

Leo's Genetic Report

Summary Page

Breeds Group

Western 96.05%

Maine Coon Broadly	80.45%
Western Siberian	13.53%
Ragdoll	2.08%
Russian Blue	0%
Norwegian Forest Cat	0%
American Shorthair	0%
Abyssinian	0%
Turkish Van	0%
Turkish Angora	0%

Eastern 0%

Broadly Eastern	13.53%
Oriental	0%
Peterbald	0%
Burmese	0%
Birman	0%
Thai	0%

Persian 0%

Broadly Persian	13.53%
Persian	0%
Exotic Shorthair	0%
British Shorthair	0%
Himalayan	0%

Exotic 0%

Broadly Exotic	13.53%
Bengal	0%
Savannah	0%
Egyptian Mau	0%

Polycat 3.95%

A domestic polycat is a remarkable result of many generations of mixed breeding between different types of cats, which is why the ancestry and origin of these kitties can be very difficult to determine.



NAME

Leo

AGE:

less than 1

GENDER:

male

REPORT DATE:

1-1-22

PARENT NAME:

John Smith

PRIMARY VET

HOSPITAL: My Vet Clinic

Oral Health Report



LEO'S RISK OF PERIODONTAL DISEASE

Periodontal disease affects the tissues surrounding the teeth. Initial stages are classified as gingivitis, while advanced stages are known as periodontitis.

RISK:

HIGH

MEDIUM

LOW

Risk score

2.16

LEO'S RISK OF TOOTH RESORPTION

Tooth resorption, also known as feline odontoclastic resorptive lesion (FORL), is characterized by progressive dentin erosion.

RISK:

HIGH

MEDIUM

LOW

Risk score

0.8

LEO'S RISK OF BAD BREATH

When bad breath is a persistent problem for a cat, this could be indicative of more serious oral health or general health issues.

RISK:

HIGH

MEDIUM

LOW

Risk score

6.9

Health Markers

CLEAR

Your cat is negative for genetic markers associated with this number of diseases:

39

AT RISK

Your cat is positive for genetic markers that increase their risk for developing this number of diseases:

0

CARRIER

Your cat is not at risk, but carries genetic markers associated with this number of diseases:

2

AT HIGH RISK

Your cat's genotype puts them at high risk for developing this number of diseases:

1

LIKELY BLOOD TYPE:
(based on genotype)

A

Trait Markers

LIKELY TO HAVE

2

NOT LIKELY TO HAVE

18

CARRIER

1

Breed Analysis

We analyzed your cat's genome and compared it to the genomes of purebred cats in our reference panel. There are 21 popular purebred breeds represented in our panel, categorized into the four main breed groups. Below is the percentage breakdown of the genomic similarity that your cat shares with each purebred breed associated with a main breed group.



Western 96.05%

Despite its name, this breed group contains breeds with diverse geographic origins, spanning Turkey, Russia, Europe, and the Americas.

80.45%	13.53%	2.08%	0%	0%	0%	0%	0%	0%	0%
Maine Coon	Broadly Western	Siberian	Ragdoll	Russian Blue	Norwegian Forest Cat	American Shorthair	Abyssinian	Turkish Van	Turkish Angora

Eastern 0%

Eastern breeds form their own tight, genetically distinct group. The origins of the breeds in this group can be traced back to Asian countries, with Southeast Asia playing a prominent role.

13.53%	0%	0%	0%	0%	0%
Broadly Eastern	Oriental	Peterbald	Burmese	Birman	Thai

Persian 0%

The Persian breed is among the oldest in the world. The Persian breed group contains breeds that are derived from the Persian breed.

13.53%	0%	0%	0%	0%
Broadly Persian	Persian	Exotic Shorthair	British Shorthair	Himalayan

Exotic 0%

The Exotic breed group contains Egyptian Mau and hybrid cats, which are the result of crossing domestic cats with certain wildcats. These cats often have a high degree of genetic separation from other breeds.

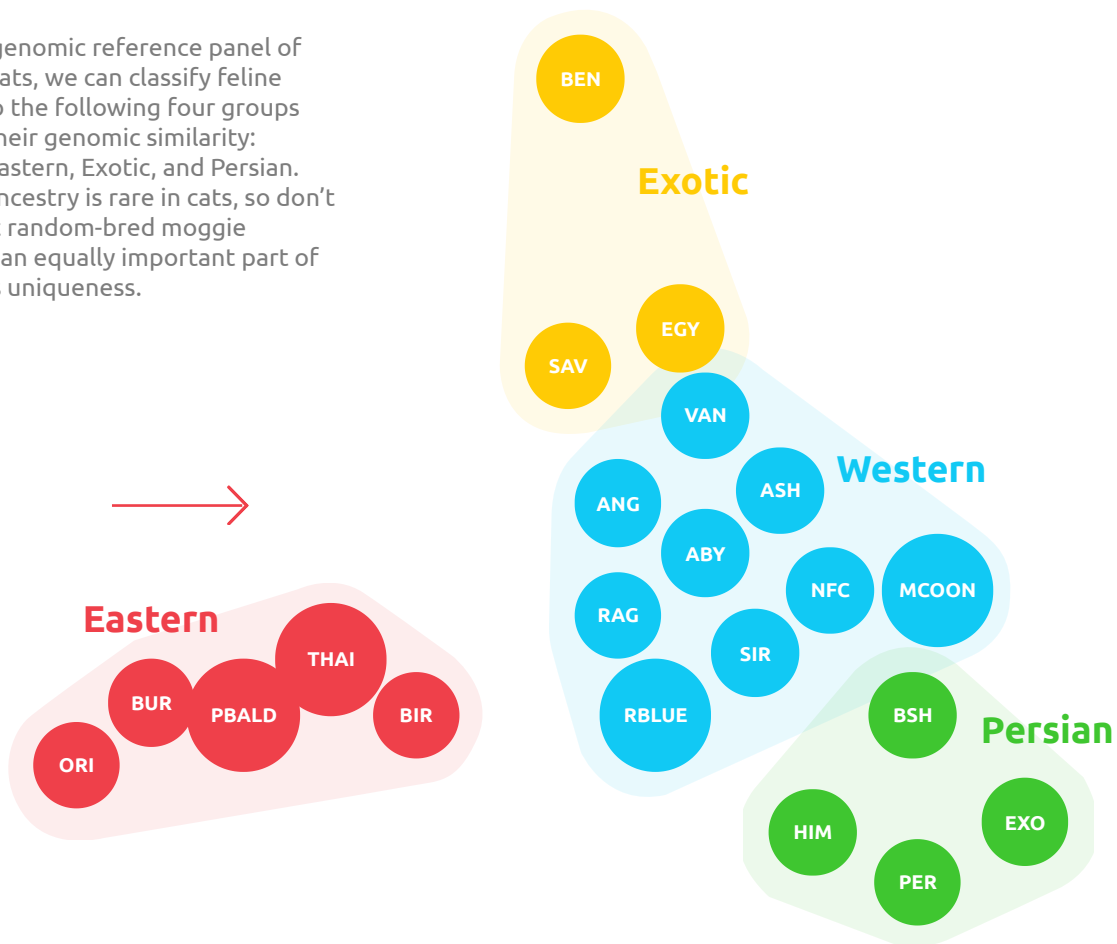
13.53%	0%	0%	0%
Broadly Exotic	Bengal	Savannah	Egyptian Mau

