MaineMarie Guacamole's genetic health and trait markers

VERSION 4.0

basepaws

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@basepaws.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.

Metabolic disorders

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

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	del(C)	Burmese, Korat	0 copies
HEXB	C>T	Burmese, Korat	0 copies
HEXB	del(TAA..)	Burmese, Korat	0 copies
HEXB	inv(TAC..)	Burmese, Korat	0 copies

Clear

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	C>T	Birman, Siamese	0 copies
ARSB	A>G	Birman, Siamese	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

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	del(ACA..)	None	0 copies
HMBS	G>A	None	0 copies
HMBS	C>T	None	0 copies
HMBS	G>A	None	0 copies
HMBS	del(GAG)	None	0 copies

Clear

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

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	C>T	None	0 copies
UROS	G>A	None	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	C>G	None	0 copies
NPC1	T>G	None	0 copies

Clear

Metabolic disorders

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

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(GTC)	None	0 copies	Clear
IDUA	del(TCG)	None	0 copies	

Neuronal Ceroid Lipofuscinosis

Neuronal ceroid lipofuscinosis (NCL) is a neurodegenerative lysosomal storage disease. It is characterized by abnormal accumulation of lipopigments in the neurons, liver, spleen, kidneys, and heart. NCL is associated with progressive blindness and neurologic deficits.

Gene	Mutation	Seen in	Status	
CLN6	G>A	None	0 copies	Clear

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	Mutation	Seen in	Status	
NPC2	C>T	None	0 copies	Clear

Blood disorders

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	0 copies

Clear

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 dysfuctional 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

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>T	None	0 copies
CYB5R3	C>G	None	0 copies

Clear

Musculoskeletal and connective tissue disorders

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	Mutation	Seen in	Status	
CYP2R1	del(A)	None	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

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

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	Clear
CYP27B1	C>A	None	0 copies	

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	Mutation	Seen in	Status	
ACVR1	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	
MYBPC3	G>A	Ragdoll	0 copies	
MYBPC3	C>G	Maine Coon	0 copies	Clear
MYH7	C>T	None	0 copies	

Renal disorders

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

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	G>A	None	0 copies
SLC7A9	T>A	None	0 copies
SLC7A9	C>T	None	0 copies
SLC7A9	G>A	None	0 copies
SLC7A9	A>G	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

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

Eye disorders

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	Mutation	Seen in	Status	
AIPL1	C>T	Persian	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	Mutation	Seen in	Status	
KIF3B	C>T	Bengal	0 copies	Clear

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	Mutation	Seen in	Status	
LTBP2	ins(CTC..)	Siamese	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	Mutation	Seen in	Status	
CEP290	A>C	Abyssinian, Somali	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	Mutation	Seen in	Status	
KRT14	G>A	None	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	Mutation	Seen in	Status	
NSDHL	A>G	None	0 copies	Clear

Autoimmune disorders

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	Mutation	Seen in	Status	
FASLG	ins(A)	British Shorthair	0 copies	Clear

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	Mutation	Seen in	Status	
ITGB2	del(GCC..)	None	0 copies	Clear

Endocrine disorders

Congenital Adrenal Hyperplasia

Congenital adrenal hyperplasia is an autosomal recessive disease characterized by an inbalance 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	Mutation	Seen in	Status	
CYP11B1	G>A	None	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	Mutation	Seen in	Status	
TPO	C>T	None	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	Mutation	Seen in	Status	
TAC3	C>T	None	0 copies	Clear

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@basepaws.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 texture

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
LPAR6	del(GTT..)	Cornish Rex, German Rex	0 copies
KRT71	TCC..>ATC..	Devon Rex	0 copies
KRT71	C>T	Devon Rex	0 copies
KRT71	C>G	Devon Rex	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

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	ins(GT)	Lykoi	0 copies
HR	GT>GCA..	Lykoi	0 copies
HR	G>A	Lykoi	0 copies
HR	C>T	Lykoi	0 copies
HR	C>T	Lykoi	0 copies
HR	ins(GAC..)	Lykoi	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	Mutation	Seen in	Status
KRT71	TCC..>ATC..	Sphynx, Kohana	0 copies
KRT71	C>T	Sphynx, Kohana	0 copies

Not Likely To Have

Coat color and pattern

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	C>A	None	0 copies
LVRN	G>A	None	0 copies
LVRN	A>G	None	0 copies

Not Likely To Have

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	Mutation	Seen in	Status
LVRN	C>A	None	0 copies
LVRN	A>G	None	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	Mutation	Seen in	Status
TYR	G>A	None	0 copies
TYR	del(G)	None	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	G>A	Siamese, Birman, Himalayan	0 copies
TYR	G>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	0 copies
TYR	C>A	Siamese, Birman, Himalayan	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	0 copies
TYRP1	G>A	None	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

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

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

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. An MC1R mutation described in Kurilian Bobtail cats is associated with the copal coat color, expressed as warm red at first, followed by a transition to an apricot-like shade during the first year of life.

Gene	Mutation	Seen in	Status	
MC1R	del(GCG..)	Kurilian Bobtail	0 copies	Not Likely To Have

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	G>T	Bengal	0 copies	
ASIP	T>C	Bengal	0 copies	
ASIP	A>T	Bengal	0 copies	
ASIP	T>C	Bengal	0 copies	
ASIP	A>G	Bengal	0 copies	Not Likely To Have
ASIP	C>T	Bengal	0 copies	
ASIP	A>G	Bengal	0 copies	
ASIP	del(CA)	Bengal	0 copies	
ASIP	C>G	Bengal	0 copies	

Coat color and pattern

Black coat color

The gene agouti/ASIP has been implicated in pigmentation, including melanism, in mice and other animals. If a cat carries 2 copies of a particular 2-basepair deletion in the ASIP gene, their coat is likely to be a solid or black color.

Gene	Mutation	Seen in	Status	
ASIP	★★★★☆ del(CA)	None	0 copies	Not Likely To Have

Dilute coat color

The dilute coat color phenotype in cats is related to both the eumelanin and phaeomelanin 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

Body morphology

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

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	T>A	None	0 copies	
ZRS	T>C	None	0 copies	Not Likely To Have
ZRS	C>G	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	GGC..>CTG..	Manx, American Bobtail, Pixie-bob	0 copies
T	del(A)	Manx, American Bobtail, Pixie-bob	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	T>G	Maine Coon, Persian, Ragdoll, Somali	0 copies
FGF5	del(A)	Maine Coon, Persian, Ragdoll, Somali	0 copies
FGF5	A>T	Maine Coon, Persian, Ragdoll, Somali	0 copies

Not 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

Thank you!
Stay healthy.

VERSION 4.0

basepaws