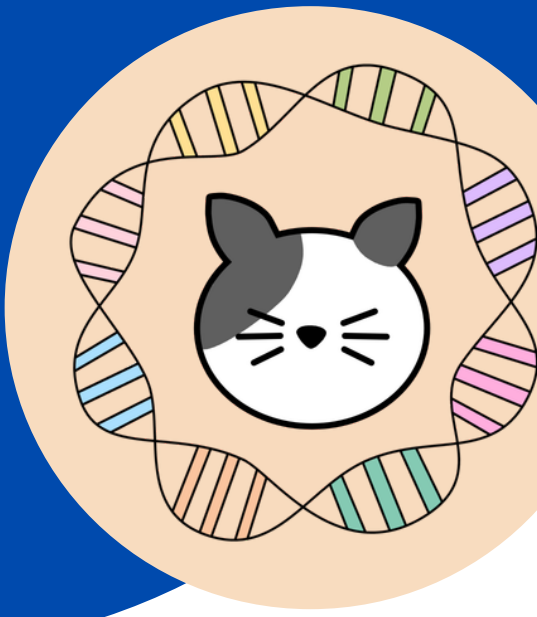
FELOME GENETICS

Premium DNA Test

Felome

version 20251201



- **300+** Genetic Variants
- **70+** Disease Variants
- **50+** Traits and Colors
- Blood Type (genetic)
- MDR1 Drug Sensitivity
- Genetic Diversity (heterozygosity)

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CONTENTS

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How to use this genetic analysis for your cat

Screening

These results indicate your cat's likelihood of developing a disease or trait. Only a veterinarian can diagnose a disease in your cat. This genetic analysis is intended for informative and preventative use only.

Consultation

Discuss the results with your veterinarian, who can help you plan your cat's health journey to promote early detection and, if necessary, management of certain diseases.

Action

Anticipate your cat's needs to improve their well-being and life expectancy. For breeders, these results are a valuable tool to guide responsible breeding practices and preserve the health of pedigrees.

Recently discovered genetic variants...

Some genetic variants have been published by the scientific community in the **last 5 years**. Due to their novelty, the frequency and importance of these variants in cat populations is not always clear and there may be a lack of data concerning their correlation with symptoms. Therefore, it is important that we continue to screen for these variants and monitor their impact. Meanwhile, we will provide the complete results in this report.

Guide to interpreting results

Hereditary Diseases

Non-Carrier

Your cat will not develop the disease associated with this genetic variant.
However, your cat may develop this disease due to other untested variants.

Carrier

Your cat will not develop the disease associated with this genetic variant.

- However, your cat may develop this disease due to other untested variants.
- Your cat can transmit the variant to their offspring.

At Risk

Your cat is at risk of developing the disease associated with this genetic variant.
The probability of developing the disease varies depending on the variant: in some cases it is close to 100%, while in others it can be around 50 to 75%.

- Your cat can transmit the variant to their offspring.
- Talk to your veterinarian for early detection and appropriate treatment.

Hereditary Traits

Non-Carrier

Your cat will not exhibit the trait associated with this genetic variant.

- However, your cat may exhibit this trait due to other untested variants.

Carrier

Your cat will not exhibit the trait associated with this genetic variant.

- However, your cat may exhibit this trait due to other untested variants.
- Your cat can transmit the variant to their offspring.

Expressed

Your cat will exhibit the trait associated with this genetic variant.
Your cat will normally express this trait, but the overall appearance also depends on the interaction between other genetic variants. For some traits, other genes or variants can modify or mask their appearance!

- Your cat can transmit the variant to their offspring.

ID number : 000 000 000 000 000
Sex : Male
Cat name : **Milo**

Date : November 14, 2025
ID kit : S00KIT00000
Client Name : James Smith

Health Summary

Kidney & Urinary Diseases
- 5 Non-carrier
- 0 Carrier
- 0 At Risk

Hypertrophic Cardiomyopathy
- 4 Non-carrier
- 0 Carrier
- 0 At Risk

Eye Diseases
- 5 Non-carrier
- 0 Carrier
- 0 At Risk

Metabolic & Endocrine Disorders
- 31 Non-carrier
- 0 Carrier
- 0 At Risk

Blood & Immune Disorders
- 10 Non-carrier
- 0 Carrier
- 0 At Risk

Musculoskeletal Disorders
- 9 Non-carrier
- 0 Carrier
- 0 At Risk

Dermatological & Nervous System Disorders
- 6 Non-carrier
- 0 Carrier
- 0 At Risk

Developmental Morphology
- 2 Non-carrier
- 0 Carrier
- 0 At Risk

The genetic **Blood Type** of your cat : **A**

Genotype of your cat: A/b

Drug Sensitivity
no

Resistance to FIV Infection
no

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Traits Summary

Coat Color & Pattern

- [x] Solid (non-Agouti)
- [ ] Charcoal
- [ ] Chocolate
- [ ] Cinnamon
- [x] Colorpoint (Siamese)
- [ ] Colorpoint (Burmese)
- [ ] Mink
- [ ] Albinism
- [x] Orange or Tortie
- [x] Dilution
- [ ] Amber
- [ ] Copal / Carnelian
- [ ] Russet
- [ ] Gloving
- [ ] Glitter
- [x] Tabby Blotched
- [ ] Tabby Ticked
- [ ] Dominant White
- [x] White Spotting
- [ ] Copper
- [ ] Extreme Sunshine
- [ ] Sunshine

Coat Type

- [ ] Lykoi coat
- [x] Long hair
- [ ] Nudity
- [ ] Rexing (Curly Hair)

Eye Color

- [ ] Dominant Blue Eyes (DBE-AGO)
- [ ] Dominant Blue Eyes (DBE-ALT)
- [ ] Dominant Blue Eyes (DBE-RE)
- [ ] Dominant Blue Eyes (DBE-CEL)
- [ ] Pink-eye (Donskoy)

Body Morphology

- [ ] Folded Ears
- [ ] Polydactyly
- [ ] Bobtail

% Genetic Diversity Heterozygosity

Your cat's percentage of heterozygosity is: **27 %**

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Blood Type

Blood Types A, B, AB

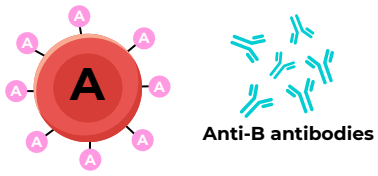
Cats have three main blood serotypes called type A, type B, and type AB (also known as type C), which are determined by the activity of an enzyme encoded by the CMAH gene. Several genetic variants can influence the activity of this enzyme..

* **It is possible to have Type B with 1 copy of 2 different variants.**
* **Cats with the genotype c/b will be Type C.**

The genetic **Blood Type** of your cat : **A**

Genotype of your cat: A/b

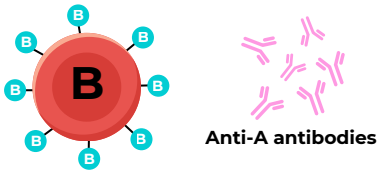
Type A



Possible Genotypes :
A/A, A/b1, A/b2, A/b3, A/c

Transfusion :
They can receive Type A blood.

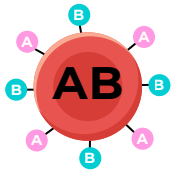
Type B



Possible Genotypes :
b1/b1, b2/b2, b3/b3, b1/b2, b1/b3, b2/3

Transfusion :
They can receive Type B blood.

Type C (AB)



There are no anti-A or anti-B antibodies present.

Possible Genotypes :
c/c, c/b1, c/b2, c/b3

Transfusion :
They can receive Type C blood, or Type A (with caution) if needed.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Type B (b1)	*CMAH*	c.179G>T	recessive	A/A	0	Non-carrier
Type B (b2)	*CMAH*	c.268T>A	recessive	A/b	1	Carrier
Type B (b3)	*CMAH*	c.1322delT	recessive	A/A	0	Non-carrier
Type C/AB (c)*	*CMAH*	c.364C>T	recessive	A/A	0	Non-carrier

*** This variant of Type C is rare and mainly seen in Ragdolls.**

Note: *DNA blood typing may not match serological results for a small percentage of individuals (<1%) due to the presence of multiple variants in the CMAH gene that may cumulatively impact its function and are still being studied. The accuracy of some variants for Type B may vary between breeds. We recommend that you ask your veterinarian for a serological test to confirm your cat's blood type if you need it for medical decisions or if you are considering breeding.*

Scientific references
2007 Bighignoli et al., "Cytidine monophospho-N-acetylneuraminic acid hydroxylase (CMAH) mutations associated with the domestic cat AB blood group." BMC Genet 8:27.
2014 Tasker et al., "Feline blood genotyping versus phenotyping, and detection of non-AB blood type incompatibilities in UK cats." Journal of Small Anim Pract 55(4):185-9.
2016 Gandolfi et al., "A Novel Variant in CMAH Is Associated with Blood Type AB in Ragdoll Cats." PLoS One 11(5):e0154973.
2018 Kehl et al., "Molecular characterization of blood type A, B, and C (AB) in domestic cats and a CMAH genotyping scheme." PLoS One 13(9):e0204287.
2022 Anderson et al., "Genetic epidemiology of blood type, disease and trait variants, and genome-wide genetic diversity in over 11,000 domestic cats." PLoS Genet 18(6).