Domestic Polycat 3.95%

A domestic polycat is the remarkable result of many generations of mixed breeding (the opposite of selective breeding) between different types of cats, which is why the ancestry and origin of these kitties can be very difficult to determine.

Feline Breeds

Using our genomic reference panel of purebred cats, we can classify feline breeds into the following four groups based on their genomic similarity: Western, Eastern, Exotic, and Persian. Pedigree ancestry is rare in cats, so don't forget that random-bred moggie heritage is an equally important part of your kitty's uniqueness.



Western

Russian Blue	Ragdoll	American Shorthair	Siberian	Norwegian Forest Cat	Maine Coon	Abyssinian	Turkish Van	Turkish Angora
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Eastern

Oriental Shorthair	Peterbald	Burmese	Birman	Siamese
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Persian

Persian	Exotic Shorthair	British Shorthair	Himalayan
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Exotic

Savannah	Bengal	Egyptian Mau
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Leo's genetic health and trait markers

Kitty Genetics 101: Critical Reading

Genotype is the portion of your cat's genome that encodes the physical expression (phenotype) of a particular trait, such as eye color, coat color, or disease predisposition. Genes comprise the genotype.

Every gene in your kitty's genome is present in two copies—one inherited from each parent. These two copies can be the same or different. If they are different, we say that the cat has two different alleles (gene variants) and is heterozygous for this gene. Whereas if the two copies of the gene are the same, the cat is homozygous for the gene of interest.

How Does It All Relate To Disease?

Genetic disorders are conditions that can be present at birth or develop later in life and are caused by one or more genetic mutations.

We tested your cat for genetic health markers associated with genetic disorders. Genetic health markers are mutated gene alleles that are associated with an increased likelihood of developing a particular genetic disorder. With the exception of hypertrophic cardiomyopathy (which has more complex genetics) and Haemophilia B (which has an X-linked inheritance pattern), the conditions included in this report have either a dominant or a recessive pattern of inheritance. Diseases with a dominant inheritance pattern means that having only one mutated gene allele will result in the cat developing the disease. For diseases with a recessive inheritance pattern, the cat will develop the disease only if it has two mutated alleles. Having only one mutated recessive allele makes the cat a disease "carrier", meaning that it will not develop the disease but can pass the mutation to its kittens.

When it comes to the phenotype, alleles can have different contributions. If the two gene alleles are the same, then they will both contribute equally to the phenotype.

If the two alleles are different, then it is the nature of their relationship that determines which allele will contribute to the phenotype. Some alleles are dominant, meaning that they can "hide" other alleles and become the sole contributor to the phenotype. In contrast, the allele that is "hidden" in a heterozygous state is known as a recessive allele. A recessive allele can only contribute to the phenotype when your cat is homozygous for that allele, in other words, there is no dominant allele that can hijack the phenotype expression.

This section of the report details your kitty's results for 115 genetic markers that are associated with 43 diseases. 'Clear' status indicates that your cat tested negative for all genetic markers associated with a disease. 'Carrier' status means that your cat has one copy of a recessive genetic marker, or that your female cat has one copy of a marker with X-linked inheritance. In either case, this should only be a concern if you plan to breed your cat. 'At risk' and 'At high risk' status means one of three things: (1) your cat has tested positive for a dominant allelic mutation, (2) your cat has two copies of a recessive allelic mutation, or (3) your male cat has one copy of a marker with X-linked inheritance.

If you see either "At risk" or "At high risk", contact your veterinarian.

Please note that results should not replace a professional evaluation and clinical diagnosis by a veterinarian. It is also important to understand that a 'Clear' result does not mean that your cat is guaranteed to not develop the disease. It simply means that your cat is negative for the particular mutation(s) that we tested them for. There may be environmental factors and other genetic mutations that are not yet known (i.e., they are not yet included in our test) which could possibly contribute to developing the disease.

Scientific evidence strength rating system

The genetic markers in this report represent a deep dive into feline genetics research, including the very latest findings. Some research findings are scientifically stronger than others. For example, a study on 100 cats has stronger statistical power, and its conclusions can be interpreted with greater confidence, than a study on fewer than five cats. The report includes a five-star rating system that denotes the strength of the scientific research evidence that is available for each genetic marker, and the interpretation of associated results. This system answers the question: 'How strong is the evidence that this marker is associated with this disease or trait?'



Consistent results reported across multiple scientific studies. More than 100 cats across studies confirmed to have the expected correlation between the genetic marker and physical presentation. Multiple scientific tools used to confirm the mechanism behind the mutation. No contradictory results reported in literature.



Results based on one or more studies reporting findings from at least 30 cats, confirming the expected correlation between the genetic marker and physical presentation. Multiple scientific tools used to confirm the mechanism behind the mutation. No contradictory results reported in literature.



Results based on one or more studies reporting findings from at least 10 cats, confirming the expected correlation between the genetic marker and physical presentation. Multiple scientific tools may or may not have been used to confirm the mechanism behind the mutation. No contradictory results reported in literature.



