

# Apollo's Genetic Report



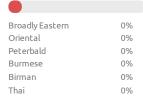
#### Summary Page

# **Breeds** Group

#### Western 100%

Maine Coon	100%
Broadly Western	0%
Ragdoll	0%
Russian Blue	0%
Norwegian Forest Cat	0%
Siberian	0%
American Shorthair	0%
Abyssinian	0%
Turkish Van	0%
Turkish Angora	0%

#### Eastern 0%



#### Persian 0%

0%
0%
0%
0%
0%

#### Exotic 0%

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

#### Polycat 0%



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 **Apollo**

AGE: **GENDER:** male

**REPORT DATE: PARENT NAME:** 06-19-2025 Deborah Raymond

PRIMARY VET HOSPITAL:

# **Oral Health** Report

Risk score legend

**HIGH RISK** 

#### **APOLLO'S RISK OF PERIODONTAL** DISFASE

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

( ) HIGH

Risk score

MEDIUM

#### APOLLO'S RISK OF TOOTH RESORPTION

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

#### **RISK:**

( ) HIGH

Risk score

MEDIUM

#### **APOLLO'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

Risk score

MEDIUM ( ) Low

4.97

# Health **Markers**

#### **CLEAR**

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

#### **CARRIER**

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

#### AT RISK

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

#### AT HIGH RISK

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

LIKELY BLOOD TYPE:

(based on genotype)

Α

# **Trait** Markers

**LIKELY TO** HAVE

NOT LIKELY TO HAVE

**CARRIER** 

20





# Apollo'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

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.

# 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.

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 Basepaws 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 Basepaws 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@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, Hypertipoproteinaemia). 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.

#### 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

#### 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

G>T

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

CLCN1

**★★★★**del(C)

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	C>A	None	0 copies
CYP27B1	<b>★★★★</b> del(C)	None	0 copies





# 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

CYP2R1

Mutation ★★★★

del(A)

Seen in

None

Status

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

WNK4

Mutation

C>T

Seen in

Burmese

Status

0 copies

Clear

# Eye disorders

#### Progressive Retinal Atrophy (AIPL1-related)

This progressive retinal atrophy is related to Leber congenital  $amauros is \textbf{--} a \ retinal \ disease \ causing \ severe \ visual \ impairment$ 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

#### Status

\*\*\*\*

Persian

0 copies

Clear

# Late-Onset Photoreceptor Degeneration

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

CFP290

#### Mutation \*\*\*

A>C

#### Seen in

#### Status

Abyssinian, Somali

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

#### Gene

KIF3B

#### Mutation \*\*\*\*

C>T

#### Seen in

Bengal



0 copies

Clear

(AIPL1).

#### 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..)

# \*\*\*\*

#### Siamese

Seen in

#### Status









#### 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

ARSB

**ARSB** 

Mutation

\*\*\*\*

C>T

#### Seen in

Status

# A>G

Birman, Siamese

0 copies

Birman, Siamese

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

ATP7B

Mutation ★★★★

C>G

Seen in

None

Status

0 copies

Clear

#### 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** CLN6

MFSD8

Mutation ★★★★ G>A

\*\*\*\*

del(T)

Seen in

Status

None 0 copies

None 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

C>T

Seen in

None

0 copies

Status

Clear



Genetic Report

#### 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

GLB1

Mutation ★★★★

C>G

Seen in

Status

None

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

GM2A

Mutation ★★★★

del(GAC..)

Seen in

Status

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** GUSB

GUSB

Mutation ★★★★ G>A

\*\*\*

T>G

Seen in

None

0 copies

Status

Status

0 copies

0 copies

Clear

GUSB C>T None 0 copies

#### 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

HEXB

HEXB

HEXB

HEXB

Mutation

del(TAA..)

\*\*\*\*

inv(TAC..)

\*\*\*\*

del(C)

\*\*\*\*

C>T

Burmese, Korat

Seen in

Burmese, Korat 0 copies

Burmese, Korat 0 copies

Burmese, Korat 0 copies

Clear





#### 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	ins(T)	None	0 copies
HMBS	<b>★★★★</b> G>A	None	0 copies
HMBS	★本本本 C>T	None	0 copies
HMBS	G>A	None	0 copies
HMBS	del(GAG)	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(TCG)	None	0 copies
IDUA	del(GTC)	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

#### 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







#### 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

NPC1

NPC1

Mutation

Seen in

None

Status

会会会会会

C>G ★★★★ T>G

None

0 copies

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

NPC2

Mutation ★★★★

C>T

Seen in

Status

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

PKLR

Mutation ★★★★

G>A

Seen in Somali.

Abyssinian

0 copies

Status

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

SMPD1

Mutation

G>A

Seen in

None

Status

0 copies

Clear



Genetic Report

#### 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

#### **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

CYB5R3

CYB5R3

Mutation

Seen in

None

Seen in

None

Status

0 copies

**★★★★** C>T

<mark>★★★★</mark> C>G

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

F12

F12

Mutation ★★★★

del(C)

\*\*\*

G>C

0 copies

Status

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

F9

Mutation

\*\*\*\*

G>A

★★★★★ C>T Seen in None

None

0 copies

Status

0 copies

о соріс

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 ITGA2B Mutation

★★★★

del(G)

Seen in

None

een in

Status
0 copies

Clear



Genetic Report

#### **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.

#### Mutation Seen in Gene Status

\*\*\*\* CYP11B1 G>A

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

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
TD 0	*nknknknk		
TPO	C>T	None	0 copies