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Drug sensitivity (Multidrug-Resistance 1)

Drug sensitivity is caused in particular by a reduced ability to eliminate certain types of drugs. The accumulation of toxic levels in the nervous system can lead to adverse reactions.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
MDR1	*ABCB1*	c.1930_1931delTC	recessive	N/N	0	Non-carrier

Breeds mainly affected: Turkish Angora, Balinese, Maine Coon, Ragdoll, Russian, Siamese

Scientific references

2015 Mealey et al., "Identification of a nonsense mutation in feline ABCB1." J Vet Pharmacol Ther 38(5):429-33.
2021 Mealey et al., "ABCB11930_1931del TC gene mutation in a temporal cluster of macrocyclic lactone-induced neurologic toxicosis in cats associated with products labeled for companion animal use." J Am Vet Med Assoc 259:72-76.

Resistance to FIV infection

A genetic variant has been identified that has undergone positive selection during evolution in cats, and may be associated with differences in susceptibility to feline immunodeficiency virus (FIV) infection.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Res	*APOBEC3*	c.193_194GC>AT	dominant	N/N	0	Non-carrier

Scientific references

2016 Yoshikawa et al., "A Naturally Occurring Domestic Cat APOBEC3 Variant Confers Resistance to Feline Immunodeficiency Virus Infection." J Virol 90(1):474-85.

Kidney & Urinary Diseases

Cystinuria

Cystinuria causes the formation of cystine crystals in the urinary system.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Type IA	*SLC3A1*	c.1342C>T	recessive	N/N	0	Non-carrier
Type B	*SLC7A9*	c.881T>A	recessive	N/N	0	Non-carrier

Breeds mainly affected (Type B): Domestic Cats, Maine Coon, Siamese, Siberian, Sphynx

Scientific references

2015 Mizukami et al., "Feline cystinuria caused by a missense mutation in the SLC3A1 gene." Scientific Reports 11(1):7159.
2016 Mizukami et al., "Cystinuria Associated with Different SLC7A9 Gene Variants in the Cat." PLoS One 11(7):e0159247.

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Kidney & Urinary Diseases

Hyperoxaluria

Hyperoxaluria causes the deposition of oxalate crystals in the urinary system.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Type II	*GRHPR*	Splicing G>A	recessive	N/N	0	Non-carrier

Scientific references

2009 Goldstein et al., "Primary Hyperoxaluria in Cats Is Caused by a Mutation in the Feline GRHPR Gene." Journal of Heredity 100:S2-S7.

Polycystic Kidney Disease

Polycystic kidney disease is a condition characterized by the presence of cysts in the kidneys that can be detected early in life by ultrasound, before symptoms of kidney failure appear. Kidney failure occurs later and can begin over a long period of time (3-10 years) with an average of 7 years.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
PKD1	*PKD1*	c.10063C>A	dominant	N/N	0	Non-carrier
PKD2	*PKD2*	c.2211del	dominant	N/N	0	Non-carrier

Breeds primarily affected (PKD1): American Shorthair, American Wirehair, British Longhair, British Shorthair, Burmilla, Exotic Shorthair, Highland Fold, Persian, Scottish Fold, Selkirk Rex

Breeds mainly affected (PKD2): Siberian

What can you observe before visiting the vet?

Polycystic kidney disease is usually characterized by increased fluid intake and daily urine output, weight loss, and lethargy.

Scientific references

2004 Lyons et al., "Feline polycystic kidney disease mutation identified in PKD1." J Am Soc Nephrol 15(10):2548-55.
2021 Rodney et al., "A domestic cat whole exome sequencing resource for trait discovery." Scientific Reports 11(1):7159.

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Cardiomyopathy

Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is the most common heart disease in cats, with varying frequencies depending on the breed. It is characterized by an increase in cardiac muscle volume, which can be seen on a chest ultrasound. Electrocardiography (ECG) or Doppler imaging can also be performed, and when the disease is advanced, a heart murmur, galloping sound, or arrhythmia may be heard on cardiac auscultation.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
HCM-MC	*MYBPC3*	c.91G>C (A31P)	dominant *	N/N	0	Non-carrier

*** Dominant with incomplete penetrance: Carriers of 1 copy are at moderate risk of developing hypertrophic cardiomyopathy and veterinary monitoring is recommended, but the exact mode of transmission remains controversial.**

Breeds mainly concerned (HCM-MC): Maine Coon, Munchkin, Scottish Fold

What can you observe before visiting the vet?
Affected cats (HCM-MC) are mostly asymptomatic, sometimes with signs of heart failure such as exercise intolerance, loss of appetite, respiratory effort, or even pleural edema or effusion. These symptoms can develop from 6 months to 18 years of age, peaking at 4 years of age. HCM may also cause thromboembolism, which would be characterized by posterior paralysis or even sudden death.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
HCM-R	*MYBPC3*	c.2460C>T (R820W)	dominant *	N/N	0	Non-carrier

*** Dominant with incomplete penetrance: Carriers of 1 copy are at moderate risk of developing hypertrophic cardiomyopathy and veterinary monitoring is recommended, but the exact mode of transmission remains controversial.**

Breeds primarily affected (HCM-R): Ragdoll

What can you observe before visiting the vet?
Most affected cats (70%) show no symptoms (HCM-R), but signs of heart failure (such as exercise intolerance, loss of appetite, respiratory effort, or even pleural edema or effusion) or hindlimb paralysis may be observed. These symptoms can develop from 3 months to 17 years of age.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
HCM-A	*ALMS1*	c.7384G>C	unknown *	N/N	0	Non-carrier
HCM-H	*MYH7*	c.5647G>A	unknown *	N/N	0	Non-carrier

*** The importance of this variant in the development of hypertrophic cardiomyopathy and the exact mode of transmission remain controversial.**

Breeds primarily affected (HCM-A): American Shorthair, Exotic Shorthair, Munchkin, Scottish Fold, Sphynx

Scientific references
2005 Meurs et al., "A cardiac myosin binding protein C mutation in the Maine Coon cat with familial hypertrophic cardiomyopathy." Hum Mol Genet 14(23):3587-93.
2007 Meurs et al., "A substitution mutation in the myosin binding protein C gene in ragdoll hypertrophic cardiomyopathy." Genomics 90(2):261-4.
2019 Schipper et al., "A feline orthologue of the human MYH7 c.5647G>A (p.(Glu1883Lys)) variant causes hypertrophic cardiomyopathy in a Domestic Shorthair cat." Eur J Hum Genet 27(11):1724-1730.
2021 Meurs et al., "A deleterious mutation in the ALMS1 gene in a naturally occurring model of hypertrophic cardiomyopathy in the Sphynx cat." Orphanet J Rare Diseases 16(108).
2024 Boeykens et al., "Classification of feline hypertrophic cardiomyopathy-associated gene variants according to the American College of Medical Genetics and Genomics guidelines." Front Vet Sci 11(1327081).

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Eye Diseases

Progressive Retinal Atrophy, Type B

Progressive retinal atrophy (PRA or PRA) type b is an eye disorder that affects many cats worldwide. 18% of Bengals are affected in Europe. Retinal degeneration can be observed early in their life (around 9 weeks) by electroretinography. A visual deficit gradually sets in, between 2 and 5 months, until a later loss of vision, around 1 year, first at night and then during the day. It is possible to assess the cat's vision by an eye examination and a so-called "obstacle" test.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
PRA-b	*KIF3B*	c.1000G>A	recessive	N/N	0	Non-carrier

Breeds mainly affected: Bengal

What can you observe before visiting the vet?
Affected cats have dilated pupils and difficulty moving.

Scientific references
2020 Cogné et al., "Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy." Am J Hum Genet 106(6):893-904.

Progressive Retinal Atrophy, PRA-pd type

Progressive retinal atrophy (PRA) type pd is characterized by a gradual loss of vision. The onset of the disease begins around 5 weeks.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
PRA-pd	*AIPL1*	c.577C>T	recessive	N/N	0	Non-carrier

Breeds mainly affected: Persian/Exotic shorthair

Scientific references
2016 Lyons et al., "Whole genome sequencing in cats, identifies new models for blindness in AIPL1 and somite segmentation in HES7." BMC Genomics 17:265.

Progressive Retinal Atrophy, rdAc type

Progressive retinal atrophy (PRA or PRA) of the rdAc type affects the photoreceptor cells of the eyes (rods), leading to a progressive loss of their night vision. Then, daytime vision is also reduced following the damage to certain cells (cones) which are responsible for detecting colors. The onset of the disease begins around 2 years of age and total blindness occurs between 3 and 5 years. Electroretinography diagnoses the disease from the age of 8 to 12 weeks. A histological examination reveals deformations of the rods from 5 months, and of the cones from 1 year. An ophthalmoscopic analysis also allows observation of the back of the eye.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
PRA-rdAc	*CEP290*	IVS50+9T>G	recessive	N/N	0	Non-carrier

Breeds mainly concerned: Abyssinian, American Curl, Californian Rex, Cornish Rex, Munchkin, Chausie, German Rex, Havana Brown, Ocicat, Savannah, Snowshoe, Siamese, Singapura, Somali, Thai, Tonkinese

What can you observe before visiting the vet?
Clinical signs are difficult to perceive except for changes in behavior such as frequent bumping into furniture, etc.