Results based on a study that reports findings from fewer than 10 cats, confirming the expected correlation between the genetic marker and physical presentation. Multiple scientific tools may or may not have been used to confirm the mechanism behind the mutation. Potentially inconclusive inheritance pattern. No contradictory results reported in literature.



Preliminary results to be interpreted with extreme caution. Based on a scientific study that either focuses on a single cat or on a few studies that report contradictory results.

Health Summary

In this section, you will find a brief description of the genetic diseases for which your cat currently has results. Each of these diseases is represented by at least one known health marker. We also provide details on the genes and genetic mutations included in our test, with our five-star scientific evidence grading system for each marker. This grading system assesses the strength of the evidence that links each marker to each disease, based on the amount and quality of scientific literature available.

IMPORTANT: Results for some of the 115 health markers we currently test for may not be available in your first report. Your cat's results for these missing markers may be added over the coming weeks or months. Check results often for potential updates on missing markers.

It's important to share this information with your veterinarian so it can be added to your pet's medical records. If you are a veterinarian, please email vet@canix.com or visit basepaws.com/vets.

Clear

The cat is negative (has zero copies) for all of the markers for which we tested that are known to be associated with a particular disease. This result, however, should not rule out the need to seek a professional diagnosis by a veterinarian, should the cat develop symptoms of the disease. It is still possible that the cat is positive for markers yet to be discovered that could be associated with the disease, or in some cases, environmental factors could contribute to a cat's potential to develop the disease.

Carrier

In most cases, this means that the cat has one copy of an autosomal recessive disease-associated marker (mutated gene allele). As a refresher from the health markers intro section, for diseases with a recessive inheritance pattern, the cat will develop the disease only if it has two mutated alleles (two copies of the marker). Alternatively, the 'Carrier' result may mean that a female cat has one copy of a marker associated with the disease that follows an X-linked inheritance pattern. As a carrier, your cat is not at risk for developing the disease, but its offspring may be at risk.

At Risk

The cat has one copy of a marker for a disease where there is a dosage component to disease presentation (e.g., Hypertrophic Cardiomyopathy, Mucopolysaccharidosis VI, Hyperlipoproteinaemia). Gene dosage refers to the number of copies of an allele present in a cat's genome (entire set of genetic instructions stored in their chromosomes). A higher gene dosage results in an increased expression of a disease. This means that having one copy of the disease-associated marker could result in a milder disease presentation, as compared to having two copies where the disease presentation may be more severe.

At High Risk

This designation indicates that there is a very strong chance that the cat will develop the disease. It can mean one of four things: (1) the cat has one or two copies of a marker associated with an autosomal dominant disease (a single copy is enough to cause the disease); (2) the cat has two copies of a marker associated with an autosomal recessive disease; (3) the cat has two copies of a marker associated with a disease, where having these two copies results in a more severe disease presentation than if there was only one copy (the gene dosage effect); or (4) a male cat has one copy of a disease with an X-linked (sex-linked and therefore non-autosomal) inheritance pattern, meaning they are affected because they only have a single copy of the X chromosome that carries the mutation.

Eye disorders

Glaucoma

Primary congenital glaucoma is an autosomal recessive disease characterized by high fluid pressure in the eye, which damages the optic nerve and ultimately leads to blindness. Early diagnosis and treatment may slow the progression of the disease.

Gene

LTBP2

Mutation

★★★★★
ins(CTC..)

Seen in

Siamese

Status

0 copies

Clear

Progressive Retinal Atrophy (KIF3B-related)

This progressive retinal atrophy is related to Leber congenital amaurosis—a retinal disease causing severe visual impairment at birth. The disorder is inherited in an autosomal recessive manner, and is associated with the defective functioning of a protein called aryl hydrocarbon receptor-interacting protein-like 1 (AIPL1).

Gene

KIF3B

Mutation

★★★★★
C>T

Seen in

Bengal

Status

0 copies

Clear

Progressive Retinal Atrophy (AIPL1-related)

This progressive retinal atrophy is related to Leber congenital amaurosis—a retinal disease causing severe visual impairment at birth. The disorder is inherited in an autosomal recessive manner, and is associated with the defective functioning of a protein called aryl hydrocarbon receptor-interacting protein-like 1 (AIPL1).

Gene

AIPL1

Mutation

★★★★★
C>T

Seen in

Persian

Status

0 copies

Clear

Late-Onset Photoreceptor Degeneration (rdAc)

This late-onset type of progressive retinal atrophy (PRA) is caused by an autosomal recessive mutation "rdAc" in the CEP290 gene. The mutation causes progressive degeneration of the retina, slowly leading to vision loss and blindness.

Gene

CEP290

Mutation

★★★★★
A>C

Seen in

Abyssinian,
Somali

Status

0 copies

Clear

Autoimmune disorders

Leukocyte Adhesion Deficiency

Leukocyte adhesion deficiency (LAD) is an immunodeficiency disorder associated with poorly-functioning neutrophils. Consequently, affected cats are highly susceptible to recurrent infections. LAD is a serious disorder, and if left untreated, affected kittens rarely reach adulthood.

Gene

ITGB2

Mutation

★☆☆☆☆
del(GCC..)

Seen in

None

Status

0 copies

Clear

Autoimmune Lymphoproliferative Syndrome

Autoimmune Lymphoproliferative Syndrome (ALPS) is a lethal disease, distinguished by massive enlargement of lymphatic nodes and the spleen, caused by the accumulation of lymphocytes.

Gene

FASLG

Mutation

★☆☆☆☆
ins(A)

Seen in

British
Shorthair

Status

0 copies

Clear

Metabolic disorders

Porphyria, acute intermittent (AIP)

Porphyria is a group of diseases associated with the accumulation of porphyrins. There are two types of porphyria described: acute and congenital. The acute type primarily affects the nervous system. Clinical presentation, severity and prognosis vary.