#### 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

\*\*\*\* del(GCC..) ITGB2

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

GRHPR

Mutation ★★★★

G>A

Seen in

Status

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

PKD1

Mutation

C>A

Seen in

Status

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

SLC3A1

Mutation

C>T

Seen in

None

Status

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

SLC7A9

SLC7A9

SLC7A9

SI C7A9

Mutation

C>T

\*\*\*\*

G>A

G>A

\*\*\*\*

A>G

Seen in

Status

None

0 copies

\*\*\*\*

None

None

None

0 copies

0 copies

Clear

SLC7A9

\*\*\*\*

T>A

None

0 copies

0 copies



Genetic Report

#### 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

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
	****		
NCDHI	A > C	None	0 copies



# 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	<b>☆☆☆☆</b> G>A	Ragdoll	0 copies
MYBPC3	C>G	Maine Coon	0 copies
MVH7	★★★★★ C>T	None	0 copies



# 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
	食食食食食	
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.

# 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

#### 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

del(CA)

ASIP

None 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.

Mutation	Seen in	Status
del(CA)	Bengal	0 copies
<b>★★★★</b> C>G	Bengal	0 copies
A>G	Bengal	0 copies
<mark>★★★★</mark> C>T	Bengal	0 copies
A>G	Bengal	0 copies
T>C	Bengal	0 copies
<b>★★★★</b> A>T	Bengal	0 copies
T>C	Bengal	0 copies
<b>☆☆☆☆</b> G>T		0 copies
	del(CA)  C>G  A>G  C>T  A>G  A>C  T>C	del(CA) Bengal  C>G Bengal  A>G Bengal  C>T Bengal  T>C Bengal  T>C Bengal  Bengal  T>C Bengal  Bengal

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	A>G	None	0 copies
LVRN	C>A	None	0 copies
LVRN	<b>★★★★</b> G>A	None	0 copies



#### 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

\*\*\*\* A>G None

IVRN

\*\*\*\* LVRN C>A 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

\*\*\* Norwegian MC1R G>A Forest Cat

0 copies

0 copies

Not Likely To Have

#### 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

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





#### 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

**★★★★** del(T)

MLPH

Gene

TYR

TYR

TYR

None 1 copies

Carrier

#### 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.

Mutation

del(G)

★★★★

G>A

None

Seen in

None

0 copies

0 copies

0 copies

Status

Not Likely To Have

#### 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	del(G)	Siamese, Birman, Himalayan	0 copies
TYR	del(G)	Siamese, Birman, Himalayan	0 copies
TYR	G>A	Siamese, Birman, Himalayan	0 copies
TYR	<b>★★★★</b> G>A	Siamese, Birman, Himalayan	0 copies
TYR	★★★★ C>T	Siamese, Birman, Himalayan	0 copies

Siamese,

Birman,

Himalayan

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 G>A None 0 copies

TYRP1 C>G None 0 copies

\*\*\*\*

C>A





#### 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
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TYRP1 C>T None 0 copies



# Coat length

	Gene	Mutation	Seen in	Status	
Long-haired coat The long-haired coat can be a typical feature of Maine Coon,		****	Maine Coon, Persian, Ragdoll,		
Persian, Ragdoll, and Somali cats, as well as of some mixed-breed	FGF5	G>T	Somali	2 copies	
cats. The long-haired coat phenotype is associated with various mutations in the fibroblast growth factor 5 (FGF5) gene.	FGF5	del(A)	Maine Coon, Persian, Ragdoll, Somali	0 copies	
	FGF5	<b>★★★★</b> T>G	Maine Coon, Persian, Ragdoll, Somali	2 copies	
			Maine Coon,		Likely To Have
	FGF5	A>T	Persian, Ragdoll, Somali	0 copies	
		****	Maine Coon, Persian, Ragdoll,		
	FGF5	ins(A)	Somali	0 copies	
		*Arkirkok	Maine Coon, Persian, Ragdoll,		
	FGF5	G>A	Somali	0 copies	

#### Coat texture

#### 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
TIIX	, ,	Lykoi	o copies
HR	<del>食食食食</del> G>A	Lykoi	0 copies
HR	C>T	Lykoi	0 copies
HR	<b>☆☆☆☆</b> C>T	Lykoi	0 copies
HR	ins(GAC)	Lykoi	0 copies
HR	GT>GCA	Lykoi	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	★★★★★ TCC>ATC	Devon Rex	0 copies
KRT71	<b>☆☆☆☆</b> C>G	Devon Rex	0 copies
KRT71	<b>★★★★★</b> C>T	Devon Rex	0 copies
LPAR6	del(GTT)	Cornish Rex, German Rex	0 copies



# Coat texture

	Gene	Mutation	Seen in	Status	
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.	KRT71	TCC>ATC	Sphynx, Kohana	0 copies	
		****			Not Likely To Have
	KRT71	C>T	Sphynx, Kohana	0 copies	

# Body morphology

#### 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

#### Gene

HES7

#### Mutation

#### Seen in

#### Status

\*\*\*\* A>G

Japanese Bobtail

0 copies

Not Likely To Have

#### 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.

ie	ne		
30			

# Mutation del(A)

#### Seen in Manx, American Bobtail, Pixiebob

# 0 copies

Status

#### \*\*\*\* del(G)

\*\*\*\*

del(G)

# 0 copies

Bobtail, Pixie-

bob

# 0 copies

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

TRPV4

Т

#### Mutation 会会会会会 C>A

#### Seen in

None

# Status

0 copies

Not Likely To Have



Genetic Report

# 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>C	None	0 copies
	the state of the s		

None

0 copies

T>A

ZRS