Scientific references
2007 Menotti-Raymond et al., "Mutation in CEP290 discovered for cat model of human retinal degeneration." J Hered 98(3):211-20.

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Eye Diseases

Cone and Rod Dysplasia

Progressive retinal atrophy (PRA or PRA) type Rdy is characterized by vision loss early in life.

Phenotype	Gene	Variant(s).	Mode of Inheritance	Genotype	Copies	Result
Rdy	*CRX*	c.546delC	dominant *	N/N	0	Non-carrier

*** Dominant with incomplete penetrance**

Breeds mainly affected: Abyssinian, Somali

Scientific references
2010 Menotti-Raymond et al., "Mutation discovered in a feline model of human congenital retinal blinding disease." Invest Ophthalmol Vis Sci 51(6):2852-9.

Glaucoma (hereditary)

Glaucoma is characterized by damage to the optic nerve.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
PCG	*LTBP2*	c.1449_1452dup	recessive	N/N	0	Non-carrier

Breeds mainly concerned: Siamese

Scientific references
2016 Kuehn et al., "A Mutation in LTBP2 Causes Congenital Glaucoma in Domestic Cats (Felis catus)." PLoS One 11(5):e0154412.

Metabolic & Endocrine Disorders

Aciduria (L-2-hydroxyglutaric)

L-2-hydroxyglutaric aciduria is a condition caused by a metabolic defect that results in nervous symptoms (including seizures).

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
2HGA	*L2HGDH*	c.1301A>G	recessive	N/N	0	Non-carrier

Scientific references
2021 Christen et al., "L2HGDH Missense Variant in a Cat with L-2-Hydroxyglutaric Aciduria." Genes 12(5):682.

Neuronal Ceroid Lipofuscinosis

Neuronal ceroid lipofuscinoses are neurodegenerative diseases caused by the accumulation of lipofuscin in cells of the central nervous system.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Type 6A	*CLN6*	c.668G>A	recessive	N/N	0	Non-carrier
Type 7	*MFSD8*	c.780delT	recessive	N/N	0	Non-carrier

Scientific references
2020 Katz et al., "Neuronal Ceroid Lipofuscinosis in a Domestic Cat Associated with a DNA Sequence Variant That Creates a Premature Stop Codon in CLN6." G3 10(8):2741-2751.
2020 Guevar et al., "A major facilitator superfamily domain 8 frameshift variant in a cat with suspected neuronal ceroid lipofuscinosis." J Vet Intern Med 34(1):289-293.

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Metabolic & Endocrine Disorders

Dihydropyrimidinase Deficiency

Dihydropyrimidinase deficiency is a rare disease that causes vomiting, malnutrition, and weakness.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
DHP	*DPYS*	c.1303G>A	recessive	N/N	0	Non-carrier

Scientific references

2012 Chang et al., "Dihydropyrimidinase deficiency: the first feline case of dihydropyrimidinuria with clinical and molecular findings." JIMD Reports 6:21-6.

Gangliosidosis

Gangliosidoses are neurodegenerative conditions affecting different races, due to various mutations.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Type I (GM1)	*GLB1*	c.1448G>C	recessive	N/N	0	Non-carrier
Type IIA (GM2A)	*GM2A*	c.516_519delGGTC	recessive	N/N	0	Non-carrier
Type II (GM2)	*HEXB*	c.1467_1491inv	recessive	N/N	0	Non-carrier
Type II (GM2)	*HEXB*	c.667C>T	recessive	N/N	0	Non-carrier
Type II (GM2-B)	*HEXB*	c.1244-8_1250del	recessive	N/N	0	Non-carrier
Type II (GM2-K)	*HEXB*	c.39delC	recessive	N/N	0	Non-carrier

What can you observe before visiting the vet?

Clinical signs (GM1) appear from the age of 4 to 6 months and are manifested by vision disorders, loss of balance, head tremors, opacification of the cornea and especially ataxia (lack of coordination of movements) of the hind limbs which quickly affects all four limbs and ends up preventing the animal from moving.

Affected cats (GM2) suffer from neurological disorders, such as loss of balance, head tremors, ataxia and vision disturbances. More generally, the disease occurs in the cat's early life between birth and 2 years of age.

Clinical signs (GM2-B) generally appear between 6 and 8 weeks of age and are manifested by loss of balance, head tremors, progressive loss of coordination of movements (ataxia) until an inability to stand and move. Corneal opacification may also occur as well as vision disturbances.

Symptoms of the disease (GM2-K) appear between 1 and 3 months of age; loss of balance, head tremors, loss of coordination of movements (ataxia) progressively leading to the inability to stand and move, clouding of the cornea, and finally vision disturbances.

Breeds mainly concerned (GM1): Balinese, Havana Brown, Korat, Siamese, Oriental, Mandarin, Peterbald, Thai

Breeds mainly concerned (GM2-B): Asian, Burmese, Burmilla, Bombay, Tonkinese

Breeds mainly concerned (GM2-K): Korat

Scientific references

1994 Muldoon et al., "Characterization of the molecular defect in a feline model for type II GM2-gangliosidosis (Sandhoff disease)." Am J Pathol 144(5):1109-18.
2004 Martin et al., "An inversion of 25 base pairs causes feline GM2 gangliosidosis variant." Exp Neurol 187(1):30-7.
2005 Martin et al., "Mutation of the GM2 activator protein in a feline model of GM2 gangliosidosis." Acta Neuropathology 110(5):443-50.
2007 Hasegawa et al., "Clinical and molecular analysis of GM2 gangliosidosis in two apparent littermate kittens of the Japanese domestic cat." J Feline Med Surg 9:232-7.
2007 Kanae et al., "Nonsense mutation of feline beta-hexosaminidase beta-subunit gene causing Sandhoff disease in a family of Japanese domestic cats."Res Vet Sci 82:54-60.
2008 Martin et al., "Molecular consequences of the pathogenic mutation in feline GM1 gangliosidosis." Mol Genet Metab 94(2):212-21.
2009 Bradbury et al., "Neurodegenerative lysosomal storage disease in European Burmese cats with hexosaminidase beta-subunit deficiency." Mol Genet Metab 97(1):53-9.

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Metabolic & Endocrine Disorders

Glycogen Storage Disease, Type IV (GSD4)

Glycogen storage disease is a glycogen storage disease caused by an enzyme deficiency. This storage causes progressive organ dysfunction.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
GSD4	*GBE1*	334 bp ins 5'-6.2kb del	recessive	N/N	0	Non-carrier

Breeds mainly affected: Norwegian Forest Cat

What can you observe before visiting the vet?

Kittens may appear healthy until 5 months of age, followed by a gradual onset of clinical signs such as persistent fever, tremors, involuntary muscle contractions and an inability to move. More severely affected kittens may die within a few hours of birth. Some symptoms that are present in the animal are not clinically visible, such as cirrhosis or liver weakness.

Scientific references

2007 Fyfe et al., "A complex rearrangement in GBE1 causes both perinatal hypoglycemic collapse and late-juvenile-onset neuromuscular degeneration in glycogen storage disease type IV of Norwegian Forest Cats." Mol Genet Metab 90:383-92.

Hyperlipoproteinemia

Hyperlipoproteinemia leads to growth defects, fatty deposits in the skin, and retinal defects.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
HLP	*LPL*	c.1315G>A	recessive	N/N	0	Non-carrier

Scientific references

1996 Ginzinger et al., "A mutation in the lipoprotein lipase gene is the molecular basis of chylomicronemia in a colony of domestic cats." J Clin Invest 97(5):1257-66.

Congenital Adrenal Hyperplasia

Congenital adrenal hyperplasia is a hereditary endocrine disorder caused by a deficiency of steroidogenic enzymes and characterized by adrenal insufficiency and changes in male hormone levels.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
HCS	*CYP11B1*	c.1151G>A	recessive	N/N	0	Non-carrier

Scientific references

2012 Owens et al., "Congenital adrenal hyperplasia associated with mutation in an 11β-hydroxylase-like gene in a cat". J Vet Intern Med Sep-Oct;26(5):1221-6.

Hypogonadisme

Hypogonadotropic hypogonadism is a cause of incomplete puberty.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
HH	*TAC3*	c.220G>A	recessive	N/N	0	Non-carrier

Scientific references

2019 Hug et al., "A TAC3 Missense Variant in a Domestic Shorthair Cat with Testicular Hypoplasia and Persistent Primary Dentition." Genes 14(10):806.

Hypothyroidism

Congenital (hereditary) hypothyroidism results in reduced thyroid hormone production. Affected cats have disproportionate dwarfism and morphological abnormalities, sometimes with an enlarged thyroid (goiter).