Gene	Mutation	Seen in	Status
HMBS	★☆☆☆☆ C>T	None	0 copies
HMBS	★☆☆☆☆ G>A	None	0 copies
HMBS	★☆☆☆☆ ins(T)	None	0 copies
HMBS	★☆☆☆☆ del(GAG)	None	0 copies
HMBS	★☆☆☆☆ G>A	None	0 copies
HMBS	★☆☆☆☆ del(ACA..)	None	0 copies

Clear

Mucopolysaccharidosis VII

Mucopolysaccharidosis is a metabolic deficiency in the production or functioning of the enzymes required for digestion of glycosaminoglycans (GAGs). Consequently, GAGs start accumulating in the cells, causing progressive cellular damage. The prognosis and life expectancy are variable.

Gene	Mutation	Seen in	Status
GUSB	★☆☆☆☆ T>G	None	1 copies
GUSB	★☆☆☆☆ C>T	None	1 copies
GUSB	★☆☆☆☆ G>A	None	0 copies

At High Risk

Niemann-Pick disease, type A

Niemann-Pick disease is a group of autosomal recessive disorders characterized by deficiencies of specific enzymes that are involved in the intracellular transport of lipids. This leads to accumulation of cholesterol in lysosomes, causing neurological deterioration.

Gene	Mutation	Seen in	Status
SMPD1	★☆☆☆☆ G>A	None	0 copies

Clear

Metabolic disorders

Mucopolysaccharidosis I

Mucopolysaccharidosis is a metabolic deficiency in the production or functioning of the enzymes required for digestion of glycosaminoglycans (GAGs). Consequently, GAGs start accumulating in the cells, causing progressive cellular damage. The prognosis and life expectancy are variable.

Gene	Mutation	Seen in	Status
IDUA	★★★★★ del(TCG)	None	0 copies
IDUA	★★★★★ del(GTC)	None	0 copies

[Clear](#)

Wilson's Disease

Wilson's Disease is a rare autosomal recessive genetic disorder associated with accumulation of copper in the vital organs, such as the liver and the brain. If left untreated, this can be a lethal disease. Affected cats may show signs of jaundice, lethargy, weight loss, hyperammonemia, and elevated hepatic enzymes.

Gene	Mutation	Seen in	Status
ATP7B	★★★★★ C>G	None	0 copies

[Clear](#)

Alpha-mannosidosis

Alpha-mannosidosis is a lysosomal storage disorder characterized by the deficiency of the alpha-D-mannosidase enzyme and the progressive accumulation of oligosaccharides in all tissues, consequently leading to cell death. Treatment options are scarce.

Gene	Mutation	Seen in	Status
MAN2B1	★★★★★ del(CTG..)	Persian	0 copies

[Clear](#)

Dihydropyrimidinuria

Dihydropyrimidinuria is a rare disease associated with a deficiency of the enzyme dihydropyrimidinase. Cats with this disease can suffer from weight loss, vomiting, and lethargy.

Gene	Mutation	Seen in	Status
DPYS	★★★★★ C>T	None	0 copies

[Clear](#)

Metabolic disorders

Mucopolysaccharidosis VI

Mucopolysaccharidosis is a metabolic deficiency in the production or functioning of the enzymes required for digestion of glycosaminoglycans (GAGs). Consequently, GAGs start accumulating in the cells, causing progressive cellular damage. The prognosis and life expectancy are variable.

Gene	Mutation	Seen in	Status
ARSB	★★★★★ A>G	Birman, Siamese	0 copies
ARSB	★★★★★ C>T	Birman, Siamese	0 copies

Clear

Niemann-Pick disease, type C1

Niemann-Pick disease is a group of autosomal recessive disorders characterized by deficiencies of specific enzymes that are involved in the intracellular transport of lipids. This leads to accumulation of cholesterol in lysosomes, causing neurological deterioration.

Gene	Mutation	Seen in	Status
NPC1	★★★★★ T>G	None	0 copies
NPC1	★★★★★ C>G	None	0 copies

Clear

Pyruvate Kinase Deficiency

Pyruvate kinase (PK) deficiency is an inherited metabolic disorder characterized by disrupted survival of erythrocytes, leading to anemia, lethargy, weakness, weight loss, abdominal enlargement, and in some cases, jaundice, tachycardia, and muscle wasting. The prognosis and severity are variable.

Gene	Mutation	Seen in	Status
PKLR	★★★★★ G>A	Somali, Abyssinian	0 copies

Clear

Gangliosidosis, type GM2 (variant AB)

Gangliosidosis is a group of lipid storage disorders characterized by the accumulation of gangliosides in neurons. This eventually leads to progressive destruction of the nerve cells in the brain and spinal cord. The disease is not treatable, and typically leads to death within the first year of life.

Gene	Mutation	Seen in	Status
GM2A	★★★★★ del(GAC..)	None	0 copies

Clear

Metabolic disorders

Gangliosidosis, type GM2 (variant 0)

Gangliosidosis is a group of lipid storage disorders characterized by the accumulation of gangliosides in neurons. This eventually leads to progressive destruction of the nerve cells in the brain and spinal cord. The disease is not treatable, and typically leads to death within the first year of life.

Gene	Mutation	Seen in	Status
HEXB	★★★★★ inv(TAC..)	Burmese, Korat	0 copies
HEXB	★★★★★ del(TAA..)	Burmese, Korat	0 copies
HEXB	★★★★★ del(C)	Burmese, Korat	0 copies
HEXB	★★★★★ C>T	Burmese, Korat	0 copies

[Clear](#)

Porphyria, congenital erythropoietic (CEP)

Porphyria is a group of diseases associated with the accumulation of porphyrins. There are two types of porphyria described: acute and congenital. The acute type primarily affects the nervous system. Clinical presentation, severity and prognosis vary.

Gene	Mutation	Seen in	Status
UROS	★★★★★ G>A	None	0 copies
UROS	★★★★★ C>T	None	0 copies

[Clear](#)

Hyperlipoproteinaemia

Hyperlipoproteinemia is a disease characterized by a deficiency in the digestion of lipids and lipoproteins, mainly cholesterol and triglycerides. Their persistently elevated levels in the blood pose multiple health threats, such as heart disease. Timely diagnosis improves prognosis.

