as a chronic, intermittent, hemolytic anemia. The disorder has a high variability of age of onset and severity of clinical signs. The age of onset of clinical signs varies from six months to five years of age. Clinical signs of the disorder are highly variable but may include lethargy, weakness, diarrhea, pale mucous membranes, anorexia, poor coat quality, weight loss, icterus (jaundice), splenomegaly, and ascites in severe cases. The severity of clinical signs also varies greatly with some cats maintaining adequate quality of life and others requiring euthanasia. The disorder has been reported in multiple cat breeds.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to be shown. A carrier cat with one copy of the Pyruvate Kinase Deficiency mutation can be safely bred with a clear cat with no copies of the Pyruvate Kinase Deficiency mutation. About half of the kittens will have one copy (carriers) and half will have no copies of the Pyruvate Kinase Deficiency mutation. Kittens in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected kittens. Please note: It is possible that disease signs similar to the ones caused by the Pyruvate Kinase Deficiency mutation could develop due to a different genetic or clinical cause.

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Health conditions known in the breed

	Gene	Risk Variant	Copies	Inheritance	Result
Spinal Muscular Atrophy (Discovered in the Maine Coon)	LIX1	Deletion	0	AR	Clear

Information about the genetic condition

Spinal Muscular Atrophies are hereditary diseases caused by the degeneration of lower motor neurons of the spinal cord. In cats, the disease has an early onset. Progressive weakness and atrophy of skeletal muscles is visible in kittens around twelve weeks of age. The changes are first seen in the proximal musculature of the hindlimbs. Clinical signs progress to gait instabilities, postural abnormalities, and visible muscle loss. Muscle loss and an inability to jump is typically observed at five months of age. In time, distal musculature may also become atrophied. After an initial progression of disease for several months, the ongoing loss of muscle function will likely slow with affected cats reaching a plateau of variable muscle atrophy, weakness, and mobility. Despite the severe clinical signs, the disease is not painful and affected cats may survive with varying degrees of disability into adulthood. The mode of inheritance is autosomal recessive. Please note that this test detects presence or absence of the disease mutation and cannot distinguish cats that have one copy of the disease mutation from cats with two copies of the disease mutation.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to be shown. A carrier cat with one copy of the SMA mutation can be safely bred with a clear cat with no copies of the SMA mutation. About half of the kittens will have one copy (carriers) and half will have no copies of the SMA mutation. Kittens in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected kittens. Please note: It is possible that disease signs similar to the ones caused by the SMA mutation could develop due to a different genetic or clinical cause.

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Traits

Coat Color

	Gene	Variant	Copies	Result
Charcoal (Discovered in the Bengal)	ASIP	A ^{Pb}	0	No effect
Solid Color Two copies of the Solid Color variant are needed for a cat to have solid colored hair. However, orange coloration overrides this effect, meaning that cats with partial or full orange coats can show tabby patterning in orange areas. Cats with zero or one copy of this variant are likely to have a tabby pattern due to color banding of the hairs.	ASIP	a	2	Solid color hairs likely
Gloving (Discovered in the Birman) Two copies of the Gloving variant and Birman ancestry are needed for a cat to show white feet due to this variant. The Gloving variant can be found in non-Birman cats without having an effect on appearance and cats can have white feet without this variant suggesting other causes are yet to be discovered.	KIT	w ^g	1	No effect
Partial and Full White	KIT	W or w ^s	0	No effect
Amber (Discovered in the Norwegian Forest Cat)	MC1R	e	0	No effect
Russet (Discovered in the Burmese)	MC1R	e ^r	0	No effect
Dilution Two copies of the Dilution variant are required to have a lightening effect on the coat.	MLPH	d	1	No effect
Albinism (Discovered in Oriental breeds)	TYR	c ^a	0	No effect
Colorpoint (Discovered in the Burmese)	TYR	c ^b	0	No effect
Colorpoint (Discovered in the Siamese)	TYR	c ^s	0	No effect
Mocha (Discovered in the Burmese)	TYR	c ^m	0	No effect
Chocolate	TYRP	b	0	No effect
Cinnamon	TYRP	b ^l	0	No effect