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
HT (2015)	*TPO*	c.1333G>A	recessive	N/N	0	Non-carrier
HT (2022)	*TPO*	c.430G>A	recessive	N/N	0	Non-carrier

Breeds mainly concerned: Birman, British Longhair, British Shorthair, Chartreux, Exotic Shorthair, Maine Coon, Norwegian Forest Cat, Russian Blue, Siamese, Siberian, Sphynx

Scientific references

2015 Giger et al., "Congenital hypothyroidism with goiter in cats due to a TPO mutation." J Vet Intern Med 29:448.
2022 Van Poucke et al., "Association of recessive c.430G>A (p.(Gly144Arg)) thyroid peroxidase variant with primary congenital hypothyroidism in cats." J Vet Intern Med 36(5):1597-1606.

ID number : 000 000 000 000 000
Sex : Male
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Metabolic & Endocrine Disorders

Wilson's Disease

Wilson's disease is a rare genetic disorder of copper metabolism. Copper accumulation can lead to liver failure.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
WD	*ATP7B*	c.3890C>G	recessive	N/N	0	Non-carrier

Scientific references

2018 Asada et al., "Hepatic copper accumulation in a young cat with familial variations in the ATP7B gene." J Vet Intern Med 33(2):874-878.

Mucopolysaccharidoses

Mucopolysaccharidosis is caused by a deficiency of the enzyme alpha-L-iduronidase (Type I), a deficiency of N-acetylgalactosamine 4-sulfatase (Type VI), or a deficiency of beta-glucuronidase (Type VII). Symptoms vary.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Type I (MPSI)	*IDUA*	c.1042_1044delCGA	recessive	N/N	0	Non-carrier
Type VI (MPSVI)	*ARSB*	c.1427T>C	recessive	N/N	0	Non-carrier
Type VI (MPSVI)	*ARSB*	c.1558G>A	recessive	N/N	0	Non-carrier
Type VII (MPSVII)	*GUSB*	c.1051G>A	recessive	N/N	0	Non-carrier
Type VII (MPSVII)	*GUSB*	c.[1423T>G;1426C>T]	recessive	N/N	0	Non-carrier

Breeds mainly affected (Type VI): Siamese

Scientific references

1996 Yogalingam et al., "Feline mucopolysaccharidosis type VI ... identification of a mutation causing the disease." J Biol Chem 271(44):27259-65.
1998 Crawley et al., "Two mutations within a feline mucopolysaccharidosis type VI colony cause three different clinical phenotypes." J Clin Invest 101(1):109-19.
1999 He et al., "Identification and characterization of the molecular lesion causing mucopolysaccharidosis type I in cats." Mol Genet Metab 67(2):106-12.
2015 Wang et al., "Mucopolysaccharidosis VII in a Cat Caused by 2 Adjacent Missense Mutations in the GUSB Gene." J Vet Intern Med 29(4):1022-8.

Acute Intermittent Porphyria

Acute intermittent porphyria is caused by low levels of an enzyme needed for the synthesis of heme (a component of hemoglobin) which causes a reddish-brown discoloration of the teeth and urine.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
AIP	*HMBS*	c.826-1G>A	dominant	N/N	0	Non-carrier
AIP	*HMBS*	c.842_844delGAG	dominant	N/N	0	Non-carrier
AIP	*HMBS*	c.107_110delACAG	dominant	N/N	0	Non-carrier
AIP	*HMBS*	c.189dupT	dominant	N/N	0	Non-carrier
AIP	*HMBS*	c.250G>A	dominant	N/N	0	Non-carrier

Scientific references

2010 Clavero et al., "Feline acute intermittent porphyria: a phenocopy masquerading as an erythropoietic porphyria due to dominant and recessive hydroxymethylbilane synthase mutations." Hum Mol Genet 19(4):584-96.
2013 Clavero et al., "Diagnosis of feline acute intermittent porphyria presenting with erythrodontia requires molecular analyses." Vet J 198(3):720-2.

Congenital Erythropoietic Porphyria

Congenital erythropoietic porphyria is caused by low levels of an enzyme needed for the synthesis of heme (a component of hemoglobin) which causes a reddish-brown discoloration of the teeth and urine.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
PEC	*UROS*	c.[140C>T;331G>A]	recessive	N/N	0	Non-carrier

Scientific references

2010 Clavero et al., "Feline congenital erythropoietic porphyria: two homozygous UROS missense mutations cause the enzyme deficiency and porphyrin accumulation." Mol Med 16(9-10):381-8.

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Sphingomyelinosis (Niemann-Pick disease)

Sphingomyelinosis (or Niemann-Pick disease) is a lysosomal storage disease. Type A is a neurodegenerative and multiorgan disease. Type C leads to neurological deterioration and premature death.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Type A (NPA)	*SMPD1*	c.1017G>A	recessive	N/N	0	Non-carrier
Type C1 (NPC1)	*NPC1*	c.2864G>C	recessive	N/N	0	Non-carrier
Type C2 (NPC2)	*NPC2*	c.82+5G>A	recessive	N/N	0	Non-carrier

Scientific references

2003 Somers et al., "Mutation analysis of feline Niemann-Pick C1 disease." Mol Genet Metab 79(2):99-103.
2014 Zampieri et al., "Characterization of a spontaneous novel mutation in the NPC2 gene in a cat affected by Niemann Pick type C disease." PLoS One 9(11):e112503.
2020 Takaichi et al., "Feline Niemann-Pick Disease with a Novel Mutation of SMPD1 Gene." Vet Pathol 57(4):559-564.

Nervous System Disorders

Epileptic Encephalopathy (EE)

Epileptic encephalopathy (EE) is a rare disease causing seizures and behavioral disturbances, appearing as early as 3 to 4 months of age.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
EE	*not yet available - EE test is being added to our report (planned for November 2025)*					

Breeds mainly affected: Bengal

Scientific references

2025 Kaczmarska et al., "Epileptic encephalopathy in a young Bengal cat caused by CAD deficiency. Sci Rep 15:13506.

Blood & Immune Disorders

Pyruvate Kinase Deficiency (PKDef)

The enzyme pyruvate kinase controls red blood cell turnover, and a deficiency can lead to hemolytic anemia. Somali and Abyssinian cats are affected, with 10% of individuals carrying the PKLR gene mutation. Bengals are also commonly affected (approximately 20% are carriers).

Phenotype	Gene	Variant(s).	Mode of Inheritance	Genotype	Copies	Result
PKDef	*PKLR*	c.693+304 G>A	recessive	N/N	0	Non-carrier

Breeds mainly affected: Abyssinian, Bengal, Celeste, Chausie, European Shorthair, German Rex, LaPerm, Lykoi, Egyptian Mau, Munchkin, Maine Coon, Norwegian Forest Cat, Savannah, Siberian, Singapura, Somali, Toyger

What can you observe before visiting the vet?

Some cats exhibit anemia with lethargy, weight loss (due to loss of appetite), and pale or even icteric mucous membranes. The age at which symptoms appear varies greatly, ranging from 1 month to 5 years of age. The life expectancy of symptomatic cats is reduced to a maximum of 11-12 years, with sometimes very young deaths if symptoms appear early.

Scientific references

2012 Grahn et al., "Erythrocyte pyruvate kinase deficiency mutation identified in multiple breeds of domestic cats." BMC Vet Res 8:207.

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Blood & Immune Disorders

Factor IX Deficiency (Hemophilia B)

Cats with hemophilia B (factor IX deficiency) can be severely affected depending on the levels of factor IX produced, which can vary among individuals. The disease is more commonly seen in males. Females with two copies of the mutation (one on each X chromosome) are affected.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
F9	*F9*	c.383G>A	X-linked recessive	N/N	0	Non-carrier
F9	*F9*	c.1150C>T	X-linked recessive	N/N	0	Non-carrier

Scientific references

2005 Goree et al., "Characterization of the mutations causing hemophilia B in 2 domestic cats." J Vet Intern Med 19(2):200-4.

Factor XI Deficiency (Hemophilia C)

Symptoms of hemophilia C (factor XI deficiency) may include anemia, bleeding gums, or prolonged bleeding after trauma or surgery.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
F11	*F11*	c.1546G>A	recessive	N/N	0	Non-carrier

Breeds mainly affected: Maine Coon

Scientific references

2022 Kuder et al., "A Common Missense Variant Causing Factor XI Deficiency and Increased Bleeding Tendency in Maine Coon Cats." Genes 13(5):792.

Factor XII Deficiency

Factor XII deficiency in cats is a reduced activity of a clotting factor. These variants may prolong blood clotting time, but their association with an increased risk of bleeding remains to be confirmed.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
F12	*F12*	c.1631G>C	recessive	N/N	0	Non-carrier
F12	*F12*	c.1321delC	recessive	N/N	0	Non-carrier

Scientific references

2015 Bender et al., "Molecular characterization ... and identification of a mutation causing factor XII deficiency in a domestic shorthair cat colony." Vet Pathol 52(2):312-20.
2017 Maruyama et al., "A novel missense mutation in the factor XII gene in a litter of cats with factor XII deficiency." J Vet Med Sci 79(5):822-826.
2019 Maruyama et al., "Factor XII deficiency is common in domestic cats and associated with two high frequency F12 mutations. Gene 706:6-12.