Gene	Mutation	Seen in	Status
LPL	★★★★★ C>T	None	0 copies

[Clear](#)

Gangliosidosis, type GM1

Gangliosidosis is a group of lipid storage disorders characterized by the accumulation of gangliosides in neurons. This eventually leads to progressive destruction of the nerve cells in the brain and spinal cord. The disease is not treatable, and typically leads to death within the first year of life.

Gene	Mutation	Seen in	Status
GLB1	★★★★★ C>G	None	0 copies

[Clear](#)

Metabolic disorders

Niemann-Pick disease, type C2

Niemann-Pick disease is a group of autosomal recessive disorders characterized by deficiencies of specific enzymes that are involved in the intracellular transport of lipids. This leads to accumulation of cholesterol in lysosomes, causing neurological deterioration.

Gene

NPC2

Mutation

★☆☆☆☆
C>T

Seen in

None

Status

0 copies

Clear

Endocrine disorders

Congenital Adrenal Hyperplasia

Congenital adrenal hyperplasia is an autosomal recessive disease characterized by an imbalance in the production of the adrenal hormones cortisol and aldosterone. The severity of the condition depends on the nature of the deficiency, and common presentations include altered development of primary and secondary sex characteristics.

Gene
CYP11B1

Mutation
★☆☆☆☆
G>A

Seen in
None

Status
0 copies

Clear

Hypogonadotropic Hypogonadism

Hypogonadotropic hypogonadism is associated with a gonadotropic releasing hormone (GnRH) deficiency. The presentation of the disease depends on the severity, and affected cats typically suffer from hindered sexual development.

Gene
TAC3

Mutation
★☆☆☆☆
C>T

Seen in
None

Status
0 copies

Clear

Hypothyroidism

Hypothyroidism in cats is a rare and complex glandular disorder caused by an under-active thyroid gland. As a result, thyroid hormones triiodothyronine (T3) and thyroxine (T4) are under-produced. If diagnosed correctly and as early as possible, the prognosis and management of the disease can be promising.

Gene
TPO

Mutation
★☆☆☆☆
C>T

Seen in
None

Status
0 copies

Clear

Renal disorders

Cystinuria, type B

Cystinuria is an inherited metabolic disease characterized by defective amino acid reabsorption, leading to the formation of cystine stones in the kidneys, ureter and bladder, and eventually, to urinary obstruction. There are no available treatments for this condition.

Gene	Mutation	Seen in	Status
SLC7A9	★★★★★ T>A	None	0 copies
SLC7A9	★★★★★ A>G	None	0 copies
SLC7A9	★★★★★ G>A	None	0 copies
SLC7A9	★★★★★ C>T	None	0 copies
SLC7A9	★★★★★ G>A	None	0 copies

Clear

Cystinuria, type IA

Cystinuria is an inherited metabolic disease characterized by defective amino acid reabsorption, leading to the formation of cystine stones in the kidneys, ureter and bladder, and eventually, to urinary obstruction. There are no available treatments for this condition.

Gene	Mutation	Seen in	Status
SLC3A1	★★★★★ C>T	None	0 copies

Clear

Polycystic Kidney Disease

Polycystic kidney disease (PKD) is an autosomal dominant genetic disease characterized by the formation of small fluid-filled cysts in the kidneys, which leads to kidney damage and failure. The disease is progressive and irreversible, but early diagnosis can significantly improve prognosis.

Gene	Mutation	Seen in	Status
PKD1	★★★★★ C>A	Persian	0 copies

Clear

Primary Hyperoxaluria

Primary hyperoxaluria is a severe, autosomal recessive disorder associated with an elevated deposition and excretion of oxalates. This leads to formation of renal and bladder oxalate stones, and eventually, kidney damage and failure. Therapy is restricted to the treatment of symptoms.

Gene	Mutation	Seen in	Status
GRHPR	★★★★★ G>A	None	0 copies

Clear

Musculoskeletal and connective tissue disorders

Myotonia Congenita

Myotonia Congenita (MC) is a hereditary neuromuscular disorder characterized by persistent muscle contraction (or delayed relaxation of muscles). The overall prognosis of the disease is poor, and treatment options are scarce.

Gene	Mutation	Seen in	Status
CLCN1	★★★★★ G>T	None	0 copies

Clear

Vitamin D-deficiency rickets, type IA

Vitamin D-dependent rickets is a disease associated with the softening of bones and an increased rate of bone deformities and fractures.

Gene	Mutation	Seen in	Status
CYP27B1	★★★★★ del(C)	None	0 copies
CYP27B1	★★★★★ C>A	None	0 copies

Clear

Hypokalemic Periodic Paralysis

Hypokalemic periodic paralysis is a genetic disease of the Burmese and closely related cat breeds that is characterized by low potassium ion (K+) levels in the blood. The condition is marked by either generalized or localized skeletal muscle weakness, often episodic in nature.

Gene	Mutation	Seen in	Status
WNK4	★★★★★ C>T	Burmese	0 copies

Clear

Ehlers-Danlos Syndrome

Ehlers-Danlos syndrome is a genetic disorder of the connective tissue that is associated with defective formation of collagen. The disease causes abnormally extensible skin, hindered wound healing, and hypermobility of joints. Treatment is focused on relieving symptoms and preventing complications.

Gene	Mutation	Seen in	Status
COL5A1	★★★★★ del(C)	None	0 copies

Clear

Musculoskeletal and connective tissue disorders

Fibrodysplasia Ossificans

Fibrodysplasia ossificans is a connective tissue disease that results in severe disability. This condition causes muscles, filaments, and tendons to ossify (turn into bony tissues), either spontaneously or post-injury. Due to the formation of a secondary skeleton in place of damaged muscle tissues, the cat's ability to move is progressively hindered.

Gene

ACVR1

Mutation

★☆☆☆☆
C>T

Seen in

None

Status

0 copies

Clear

Vitamin D-deficiency rickets, type IB

Vitamin D-dependent rickets is a disease associated with the softening of bones and an increased rate of bone deformities and fractures.