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Coat Type

	Gene	Variant	Copies	Result
Long Hair (Discovered in many breeds) Two copies of any Long Hair variant must be inherited for a cat to have a long coat. This can either be two copies of a particular variant, such as this one, or two of any combination of Long Hair variants.	FGF5	M4	1	Long coat possible, short coat likely
Long Hair (Discovered in the Norwegian Forest Cat)	FGF5	M2	0	No effect
Long Hair (Discovered in the Ragdoll and Maine Coon) Two copies of any Long Hair variant must be inherited for a cat to have a long coat. This can either be two copies of a particular variant, such as this one, or two of any combination of Long Hair variants.	FGF5	M3	1	Long coat possible, short coat likely
Long Hair (Discovered in the Ragdoll)	FGF5	M1	0	No effect
Lykoi Coat (Variant 1)	HR	hr ^{Ca}	0	No effect
Lykoi Coat (Variant 2)	HR	hr ^{VA}	0	No effect
Hairlessness (Discovered in the Sphynx)	KRT71	re ^{hr}	0	No effect
Rexing (Discovered in the Devon Rex)	KRT71	re ^{dr}	0	No effect
Rexing (Discovered in the Cornish Rex and German Rex)	LPAR6	r	0	No effect
Glitter	Pending	gl	0	No effect

Tail Length

	Gene	Variant	Copies	Result
Short Tail (Variant 3)	HES7	jb	0	No effect
Short Tail (Variant 1)	T	C1199del	0	No effect
Short Tail (Variant 2)	T	T988del	0	No effect

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Extra Toes

	Gene	Variant	Copies	Result
Polydactyly (Variant 1)	LIMBR1	HW	0	No effect
Polydactyly (Variant 2)	LIMBR1	UK1	0	No effect
Polydactyly (Variant 3)	LIMBR1	UK2	0	No effect

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Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Acute Intermittent Porphyria (Variant 1)	HMBS	Deletion	0	AD	Clear
Acute Intermittent Porphyria (Variant 2)	HMBS	G>A	0	AD	Clear
Acute Intermittent Porphyria (Variant 3)	HMBS	Insertion	0	AD	Clear
Acute Intermittent Porphyria (Variant 4)	HMBS	Deletion	0	AD	Clear
Acute Intermittent Porphyria (Variant 5)	HMBS	G>A	0	AR	Clear
Autoimmune Lymphoproliferative Syndrome (Discovered in British Shorthair)	FASL	Insertion	0	AR	Clear
Burmese Head Defect (Discovered in the Burmese)	ALX1	Deletion	0	AD	Clear
Chediak-Higashi Syndrome (Discovered in the Persian)	LYST	Insertion	0	AR	Clear
Congenital Adrenal Hyperplasia	CYP11B1	G>A	0	AR	Clear
Congenital Erythropoietic Porphyria	UROS	G>A	0	AR	Clear
Congenital Myasthenic Syndrome (Discovered in the Devon Rex and Sphynx)	COLQ	G>A	0	AR	Clear
Cystinuria Type 1A	SCL3A1	C>T	0	AR	Clear
Cystinuria Type B (Variant 1)	SCL7A9	C>T	0	AR	Clear
Cystinuria Type B (Variant 2)	SCL7A9	G>A	0	AR	Clear
Dihydropyrimidinase Deficiency	DPYS	G>A	0	AR	Clear
Earfold and Osteochondrodysplasia (Discovered in the Scottish Fold)	TRPV4	G>T	0	AD	Clear
Familial Episodic Hypokalemic Polymyopathy (Discovered in the Burmese)	WNK4	C>T	0	AR	Clear
Glutaric Aciduria Type II	ETFDH	T>G	0	AR	Clear
Glycogen Storage Disease (Discovered in the Norwegian Forest Cat)	GBE1	Insertion	0	AR	Clear
GM1 Gangliosidosis	GLB1	G>C	0	AR	Clear