Methemoglobinemia

Methemoglobinemia is a blood disorder that can be caused by a deficiency of an enzyme that controls the amount of iron in red blood cells.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
MetHb	*CYB5R3*	c.625G>A	recessive	N/N	0	Non-carrier
MetHb	*CYB5R3*	c.232-1G>C	recessive	N/N	0	Non-carrier

Scientific references

2019 Jaffey et al., "Clinical, metabolic, and genetic characterization of hereditary methemoglobinemia ... in cats." J Vet Intern Med 33(6):2725-2731.

Autoimmune Lymphoproliferative Syndrome (ALPS)

Autoimmune lymphoproliferative syndrome is a rare inherited disease causing enlarged lymph nodes and anemia.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
ALPS	*FASLG*	c.418dupA	recessive	N/N	0	Non-carrier

Breeds mainly affected: British Shorthair

Scientific references

2017 Aberdein et al., "A FAS-ligand variant associated with autoimmune lymphoproliferative syndrome in cats." BMC Vet Res 8:207.

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Blood & Immune Disorders

Nude Syndrome: Nudity and Reduced Life Expectancy

This form of nudity is associated with severe immunodeficiency in Birman kittens. The genetic mutation results in immunodeficiency caused by the absence of the thymus gland; this organ is responsible for producing white blood cells. Affected kittens generally have a weak immune response to respiratory or digestive infections and die quickly.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
NUDE	*FOXN1*	c.1030_1033 delCTGT	recessive	N/N	0	Non-carrier

Breeds mainly concerned: Birman

What can you observe before visiting the vet?

Diagnosable from birth, the hair takes on a downy appearance on the upper part of the body. The cat's wrinkled skin secretes smelly, yellowish-brown oily substances and requires specific, regular care. The whiskers and vibrissae are reduced and curved. In some kittens, the claws may even be thinner and softened.

Scientific references

2015 Abitbol et al., "A deletion in FOXN1 is associated with a syndrome characterized by congenital hypotrichosis and short life expectancy in Birman cats." PLoS One 10(3):e0120668.

Musculoskeletal Disorders

Spinal Muscular Atrophy (SMA)

Spinal muscular atrophy is a neurodegenerative disease characterized by progressive muscle weakness and wasting. It particularly affects Maine Coons, which are genetically predisposed. This condition causes neuron loss in the first few months of life.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
SMA	*LIX1 / LNPEP*	140kb del exons 4-6	recessive	N/N	0	Non-carrier

Breeds mainly affected: Maine Coon

What can you observe before visiting the vet?

Symptoms begin to develop as early as 3 months of age, with tremors in the hind limbs. As the disease progresses, muscle atrophy may occur, leading to difficulty moving, jumping, or running. Most kittens lose their ability to jump around 5 months of age. After rapid progression and loss of function, the disease stabilizes around 8 months of age, with variable motor loss depending on the cat. The life expectancy of affected cats is 8-9 years.

Scientific references

2006 Fyfe, et al., "An approximately 140-kb deletion associated with feline spinal muscular atrophy implies an essential LIX1 function for motor neuron survival." Genome Res 16:1084-90.

Fibrodysplasie Ossifiante Progressive

Fibrodysplasia ossificans progressiva is a rare disease in which muscles and tendons gradually ossify and stiffen. This causes muscle problems and deformities in cats as young as 4 to 6 months old.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
FOP	*ACVR1*	c.617G>A	dominant	N/N	0	Non-carrier

Scientific references

2019 Casal et al., "Identification of the Identical Human Mutation in ACVR1 in 2 Cats With Fibrodysplasia Ossificans Progressiva." Vet Pathol 56(4):614-618.

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Musculoskeletal Disorders

Hypokalemic Myopathy

Hypokalemic myopathy is a hereditary disease caused by low potassium levels that can lead to muscle pain and weakness that can progress to attacks or be permanent. This genetic disease is found primarily in Birman cats in Australia, Europe, and South Africa, with 20% of cats carrying a mutated allele and 2% of cats affected. In this genetic disease, potassium deficiency is due to excessive kidney loss.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
HK	*WNK4*	c.2899C>T	recessive	N/N	0	Non-carrier

Breeds mainly concerned: Asian, Bombay, Burmese, Burmilla, Tonkinese

What can you observe before visiting the vet?

Generally, the signs of this disease are episodic, but they can be chronic in some cats. In the first year of a cat's life, signs of weakness and muscle pain may be observed. Muscle pain, especially in the cervical area with ventroflexion of the head and neck, is a very characteristic sign, but it is not present in all affected cats.

Scientific references

2012 Gandolfi et al., "First WNK4-hypokalemia animal model identified by genome-wide association in Burmese cats." PLoS One 7(12):e53173.

Myotubular Myopathy

Myotubular myopathy is characterized by progressive difficulty walking and general weakness.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
XLMTM	*MTM1*	c.455C>T	X-linked recessive	N/N	0	Non-carrier

Breeds mainly affected: Maine Coon

Scientific references

2022 Kopke et al., "X-linked myotubular myopathy associated with an MTM1 variant in a Maine coon cat." J Vet Intern Med 36(5):1800-1805.

Myotonia Congenita

Cats with myotonia congenita exhibit rigid limbs, a stiff gait, and may have facial spasms or difficulty opening their jaws.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
MC	*CLCN*	c.1930+1G>T	recessive	N/N	0	Non-carrier

Scientific references

2014 Gandolfi et al., "A novel mutation in CLCN1 associated with feline myotonia congenita." PLoS One 9(10):e109926.

Osteochondromatose

Osteochondromatosis causes the appearance of lumps on the bones, which can lead to joint deterioration.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
OC	*EXT1*	c.1468dupC	dominant	N/N	0	Non-carrier

Scientific references

2022 Fujii et al., "A frameshift variant in the EXT1 gene in a feline leukemia virus-negative cat with osteochondromatosis." Anim Genet 53(5):696-699.

Osteogenesis Imperfecta

Osteogenesis imperfecta is an inherited disease that causes brittle bones that break easily. It is also called brittle bone disease.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
OI	*CREB3L1*	c.370_371delTG	recessive	N/N	0	Non-carrier

Scientific references

2022 Takanosu et al., "Severe osteogenesis imperfecta caused by CREB3L1 mutation in a cat." J Vet Diag Invest 34(3):558-563.

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Musculoskeletal Disorders

Vitamin D-dependent Rickets, Type I

Vitamin D deficiency due to genetic causes can lead to rickets and skeletal problems in kittens.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
VDDR	*CYP27B1*	c.637G>T	recessive	N/N	0	Non-carrier

Scientific references

2012 Grahn et al., "A novel CYP27B1 mutation causes a feline vitamin D-dependent rickets type IA." J Feline Med Surg 14(8):587-90.

Spasticity (CMS) or Congenital Myasthenia Syndrome

Spasticity (or CMS) is an inherited neuromuscular disorder that causes weakness of the pelvic and shoulder muscles and a risk of choking due to problems with swallowing. Medical treatment is available.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
CMS	*COLQ*	c.1190G>A	recessive	N/N	0	Non-carrier

Breeds mainly concerned: Devon Rex, Sphynx

What can you observe before visiting the vet?

Symptoms appear between 3 and 23 weeks and can include muscle weakness, difficulty eating, joint problems, difficulty breathing, strabismus, and in severe cases, neurological disorders or seizures. Medical treatment is available.

Scientific references

2015 Abitbol et al., "A COLQ Missense Mutation in Sphynx and Devon Rex Cats with Congenital Myasthenic Syndrome." PLoS One 10(9):e0137019.
2015 Gandolfi et al., "COLQ variant associated with Devon Rex and Sphynx feline hereditary myopathy." Anim Genet 46(6):711-5.

Dermatological Disorders

Acrodermatitis Enteropathica

Acrodermatitis enteropathica is a hereditary form of zinc deficiency that primarily causes skin damage and diarrhea.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
AE	*SLC39A4*	c.1057G>C	recessive	N/N	0	Non-carrier

Breeds mainly concerned: Turkish Van

Scientific references

2021 Kiener et al., "A Missense Variant in SLC39A4 in a Litter of Turkish Van Cats with Acrodermatitis Enteropathica." BMC Vet Res 8:207.

Ehlers-Danlos Syndrome (EDS)

EDS is an inherited connective tissue disorder. Affected cats have overly elastic, thin skin that can form excessive folds around the face and is prone to lesions. They may also show gait abnormalities and pain when palpating the joints.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
EDS	*COL5A1*	c.3514A>T	dominant	N/N	0	Non-carrier

Breeds mainly affected: Bombay

Scientific references

2022 Kiener et al., "Independent COL5A1 Variants in Cats with Ehlers-Danlos Syndrome." BMC Vet Res 8:207.

Epidermolysis Bullosa Simplex

Epidermolysis bullosa affects the skin and mucous membranes and can lead to ulcers of the tongue and pads.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
EBS	*KRT14*	c.979C>T	recessive	N/N	0	Non-carrier

Scientific references

2020 Dettwiler et al., "A nonsense variant in the KRT14 gene in a domestic shorthair cat with epidermolysis bullosa simplex." Anim Genet 51(5):829-832.