Gene

CYP2R1

Mutation

★☆☆☆☆
del(A)

Seen in

None

Status

0 copies

Clear

Blood disorders

Methemoglobinemia

Methemoglobinemia is a disease characterized by elevated levels of methemoglobin in the blood. The disease presentation is variable, but usually includes brownish discoloration of mucous membranes and blood with a brownish hue. Complications can involve arrhythmias and seizures.

Gene	Mutation	Seen in	Status
CYB5R3	★☆☆☆☆ C>G	None	0 copies
CYB5R3	★☆☆☆☆ C>T	None	0 copies

Clear

Factor XII Deficiency

Factor XII deficiency, or Hageman deficiency, is a blood clotting disorder characterized by deficiency in the coagulation factor XII. Luckily, unlike other types of haemophilias, this condition is not severe and prognosis is typically very good.

Gene	Mutation	Seen in	Status
F12	★★★★★ del(C)	None	0 copies
F12	★★★★★ G>C	None	1 copies

Carrier

Glanzmann Thrombasthenia

Glanzmann thrombasthenia is a rare genetic coagulation disorder. The disease is caused by a mutation in an integrin gene, resulting in the inability of thrombocytes to clot blood. The severity and presentation of the disease are variable.

Gene	Mutation	Seen in	Status
ITGA2B	★★★★★ del(G)	None	0 copies

Clear

Haemophilia B

Haemophilia B, also known as Christmas disease, is an X chromosome-linked recessive deficiency of coagulation factor IX. Like other haemophilias, the disorder is characterized by dysfunctional blood coagulation in a case of injury. The prognosis and treatment depend on the severity of the disease.

Gene	Mutation	Seen in	Status
F9	★★★★★ G>A	None	0 copies
F9	★★★★★ C>T	None	0 copies

Clear

Cardiovascular disorders

Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is the most common feline heart disease, and it is characterized by tachycardia and the thickening of the heart's muscular walls. The severity of the disease is variable, but if diagnosed early, different treatment options are available.

Gene	Mutation	Seen in	Status
TNNT2	★★★★★ C>T	Maine Coon	0 copies
MYBPC3	★★★★★ G>A	Maine Coon	0 copies
MYH7	★★★★★ C>T	None	0 copies
MYBPC3	★★★★★ C>G	Ragdoll	0 copies

Clear

Skin disorders

Epidermolysis Bullosa Simplex

Epidermolysis bullosa simplex (EBS) is a genetic disease associated with highly fragile skin and mucous membranes, due to mutations in genes associated with cytoskeletal cell functions. Common issues include blistering and wounding of the skin, ulcer formation on mucous membranes, and nail deformities in response to mild traumas.

Gene

KRT14

Mutation

★☆☆☆☆
G>A

Seen in

None

Status

0 copies

Clear

Inflammatory Linear Verrucous Epidermal Nevus

Inflammatory Linear Verrucous Epidermal Nevus (ILVEN) is an inherited disease characterized by skin overgrowth and the formation of pigmented, itchy cutaneous lesions. These wart-like lesions are called epidermal nevi, and affected areas are prone to inflammation. Due to the rarity of this disease in cats, there is no established treatment protocol.

Gene

NSDHL

Mutation

★☆☆☆☆
A>G

Seen in

None

Status

1 copies

Carrier

Blood type and transfusion risk

Only one blood type system has been established in domestic cats: the AB system. There are three feline blood types: A, B, and AB. There is a fourth blood type, MiK, that has also been identified, but not much is known other than the fact that it is very rare and occurs in less than 1 percent of cats.

The genetics behind feline blood types is rather complicated. There is scientific literature on seven different markers that may play a role in determining a cat's blood type. However, only four of them are considered the most reliable and consistent predictors and these four are the markers for which we screen in this test. They are all mutations in the Cytidine monophospho-N-acetylneuraminic acid hydroxylase (CMAH) gene.

IMPORTANT: Due to the imperfect relationship between genetics and blood type, we strongly recommend performing a serological blood typing test with your veterinarian, especially if blood type information is needed for making any medical decisions.

Blood Group A

Most cats of European or American descent have blood type A. This is considered the most prevalent feline blood type, with more than 70 percent of cats estimated to have blood type A.

Blood Group B

The prevalence of blood type B varies widely depending on the cat's pedigree. This blood type is rarely seen in Maine Coons and Norwegian Forest Cats, and more frequently seen in the British Shorthair and Exotic and rexoid breeds.

Blood Group AB

Blood type AB is extremely rare, with an estimated prevalence across pedigreed and non-pedigreed cats of less than 1 percent.

Likely blood type based on genotype

Gene	Mutation	Status
CMAH	★★★★★ G>T	0
CMAH	★★★★★ T>A	0
CMAH	★★★★★ C>T	0
CMAH	★★★★★ del(T)	0

Blood type A

Blood transfusion risk

It is recommended that the blood types of both the donor and the recipient are established before a transfusion. Cats with blood type A should only receive blood from cats of the same blood type. Transfusion from type B donors to type A recipients does not work efficiently and may be accompanied by mild incompatibility symptoms such as restlessness, tachycardia, and tachypnea.

Medium

Traits

This section presents your cat's results for a variety of genetic markers associated with physical traits that can be seen. Additionally, we screened your cat for an important trait that can't be seen with the naked eye: resistance to Feline Immunodeficiency Virus (FIV).

For many traits in this section, there are multiple known variants associated with each trait. We included our five-star scientific evidence grading system for each marker which assesses the strength of evidence linking each marker to each trait, based on the amount and quality of scientific literature available. In rare cases, it is possible that your cat is positive for a marker but does not exhibit the physical trait associated with it due to complex genetic or epigenetic interactions that may not be well understood. Epigenetic interactions include behavior and environment, which can cause changes that affect the way your cat's genes work but are reversible and do not change your cat's DNA sequence.

It is also possible that your cat does exhibit a physical trait but has tested negative for all known markers associated with the trait. This means is that, in your cat's case, the specific physical presentation may have different underlying genetics to what is currently known in the scientific literature. If this is the case, think of it as part of why your cat is so special and let us know (email us at meow@canix.com) about them, as they could potentially help contribute to the advancement of feline genetics research!

IMPORTANT: Results for some of the 50 trait markers we currently test for may not be available in your first report. Your cat's results for these missing markers may be added in the coming weeks or months. Check results often for potential updates on missing markers.