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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
GM2 Gangliosidosis	GM2A	Deletion	0	AR	Clear
GM2 Gangliosidosis Type II (Discovered in Domestic Shorthair cats)	HEXB	Insertion	0	AR	Clear
GM2 Gangliosidosis Type II (Discovered in Japanese domestic cats)	HEXB	C>T	0	AR	Clear
GM2 Gangliosidosis Type II (Discovered in the Burmese)	HEXB	Deletion	0	AR	Clear
Hemophilia B (Variant 1)	F9	C>T	0	XR	Clear
Hemophilia B (Variant 2)	F9	G>A	0	XR	Clear
Hyperoxaluria Type II	GRHPR	G>A	0	AR	Clear
Hypertrophic Cardiomyopathy (Discovered in the Ragdoll)	MYBPC	C>T	0	AD	Clear
Hypotrichosis (Discovered in the Birman)	FOXN1	Deletion	0	AR	Clear
Lipoprotein Lipase Deficiency	LPL	G>A	0	AR	Clear
Mucopolysaccharidosis Type I	IDUA	Deletion	0	AR	Clear
Mucopolysaccharidosis Type VI	ARSB	T>C	0	AR	Clear
Mucopolysaccharidosis Type VI Modifier	ARSB	G>A	0	MO	Clear
Mucopolysaccharidosis Type VII (Variant 1)	GUSB	G>A	0	AR	Clear
Mucopolysaccharidosis Type VII (Variant 2)	USB	C>T	0	AR	Clear
Myotonia Congenita	CLCN1	G>T	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Abyssinian)	CEP290	T>G	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Bengal)	KIF3B	G>A	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Persian)	AIPL1	C>T	0	AR	Clear
Sphingomyelinosis (Variant 1)	NPC1	G>C	0	AR	Clear
Sphingomyelinosis (Variant 2)	NPC2	G>A	0	AR	Clear

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Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Vitamin D-Dependent Rickets	CYP27B1	G>T	0	AR	Clear

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ID kit: FLNGQWH

Glossary of genetic terms

Test result definitions

At Risk: Based on the disorder's mode of inheritance, the cat inherited a number of genetic variant(s) which increases the cat's risk of being diagnosed with the associated disorder.

Carrier: The cat inherited one copy of a genetic variant when two copies are usually necessary to increase the cat's risk of being diagnosed with the associated disorder. While carriers are usually not at risk of clinical expression of the disorder, carriers of some complex variants may be associated with a low risk of developing the disorder.

Notable: Inheriting two copies of the genetic variant is noteworthy for specific aspects of health and breeding of the cat, but the cat should otherwise not suffer disease due to this genetic cause when in absence of other genetic variants.

Clear: The cat did not inherit the genetic variant(s) associated with the disorder and will not be at elevated risk of being diagnosed with the disorder due to this genotype. However, similar clinical signs could develop from different genetic or clinical causes.

Inconclusive: An inconclusive result indicates a confident call could not be made based on the data for that genetic variant. Health testing is performed in replicates, and on occasion the outcomes do not agree. This may occur due to an unusual sequence of DNA in the region tested, multiple cell genotypes present due to chimerism or acquired mutations, or due to quality of the DNA sample.

Inheritance mode definitions

Autosomal Recessive (AR): For autosomal recessive disorders, cats with two copies of the genetic variant are at risk of developing the associated disorder. Cats with one copy of the variant are considered carriers and are usually not at risk of developing the disorder. However, carriers of some complex variants grouped in this category may be associated with a low risk of developing the disorder. Cats with one or two copies may pass the disorder-associated variant to their kittens if bred.

Autosomal Recessive, asymptomatic (ARa): For autosomal recessive, asymptomatic disorders, cats with two copies of the variant can exhibit certain aspects of the variant-associated disorder but otherwise, they should not suffer clinical disease as typically expected with autosomal recessive disorders. Cats with one copy of the variant are called carriers and should not exhibit any aspect of the disorder. However, cats with one or two copies may pass the disorder-associated variant to their kittens if bred.

Autosomal Dominant (AD): For autosomal dominant disorders, cats with one or two copies of the genetic variant are at risk of developing the associated disorder. Inheriting two copies of the variant may increase the risk of development of the disorder or cause the condition to be more severe. These cats may pass the disorder-associated variant to their kittens if bred.

X-linked Recessive (XR): For X-linked recessive disorders, the genetic variant is found on the X chromosome. Female cats must inherit two copies of the variant to be at risk of developing the condition, whereas male cats only need one copy to be at risk. Males and females with any copies of the variant may pass the disorder-associated variant to their kittens if bred.

Modifier (MO): Genetic modifiers do not cause disease on their own but can cause disease or change the onset or severity of a disorder when combined with another disorder-associated variant. For some modifier variants only one copy is required to cause an effect, for others two copies are required. Please refer to the associated variant's breeder recommendations regarding safe breeding practices for each modifier variant.