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Dermatological Disorders

Hypotrichosis, DSG4-related

Cats with DSG4 gene mutations have large areas of hypotrichosis or alopecia. Hypotrichosis associated with the DSG4 gene is different from that of the Sphynx, Donskoy, and Peterbald cats.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Hyp	*DSG4*	c.76delA	recessive	N/N	0	Non-carrier
Hyp	*DSG4*	c.1777delC	recessive	N/N	0	Non-carrier

Scientific references
2020 Kiener et al., "Independent DSG4 frameshift variants in cats with hair shaft dystrophy." Mol Genet Genomics 297(1):147-154.

Inflammatory Linear Verrucous Epidermal Nevi

Symptoms of linear inflammatory verrucous epidermal nevus are patches of hyperpigmented, thick, erythematous skin with hair loss on the head, legs, and paw pads. The NSDHL gene is located on the X chromosome. Males with the mutation for this disease cannot survive. Females with one copy of the mutation are affected.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
ILVEN	*NSDHL*	c.397A>G	X-linked semi-dominant	N/N	0	Non-carrier

Scientific references
2019 De Lucia et al., "Genetic variant in the NSDHL gene in a cat with multiple congenital lesions resembling inflammatory linear verrucous epidermal nevi." Vet Dermatol 30(1):64-e18.

Developmental Morphology

Burmese Head Defect (BHD)

Some Burmese cats have been selected for particular facial characteristics (short nose, round head) determined by the ALX1 gene. Carriers of one copy of the variant have these facial characteristics, but homozygous carriers of two copies of the variant have severe malformations (frontonasal dysplasia) at birth and do not survive.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
BHD	*ALX1*	c.496delCTCTCAGGACTG	dominant	N/N	0	Non-carrier

Breeds mainly concerned: Burmese

What can you observe before visiting the vet?
Your cat has a rounded head with broad cheekbones. The nose is short with a broad bridge and there is difficulty with breathing.

Scientific references
2016 Lyons et al., "Aristaless-Like Homeobox protein 1 (ALX1) variant associated with craniofacial structure and frontonasal dysplasia in Burmese cats." Dev Biol 409(2):451-8.

Cerebral Dysgenesis

Cerebral dysgenesis is caused by abnormal brain development. Kittens with this disorder may have difficulty walking (ataxia, paresis) and may exhibit aggression, seizures, and sensory abnormalities later in life.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
CD	*PEA15*	c.176delA	recessive	N/N	0	Non-carrier

Scientific references
2020 Graff et al., "PEA15 loss of function and defective cerebral development in the domestic cat." PLoS Genet 16(12):e1008671.

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Coat Color & Pattern

Locus A - Agouti

The distribution of eumelanin (brown to black) and pheomelanin (yellow-orange) pigments along the hairs is determined by the ASIP gene. Agouti hairs (with bands of eumelanin and pheomelanin) are distinguished from non-agouti or solid hairs, which are colored by eumelanins. In orange cats, eumelanin is replaced by a red pigment. Disruption of the ASIP gene is a common reason for a solid black coat due to the absence of agouti hairs.
*** The phenotype may vary depending on other genetic factors.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Solid (a) non-Agouti	*ASIP*	c.122_123delCA	recessive	a/a	2	Solid *

Scientific references
2003 Eizirik et al., "Molecular genetics and evolution of melanism in the cat family." Current Biology 13:448-53.

Locus A - Agouti (Charcoal)

APb allele (Agouti of the Bengal Leopard Cat: Prionailurus bengalensis)

The charcoal modification is a coloration seen in Bengal cats that is characterized by a cape (dark dorsal stripe) and a mask (dark face). It is determined by the agouti variant of the Bengal Leopard Cat as well as the non-agouti variant of the domestic cat. *** Dominant with incomplete penetrance: Cats with the APb/a genotype should exhibit the Charcoal coloration.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Charcoal (APb)	*ASIP*	c.142T>C	dominant	A/A	0	Non-carrier

Breeds mainly affected: Bengal

Scientific references
2014 Gershony et al., "Who's behind that mask and cape? The Asian leopard cat's Agouti (ASIP) allele likely affects coat colour phenotype in the Bengal cat breed." Anim Genet 45:893-897.

Locus B - Chocolate

Eumelanin (dark pigment) color determined by the TYRP1 gene. *** Cats with the Chocolate B/b and Cinnamon B/bl genotypes will have a Chocolate coat and will be carriers of Cinnamon. If the cat is also homozygous for the Dilution variant (genotype d/d), then the Chocolate will be diluted to Lilac and the Cinnamon will be diluted to Fawn.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Chocolate (b)	*TYRP1*	c.1261+5G>A	recessive	B/B	0	Non-carrier
Cinnamon (bl)	*TYRP1*	c.298C>T	recessive	B/B	0	Non-carrier

Scientific references
2005 Lyons et al., "Chocolate coated cats: TYRP1 mutations for brown color in domestic cats." Mammalian Genome 16(5):356-66.
2005 Schmidt-Küntzel et al., " Tyrosinase and tyrosinase related protein 1 alleles specify domestic cat coat color phenotypes of the albino and brown loci." J Hered 96:289-301, 2005.

Locus C - Color (Albinism)

Albinism is a rare trait in cats, which occurs due to the lack of pigment production.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Albinism (c)	*TYR*	c.975delC	recessive	C/C	0	Non-carrier
Albinism (c2)	*TYR*	c.1204C>T	recessive	C/C	0	Non-carrier

Scientific references
2006 Imes et al., "Albinism in the domestic cat (Felis catus) is associated with a tyrosinase (TYR) mutation." Animal Genetics 37:175-8.
2017 Abitbol et al., "Allelic heterogeneity of albinism in the domestic cat." Animal Genetics 48(1):127-128.

ID number : 000 000 000 000 000
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Coat Color & Pattern

Locus C – Color (Colorpoint and Mink)

The colorpoint pattern is a characteristic inherited from Asian cats and characterized by a strong contrast in pigmentation on the cat's body: the color is darker at the extremities of the body. A less marked contrast is observed in Burmese while the Mink pattern presents a contrast intermediate between Siamese and Burmese.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Colorpoint Siamese (Point, cs)	*TYR*	p.G302R	recessive	C/cs	1	Carrier
Colorpoint Burmese (Sepia, cb)	*TYR*	c.715G>T	recessive	C/C	0	Non-carrier
Mink (cb & cs)	*TYR*	c.715G>T; p.G302R	recessive	-	-	-

Breeds mainly concerned: Burmese (Sepia), Siamese (Point), Tonkinese (Mink)

Scientific references

2005 Lyons et al., "Tyrosinase mutations associated with Siamese and Burmese patterns in the domestic cat (Felis catus)." Animal Genetics 36(2):119-26.
2019 Yu et al., "Mocha tyrosinase variant: a new flavour of cat coat coloration." Anim Genet 50(2):182-186.

Locus D - Dilution

The dilution of the coat color, determined by the MLPH gene, produces an overall lightening of the cat's shade. Black is diluted to Blue, Chocolate is diluted to Lilac, Cinnamon is diluted to Fawn. Red is diluted to Cream.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Dilution (d)	*MLPH*	c.83delT	recessive	d/d	2	Dilution

Scientific references

2006 Ishida et al., "A homozygous single-base deletion in MLPH causes the dilute coat color phenotype in the domestic cat." Genomics 88:698-705.

Locus E - Extension

Amber, Copal/Carnelian, and Russet color changes result in the gradual replacement of black pigment (eumelanin) with red pigment (pheomelanin) over time. *** In tabby cats, Copal is present in cats with the E/ec genotype, while Carnelian is present in cats with the ec/ec genotype.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Amber (e)	*MC1R*	c.250G>A	recessive	E/E	0	Non-carrier
Copal/Carnelian (ec)	*MC1R*	c.640_669del	recessive	E/E	0	Non-carrier
Russet (er)	*MC1R*	c.440_442del	recessive	E/E	0	Non-carrier

Breeds mainly affected: Burmese (Russet), Kurilian Bobtail (Copal/Carnelian), Norwegian Forest Cat (Amber)

Scientific references

2009 Peterschmitt et al., "Mutation in the melanocortin 1 receptor is associated with amber colour in the Norwegian Forest Cat." Animal Genetics 40(4):547-52.
2017 Gustafson et al., "Not another type of potato: MC1R and the russet coloration of Burmese cats." Animal Genetics 48(1):116-120.
2019 Abitbol et al., "Copal, a new MC1R allele in the domestic cat." Animal Genetics 50(5):553-554.

Locus G - Gloving

The gloving pattern refers to the presence of white feet, which is considered an essential criterion for the Birman. The gloving pattern is determined by the KIT gene.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Gloving (g)	*KIT*	c.1035_1036delinsCA	recessive	G/G	0	Non-carrier

Scientific references

2010 Lyons, "Feline genetics: clinical applications and genetic testing." Topics in Companion Animal Medicine 25(4):203-12.

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Coat Color & Pattern

Locus O - Orange

The X-linked Orange (O) variant suppresses the black pigment (eumelanin) while favoring orange coloration (pheomelanin). Since males have only one X chromosome, they are much more likely than females to be entirely Orange. *** Males with 1 copy of the Orange variant will be entirely Orange. In females, 1 copy results in a coat that is a mixture of black and orange, called Tortoiseshell or 'Tortie'. Females with 2 copies are less common and have an entirely Orange coat.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Orange (O)	*ARHGAP36*	deletion	X-linked	O/Y	1	Orange *

Scientific references
2025 Toh et al., "A deletion at the X-linked ARHGAP36 gene locus is associated with the orange coloration of tortoiseshell and calico cats." Curr Biol 35:2816-2825.e3, 2025.
2025 Kaelin et al., "Molecular and genetic characterization of sex-linked orange coat color in the domestic cat." Curr Biol 35:2826-2836.e5.