Carrier

The cat has one copy of a marker associated with a specific physical trait; however, it is unlikely to be physically exhibiting this trait. This could be because the trait has an autosomal recessive pattern of inheritance (needs two copies to present physically) or because the physical presentation of the trait is associated with a specific combination of markers, of which your cat only has one.

Likely to Have

The cat is positive for a marker (or markers) linked to a specific trait and is likely to exhibit this trait. This could be a result of the cat having one copy of a trait marker with an autosomal dominant pattern of inheritance, or the cat having two copies of a marker with an autosomal recessive pattern of inheritance. Alternatively, your cat could have the specific allelic series (combination of markers) that is likely to result in a specific trait.

Not Likely to Have

Based on the cat's genotype, it is unlikely that it is exhibiting this particular trait.

Coat color and pattern

Charcoal coat color

The charcoal colored coat seen in some Bengal cats is the product of two distinct alleles of the ASIP gene, inherited together. The first allele is the one associated with a solid black coat (melanism) in domestic cats, while the second allele is inherited directly from the Asian leopard cat.

Gene	Mutation	Seen in	Status
ASIP	★★★★★ C>G	Bengal	0 copies
ASIP	★★★★★ A>G	Bengal	0 copies
ASIP	★★★★★ A>T	Bengal	0 copies
ASIP	★★★★★ T>C	Bengal	0 copies
ASIP	★★★★★ T>C	Bengal	0 copies
ASIP	★★★★★ G>T	Bengal	0 copies
ASIP	★★★★★ C>T	Bengal	0 copies
ASIP	★★★★★ A>G	Bengal	0 copies
ASIP	★★★★★ del(CA)	Bengal	2 copies

Not Likely To Have

Amber coat color

The melanocortin 1 receptor gene (MC1R) is responsible for the deposition of pigment in hair. Recessive alleles of this gene produce bright red to yellow coat pigmentation. A mutation described in Norwegian Forest cats is associated with the "amber" red coat. These cats are born a different color, with amber shades developing over time.

Gene	Mutation	Seen in	Status
MC1R	★★★★★ G>A	Norwegian Forest Cat	0 copies

Not Likely To Have

Cinnamon coat color

Brown coat color variations are caused by various mutations in the gene coding for tyrosinase-related protein-1 (TYRP1). The cinnamon (light brown) coat color is a consequence of inheriting two mutated copies of the TYRP1 gene.

Gene	Mutation	Seen in	Status
TYRP1	★★★★★ C>T	None	0 copies

Not Likely To Have

Coat color and pattern

Russet coat color

The melanocortin 1 receptor gene (MC1R) is responsible for the deposition of pigment in hair. Recessive alleles of this gene produce bright red to yellow coat pigmentation. A mutation described in Burmese cats produces the "russet" red coat. These cats are born a different color, with red shades developing over time.

Gene	Mutation	Seen in	Status
MC1R	★★★★★ del(TCT)	Burmese	0 copies

Not Likely To Have

Blotched tabby coat color

The tabby coat is characterized by a mix of two features: (1) a light background component where individual hairs have light bands, and (2) a superimposed darker component where hairs have almost no banding. In blotched tabbies, the dark component is expanded into loosely-organized structures, forming wide whorls. The genetic determination of tabby coloration is complicated (and not fully understood), but different mutations in the gene LVRN play a key role in forming the blotched or mackerel tabby coat phenotype.

Gene	Mutation	Seen in	Status
LVRN	★★★★★ G>A	None	0 copies
LVRN	★★★★★ A>G	None	0 copies

Not Likely To Have

Copal coat color

The melanocortin 1 receptor gene (MC1R) is responsible for the deposition of pigment in hair. Recessive alleles of this gene produce bright red to yellow coat pigmentation. A mutation described in Burmese cats produces the "russet" red coat. These cats are born a different color, with red shades developing over time.

Gene	Mutation	Seen in	Status
MC1R	★★★★★ del(GCG..)	Kurilian Bobtail	0 copies

Not Likely To Have

Coat color and pattern

Siamese coat color

The Siamese 'colorpoint' coat is a form of albinism characterized by darker pigmentation at the extremities of the body (ears, tail, paws). This appearance is due to a temperature-sensitive mutation in the tyrosinase gene, causing pigment to be produced only at the cooler extremities of the body. This coat type is characteristic of Siamese, Birman and Himalayan cats.

Gene	Mutation	Seen in	Status
TYR	★☆☆☆☆ C>A	Siamese, Birman, Himalayan	0 copies
TYR	★☆☆☆☆ del(G)	Siamese, Birman, Himalayan	0 copies
TYR	★☆☆☆☆ del(G)	Siamese, Birman, Himalayan	0 copies
TYR	★☆☆☆☆ C>T	Siamese, Birman, Himalayan	2 copies
TYR	★☆☆☆☆ G>A	Siamese, Birman, Himalayan	0 copies
TYR	★☆☆☆☆ G>A	Siamese, Birman, Himalayan	0 copies

Likely To Have

Dilute coat color

The dilute coat color phenotype in cats is related to both the eumelanin and pheomelanin pigment pathways. When two copies of a mutation in the melanophilin gene (MLPH) are inherited, the pigment deposition in hair is affected and the original coat color is altered. For example, black fur becomes gray, and orange will turn cream.

Gene	Mutation	Seen in	Status
MLPH	★★★★★ del(T)	None	0 copies

Not Likely To Have

Chocolate coat color

Brown coat color variations are caused by various mutations in the gene coding for tyrosinase-related protein-1 (TYRP1). The chocolate coat color is a consequence of inheriting two mutated copies of the TYRP1 gene.

Gene	Mutation	Seen in	Status
TYRP1	★☆☆☆☆ C>G	None	1 copies
TYRP1	★☆☆☆☆ G>A	None	0 copies

Carrier

Coat color and pattern

Mackerel tabby coat color

The tabby coat is characterized by a mix of two features: (1) a light background component where individual hairs have light bands, and (2) a superimposed darker component where hairs have almost no banding. In mackerel tabbies, the coat has a striped appearance. The genetic determination of tabby coloration is complicated (and not fully understood), but different mutations in the gene LVRN play a key role in forming the tabby coat phenotype.