Locus Ta - Tabby Blotched

Tabby cats can have different dark patterns on their coats. Two genetic variants in the LVRN gene are associated with the conversion of stripes (mackerel) to swirls (blotched). *** It is possible to be blotched tabby with 1 copy of 2 different variants. The phenotype may vary depending on other genetic factors.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Blotched (Tab2)	*LVRN*	c.176C>A (S59X)	recessive	TaM/TaM	0	Non-carrier
Blotched (Tab3)	*LVRN*	c.2522G>A (W841X)	recessive	TaM/Tab3	1	Carrier

Scientific references
2010 Eizirik et al., "Defining and mapping mammalian coat pattern genes: multiple genomic regions implicated in domestic cat stripes and spots." Genetics 184(1):267-75.
2012 Kaelin et al., "Specifying and sustaining pigmentation patterns in domestic and wild cats." Science 337(6101):1536-41.

Locus Ta - Tabby Ticked

The ticked trait is determined by the DKK4 gene. Ticked tabbies have agouti hairs and are characterized by an absence or very significant reduction of dark markings on their coat.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Ticked (TiA)	*DKK4*	c.188G>A	dominant	Ti/Ti	0	Non-carrier
Ticked (TiCK)	*DKK4*	c.53C>T	dominant	Ti/Ti	0	Non-carrier

Scientific references
2021 Lyons et al., "Mining the 99 Lives ... Consortium database implicates genes and variants for the Ticked locus in domestic cats (Felis catus)." Animal Genetics 52(3):321-332.

Locus W - Dominant White

The solid white coat (Dominant White) is determined by an allele of the KIT gene. This trait is not the same as albinism, which is caused by a mutation in a different gene. Cats with the Dominant White trait have a higher risk of hearing loss.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Dominant White (W)	*KIT*	insertion	dominant	w/w	0	Non-carrier

Scientific references
2014 David et al., "Endogenous retrovirus insertion in the KIT oncogene determines white and white spotting in domestic cats." G3 4:1881-91, 2014.

Locus W - White Spotting

The presence of variegation (white patches in the coat, White Spotting) is determined by the KIT gene. Several patterns exist depending on the amount of white, for example van (> 75%), harlequin (50-70%) or bicolor (25-50%).

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
White Spotting (Ws)	*KIT*	insertion	dominant	Ws/w	1	White Spotting

Scientific references
2014 David et al., "Endogenous retrovirus insertion in the KIT oncogene determines white and white spotting in domestic cats." G3 4:1881-91, 2014.

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Coat Color & Pattern

Locus Wb - Wideband
vwb for variable-wideband

Sunshine, Extreme Sunshine, and Copper are coat color modifications determined by the CORIN gene. These are color variations known by various names, including "golden," and are found in tabby cats.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Copper (wbBSH)	*CORIN*	c.2425C>T	recessive	Wb/Wb	0	Non-carrier
Extreme Sunshine (wbeSIB)	*CORIN*	c.839G>A	recessive	Wb/Wb	0	Non-carrier
Sunshine (wbSIB)	*CORIN*	c.2383C>T	recessive	Wb/Wb	0	Non-carrier

Breeds mainly affected: British Shorthair (Copper), Siberian (Extreme Sunshine, Sunshine)

Scientific references
2021 Beauvois et al., “Siberian cats help in solving part of the mystery surrounding golden cats.” Animal Genetics 52(4):482-491.
2022 Abitbol et al., “Golden cats: The story goes on.” Animal Genetics 53(4):543-545.
2022 Abitbol et al., “Golden cats: A never-ending story!” Animal Genetics 53(5):715-718.

Glitter

Glitter is a characteristic that gives the coat a shiny, glittery sheen and a soft texture.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Glitter (gl)	*FGFR2*	insertion	recessive	N/N	0	Non-carrier

Breeds mainly affected: Bengal, Egyptian Mau, Toyger

Scientific references
2024 Kaelin et al., “Ancestry dynamics and trait selection in a designer cat breed.” Curr Biol 34(7):1506-1518.e7.

Coat Type

Lykoi Coat (hypotrichose et rouan)

Lykoi cats have a unique coat caused by a reduction in the number of hair follicles (a form of hypotrichosis) and a mixture of pigmented and unpigmented hairs that creates the roan effect. This characteristic is determined by the HR gene. At least six different alleles are involved. They were named according to their geographic origin.
* **It is possible to have the Lykoi appearance with 1 copy of 2 different variants.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Lykoi (France-Fr)	*HR*	c.1404+2delinsCAG	recessive	HR/HR	0	Non-carrier
Lykoi (Canada-Ca)	*HR*	c.2593C>T	recessive	HR/HR	0	Non-carrier
Lykoi (USA-NC)	*HR*	c.2243C>T	recessive	HR/HR	0	Non-carrier
Lykoi (USA-TN)	*HR*	c.1255_1256dup	recessive	HR/HR	0	Non-carrier
Lykoi (USA-TX)	*HR*	c.2112G>A	recessive	HR/HR	0	Non-carrier
Lykoi (USA-VA)	*HR*	c.3389insGACA	recessive	HR/HR	0	Non-carrier

Breeds mainly concerned: Lykoi

Scientific references
2020 Buckley et al., “Werewolf, There Wolf: Variants in Hairless Associated with Hypotrichia and Roaning in the Lykoi Cat Breed.” Genes 11(6):682.

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Coat Type

Locus L - Length (Long Hair)

The presence of medium to long hair in cats is determined by the FGF5 gene.
*** It is possible to have long hair with 1 copy of 2 different variants. Long hair can be very long or medium-long.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Long hair (M1)	*FGF5*	c.ins356T	recessive	N/N	0	Non-carrier
Poil long (M2)	*FGF5*	c.406C>T	recessive	N/N	0	Non-carrier
Long hair (M3)	*FGF5*	c.474delT	recessive	N/N	0	Non-carrier
Poil long (M4)	*FGF5*	c.475A>C	recessive	N/M4	1	Carrier
Long hair (M5)	*FGF5*	c.577G>A	recessive	N/N	0	Non-carrier

M1: Ragdoll
M2: Norwegian Forest Cat
M3: Maine Coon, Ragdoll
M4: Domestic Cats
M5: Maine Coon

Scientific references
2007 Kehler et al., "Four independent mutations in the feline fibroblast growth factor 5 gene determine the long-haired phenotype in domestic cats." J Hered 98(6):555-66.
2007 Drögemüller et al., "Mutations within the FGF5 gene are associated with hair length in cats." Animal Genetics 38(3):218-21.
2021 Shaffer et al., "Identification of a novel missense mutation in the fibroblast growth factor 5 gene associated with longhair in the Maine Coon Cat." Human Genetics 140(11):1517-1523.

Locus R - Nudity (Hairless, Hr)

Hairlessness in the Sphynx is determined by the KRT71 gene. *** Cats carrying 1 copy of Hairless and 1 copy of Devon Rex (hr/re genotype) also exhibit the Hairless phenotype.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Hairless (hr)	*KRT71*	c.816+1G>A	recessive	Hr/Hr	0	Non-carrier

Breeds mainly affected: Sphynx

Scientific references
2010 Gandolfi et al., "The naked truth: Sphynx and Devon Rex cat breed mutations in KRT71." Mamm Genome 21:509-15.

Locus R - Rexing (curly hair)

A curly coat (called Rexing) is determined by several different genes. *** The Selkirk Rex variant is dominant with incomplete penetrance: some cats carrying this variant may, however, have a coat with little or no curly hair.**

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Devon Rex (re)	*KRT71*	c.1108-4_1184 delinsAGTTGGAG	recessive	Re/Re	0	Non-carrier
Selkirk Rex (Se)	*KRT71*	c.445-1G>C	dominant*	se/se	0	Non-carrier
Cornish Rex (r)	*LPAR6*	c.250_253delTTTG	recessive	R/R	0	Non-carrier
Ural Rex (r)	*LIPH*	c.477_483del	recessive	R/R	0	Non-carrier

*** Dominant with incomplete penetrance**

Breeds mainly concerned: Devon Rex (re), Cornish Rex (r), Selkirk Rex (Se), Ural Rex (r)

Scientific references
2010 Gandolfi et al., "The naked truth: Sphynx and Devon Rex cat breed mutations in KRT71." Mamm Genome 21:509-15.
2013 Gandolfi et al., "A splice variant in KRT71 is associated with curly coat phenotype of Selkirk Rex cats." Sci Rep 3:2000.
2013 Gandolfi et al., "To the Root of the Curl: A Signature of a Recent Selective Sweep Identifies a Mutation That Defines the Cornish Rex Cat Breed." PLoS One 8(6):e67105.
2020 Manakhov et al., "The curly coat phenotype of the Ural Rex feline breed is associated with a mutation in the lipase H gene." Anim Genet 51:584-9.

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Eye Color

Dominant Blue Eyes (DBE)

The Dominant Blue Eyes (DBE) trait results from several variants located in the PAX3 gene. DBE is characterized by the presence of one or two blue or parti-colored eyes (only part of the eye is blue) and reduced white markings. It is a dominant trait that can be lethal in the homozygous state (when the individual carries two mutated copies of the PAX3 gene). DBE can also be accompanied by deafness in some individuals. Furthermore, some cats carrying a DBE mutation may not express it in their coat and eyes (non-blue eyes and a coat without white markings). These cats are called "latent."

For professional breeders, recommendations are as follows :

1) Do not mate two DBE cats, regardless of their lineage.
2) Screen DBE cats for deafness and exclude deaf cats, even those deaf in only one ear, from breeding.
3) Test non-DBE breeding cats from a DBE line for DBE using DNA testing to detect "latent" carriers.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
DBE-RE	*PAX3*	c.937C>T	dominant	N/N	0	Non-carrier
DBE-ALT	*PAX3*	insertion (RD-114)	dominant	N/N	0	Non-carrier
DBE-CEL	*PAX3*	insertion (FERV1-LTR)	dominant	N/N	0	Non-carrier
DBE-AGO	*PAX3*	c.160delC	dominant	N/N	0	Non-carrier

Breeds mainly concerned :
DBE-RE : Maine Coon
DBE-ALT : Altai, British Longhair, British Shorthair, Persian, Ragdoll, Sphynx
DBE-CEL : Celestial, Maine Coon, Siberian
DBE-AGO : Maine Coon

Scientific references
2024 Garces et al., "PAX3 haploinsufficiency in Maine Coon cats with dominant blue eyes and hearing loss resembling the human Waardenburg syndrome." G3 14:9.
2024 Abitbol et al., "Different Founding Effects Underlie Dominant Blue Eyes (DBE) in the Domestic Cat." Animals 14:1845.
2024 Abitbol et al., "A PAX3 insertion in the Celestial breed and certain feline breeding lines with dominant blue eyes." Animal Genetics 55:670–675.

Pink-eye (Incomplete Oculocutaneous Albinism)

Pink eye, determined by the HPS5 gene, is a form of incomplete oculocutaneous albinism characterized by marked lightening of the skin and hair and yellow-green eyes with a red cast.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Pink-eye	*HPS5*	c.2571-1G>A	recessive	N/N	0	Non-carrier

Breeds mainly concerned : Donskoy

Scientific references
2020 Mériot et al., "Donskoy cats as a new model of oculocutaneous albinism with the identification of a splice-site variant in Hermansky-Pudlak Syndrome 5 gene." Pigment Cell Melanoma Research 33:814-825.

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Body Morphology

Folded Ears

Folded ears are determined by the TRPV4 gene. Cats carrying two copies of the variant (homozygous mutated) have osteoarticular abnormalities.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Fold (Fd)	*TRPV4*	c.1024G>T	dominant	N/N	0	Non-carrier

Breeds mainly affected: Highland Fold, Scottish Fold

Scientific references

2016 Gandolfi et al., "A dominant TRPV4 variant underlies osteochondrodysplasia in Scottish fold cats." Osteoarthritis Cartilage 24(8):1441-50.

Polydactyly

Non-syndromic polydactyly is the presence of extra toes associated with no health problems. Variants were originally identified in Europe (UK1 and UK2) and North America (Hw, Hemingway). These three variants can be found worldwide. Polydactyly variants in some cats in France have not yet been identified.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Polydactyly (UK1)	*LMBR1*	c.257G>C	dominant	N/N	0	Non-carrier
Polydactyly (UK2)	*LMBR1*	c.481A>T	dominant	N/N	0	Non-carrier
Polydactyly (Hw)	*LMBR1*	c.479A>G	dominant	N/N	0	Non-carrier

Breeds mainly affected: Domestic Cats (UK1, UK2, Hw), Pixie Bob (Hw), Maine Coon (Hw)

Scientific references

2008 Lettice et al., "Point mutations in a distant sonic hedgehog cis-regulator generate a variable regulatory output responsible for preaxial polydactyly." Hum Mol Genet 17(7):978-85.

Short tail (Bobtail)

A shortened tail can be determined by the HES7 and T genes.

Phenotype	Gene	Variant(s)	Mode of Inheritance	Genotype	Copies	Result
Bobtail (JBT)	*HES7*	c.5A>G	dominant	N/N	0	Non-carrier
Bobtail (Mx1)	*T*	c.998delT	dominant	N/N	0	Non-carrier
Bobtail (Mx2)	*T*	c.998_1014dup17delGCC	dominant	N/N	0	Non-carrier
Bobtail (Mx3)	*T*	c.1169delC	dominant	N/N	0	Non-carrier
Bobtail (Mx4)	*T*	c.1199delC	dominant	N/N	0	Non-carrier

Breeds mainly affected: Japanese Bobtail (JBT), American Bobtail (Mx1-4), Manx (Mx1-4), Pixie Bob (Mx1-4)

Scientific references

2013 Buckingham et al., "Multiple mutant T alleles cause haploinsufficiency of Brachyury and short tails in Manx cats." Mammalian Genome 24(9-10):400-8.
2016 Lyons et al., "Whole genome sequencing in cats, identifies new models for blindness in AIPL1 and somite segmentation in HES7." BMC Genomics 17:265.

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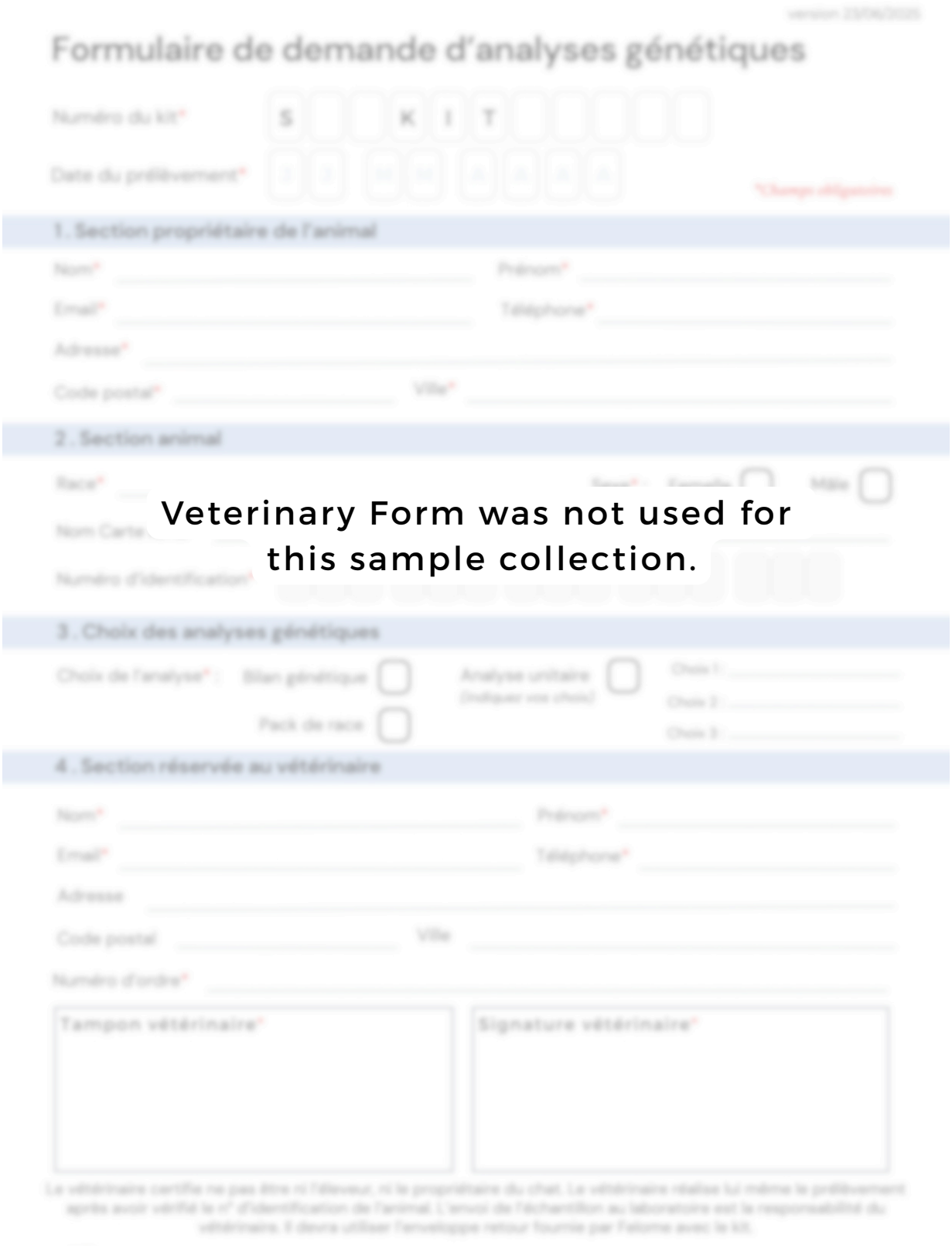
Veterinary Form



Veterinary Form was not used for this sample collection.