Gene

LVRN

Mutation

★☆☆☆☆
A>G

Seen in

None

Status

0 copies

Not Likely To Have

Albinism

Albinism is a phenotype characterized by a lack of pigmentation in hair (appearing as a solid white coat), skin, and eyes. It is associated with a recessive allele of the tyrosinase gene. Albino cats tend to have blue or pink eyes.

Gene

TYR

Mutation

★☆☆☆☆
del(G)

Seen in

None

Status

0 copies

Not Likely To Have

TYR

★☆☆☆☆
G>A

None

0 copies

Body morphology

Short tail

A variety of short tail phenotypes are associated with autosomal dominant mutations in the T gene. Some of the breeds among which these mutations can be found are Manx, American Bobtail, and Pixie-Bob.

Gene	Mutation	Seen in	Status
T	★★★★★ del(G)	Manx, American Bobtail, Pixie- bob	0 copies
T	★★★★★ del(G)	Manx, American Bobtail, Pixie- bob	0 copies
T	★★★★★ del(A)	Manx, American Bobtail, Pixie- bob	0 copies
T	★★★★★ GGC..>CTG..	Manx, American Bobtail, Pixie- bob	0 copies

Not Likely To Have

Osteochondrodysplasia

Folded ears are associated with osteochondrodysplasia, a genetic condition affecting cartilage tissues throughout the body. The phenotype is linked to an autosomal dominant allele of a gene named TRPV4. This mutation is specific to the Scottish Fold breed. Affected cats have ears folded forward and down, as well as different degrees of malformation in the distal forelimbs, distal hindlimbs, and tail, as well as progressive joint destruction.

Gene	Mutation	Seen in	Status
TRPV4	★★★★★ C>A	None	0 copies

Not Likely To Have

Short and kinked tail

A dominantly inherited mutation in a gene responsible for proper body patterning and segmentation, HES7, is the cause of the short tail phenotype characteristic of the Japanese Bobtail breed.

Gene	Mutation	Seen in	Status
HES7	★★★★★ A>G	Japanese Bobtail	0 copies

Not Likely To Have

Body morphology

Polydactyly

Cats have four toes and one dewclaw (thumb) on each front paw, and four toes on each hind paw. Polydactyly is a phenotype characterized by a higher number of toes, either on the outer or inner side of the paw. Any cat may be born polydactyl, but the trait seems to be most common among Maine Coons.

Gene	Mutation	Seen in	Status
ZRS	★★★★★ C>G	None	0 copies
ZRS	★★★★★ T>A	None	0 copies
ZRS	★★★★★ T>C	None	0 copies

Not Likely To Have

Coat texture

Lykoi coat

Lykoi (werewolf) cats have a characteristic form of partial hairlessness (hypotrichia), where there is a significant reduction in the average number of follicles per hair follicle group as compared to domestic shorthair cats. Fur on the limbs is even sparser. The phenotype is associated with a variety of mutations in the HR gene.

Gene	Mutation	Seen in	Status
HR	★★★★★ C>T	Lykoi	0 copies
HR	★★★★★ ins(GT)	Lykoi	0 copies
HR	★★★★★ G>A	Lykoi	0 copies
HR	★★★★★ ins(GAC..)	Lykoi	0 copies
HR	★★★★★ GT>GCA..	Lykoi	0 copies
HR	★★★★★ C>T	Lykoi	0 copies

Not Likely To Have

Hypotrichosis

An autosomal recessive allele of the FOXN1 gene is associated with an almost complete lack of hair (hypotrichosis) and a shortened life expectancy. Most kittens with two mutated FOXN1 gene copies die before their first birthday. This mutation was described in Birman cats.

Gene	Mutation	Seen in	Status
FOXN1	★★★★★ del(ACA..)	Birman	0 copies

Not Likely To Have

Curly coat

The genetic variations resulting in curly fur are called rex mutations. They can occur in a variety of genes, thus creating various types of curly coats in cats. These mutations cause changes in the hair structure, leading to the curly appearance.

Gene	Mutation	Seen in	Status
KRT71	★★★★★ C>G	Devon Rex	0 copies
KRT71	★★★★★ C>T	Devon Rex	0 copies
LPAR6	★★★★★ del(GTT..)	Cornish Rex, German Rex	0 copies

Not Likely To Have

Coat texture

Sphynx Coat

The Sphynx coat type is characterised by a lack of fur, with very fine hairs covering the body, particularly around the nose, tail, and toes. These hairs don't have a well-formed bulb, which makes them easily dislodged. The Sphynx coat is associated with an autosomal recessive allele of the KRT71 gene.

Gene

KRT71

Mutation

★★★★★
C>T

Seen in

Sphynx, Kohana

Status

0 copies

Not Likely To Have

Coat length

Long-haired coat

The long-haired coat can be a typical feature of Maine Coon, Persian, Ragdoll, and Somali cats, as well as of some mixed-breed cats. The long-haired coat phenotype is associated with various mutations in the fibroblast growth factor 5 (FGF5) gene.

Gene	Mutation	Seen in	Status
FGF5	★☆☆☆☆ A>T	Maine Coon, Persian, Ragdoll, Somali	0 copies
FGF5	★☆☆☆☆ G>T	Maine Coon, Persian, Ragdoll, Somali	2 copies
FGF5	★☆☆☆☆ G>A	Maine Coon, Persian, Ragdoll, Somali	0 copies
FGF5	★☆☆☆☆ ins(A)	Maine Coon, Persian, Ragdoll, Somali	0 copies

Likely To Have

Susceptibility to viral infection

Resistance to FIV

Feline Immunodeficiency Virus (FIV) is a lentivirus affecting from 2.5% to 4.4% cats worldwide, causing a disease similar to human AIDS. A variant of the APOBEC3Z3 gene was demonstrated to suppress the infectivity of FIV, thus making cats that carry this variant more likely to be resistant to infection.

Gene	Mutation	Seen in	Status
APOBEC3Z3	★☆☆☆☆ GC>AT	None	0 copies

Not Likely To Have