Your Health Marker Report

Kitty Genetics One-On-One

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, therefore, heterozygous for this gene. On the contrary, 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 to the phenotype equally. If the two alleles are different however, which allele will contribute to the phenotype depends on their relationship. Some alleles are dominant, meaning that they have the ability to "hide" other alleles and thus be 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, i.e., there is no dominant allele to hijack the phenotype expression.

How Does It All Relate To Disease?

Genetic disorders are conditions that are present at birth or develop later in life and are caused by one or more genetic mutations. We tested your furry friend for genetic health markers associated with genetic disorders. Genetic health markers are mutated gene alleles associated with an increased likelihood of developing a particular genetic disorder. With the exception of hypertrophic cardiomyopathy (HCM), which has more complex genetics, the conditions included in this report have either a dominant or a recessive pattern of inheritance. This means that for diseases with a dominant inheritance pattern, having just one mutated gene allele will result in the cat developing the disease. Conversely, for diseases with a recessive inheritance pattern, the cat will only develop the disease if it has 2 mutated alleles. Having just one mutated recessive allele makes the cat a disease carrier, meaning that it will not develop the disease, but it can pass down the mutation to its kittens.

In this part of the report, you will see your kitty's results for various genetic markers associated with diseases. Not all disease may be available right away -- make sure to check your report often in the coming weeks as more reports and markers are added. 'Clear' status indicates that your cat tested negative for a particular genetic marker. 'Carrier' status means your cat has one copy of a recessive genetic marker and should only be a concern if you plan to breed your cat. 'At risk' and 'At high risk' status means that your cat has tested positive for a dominant allelic mutation or has two copies of a recessive allelic mutation. If you see one of these two result designations, contact your veterinarian.

Please note, Basepaws results should not replace evaluation and clinical diagnosis made by a veterinarian. We also want to point out that a 'Clear' result does not mean your cat is guaranteed to not develop the disease. It simply means your cat is negative for the mutation we tested. There may be environmental factors and other not yet known genetic mutations contributing to developing the disease. If there is a positive result, please make sure to notify your veterinarian.

Health Markers

In this section, you will find a brief description for each of the genetic diseases you currently have results for. Each of these disease is represented by at least one known health marker. In addition to your cat's results, we have also included details on the genes and genetic mutations included in our test.

Clear – The cat is negative for the disease-associated marker we tested
Carrier – The cat has one copy of an autosomal recessive disease-associated marker
At Risk – The cat has one copy of a marker associated with hypertrophic cardiomyopathy
At High Risk– This designation can mean one of three things:

The cat has 1 or 2 copies of a marker associated with an autosomal dominant disease
The cat has 2 copies of a marker associated with hypertrophic cardiomyopathy
The cat has 2 copies of an autosomal recessive disease-associated marker

IMPORTANT: Not all health reports might be available initially, and more markers and results can be added over the coming weeks and even months! Stay close to your results and check often to see any new health markers and diseases added.

Polycystic kidney disease

Polycystic kidney disease (PKD) is the most common genetic disease in cats. PKD is characterized by the formation of small fluid-filled cysts in the kidneys that lead to kidney failure.

Gene	Mutation	Result	
PKD1	C>A	Negative	Clear

Cardiomyopathy, hypertrophic

Hypertrophic cardiomyopathy (HCM) is the most common feline heart disease characterized by tachycardia.

Gene	Mutation	Result	
MYPBC3	G>A *Frequent in Rag Doll	Negative	Clear
MYPBC3	C>G *Frequent in Maine Coon	Negative	Clear

Health Markers

Retinal degeneration II

Progressive retinal atrophy (PRA) is a disease marked by the deterioration of retina caused by the progressive death of retinal cells.

Gene	Mutation	Result	
CEP290	A>C	Negative	Clear

Mucopolysaccharidosis

Mucopolysaccharidoses are a group of metabolic disorders characterized by a deficiency in the production or functioning in lysosomal enzymes required for digestion of glycosaminoglycans (GAGs).

Gene	Mutation	Result	
ARSB	A>G *Type VI	Negative	Clear
IDUA	GTC>del *Type I	Negative	Clear
IDUA	TCG>del *Type I	Negative	Clear
GUSB	G>A *Type VII	Negative	Clear
GUSB	T>G *Type VII	Negative	Clear
GUSB	C>T *Type VII	Negative	Clear

Health Markers

Gangliosidosis

Gangliosidosis is a group of lipid storage disorders characterized by the accumulation of lipids – gangliosides in neurons. GM2AB gangliosidosis (type AB) is associated with a deficiency in beta hexosaminidases A and B.

Gene	Mutation	Result	
HEXB	C>T *GM2 Type II	Negative	Clear
HEXB	T>del *GM2 Type II	Negative	Clear
HEXB	TAC..>inv *GM2 Type II	Negative	Clear
GM2A	GACC>del *GM2, GM2A deficiency	Negative	Clear
GLB1	C>G *GM1	Negative	Clear

Health Markers

Cystinuria

Cystinuria is an inherited metabolic disease, relatively common in dogs and rare in cats, associated with high cysteine levels in urine.

Gene	Mutation	Result	
SLC3A1	C>T *Type I-A	Negative	Clear
SLC7A9	G>A *Type B	Negative	Clear
SLC7A9	C>T *Type B	Negative	Clear

Health Markers

Porphyria

Porphyria is a group of diseases associated with the accumulation of porphyrins. The buildup of porphyrins in the acute diseases primarily affect the nervous system.

Gene	Mutation	Result	
HMBS	ACAG>del *Acute intermittent	Negative	Clear
HMBS	T>ins *Acute intermittent	Negative	Clear
HMBS	G>A *Acute intermittent	Negative	Clear
HMBS	C>T *Acute intermittent	Negative	Clear
HMBS	G>A *Acute intermittent	Negative	Clear
HMBS	GAG>del *Acute intermittent	Negative	Clear
UROS	C>T *Congenital erythropoietic	Negative	Clear
UROS	G>A *Congenital erythropoietic	Negative	Clear

Health Markers

Factor XII deficiency

Factor XII deficiency, or Hageman deficiency, is a blood clotting disorder characterized by deficiency in the coagulation factor XII.

Gene	Mutation	Result	
F12	C>del	Negative	Clear
F12	G>C	Positive (1 copy)	Carrier

Mannosidosis, alpha

Alpha mannosidosis is a lysosomal storage disorder characterized by the deficiency of the alpha-D-mannosidase enzyme. A defective alpha-mannosidase causes progressive accumulation of mannose-rich oligosaccharides in all tissues, which subsequently disrupts the cellular functions and causes apoptosis.

Gene	Mutation	Result	
MAN2B1	CTGG>del	Negative	Clear

Myotonia

Myotonia Congenita (MC) is a hereditary neuromuscular disorder characterized by persistent contraction (or delayed relaxation of muscles), particularly during the muscle movement.

Gene	Mutation	Result	
CLCN1	G>T	Negative	Clear

Health Markers

Hypothyroidism

Feline Conginetal Hypothyroidism with Goiter is a rare autosomal recessive disease that that affects the thyroid levels in the blood.

Gene	Mutation	Result	
TPO	C>T	Negative	Clear

Vitamin D-deficiency rickets, type I

Rickets or osteomalacia is a disease associated with the softening of bones and increased rate of bone deformities and fractures.

Gene	Mutation	Result	
CYP27B1	C>del	Negative	Clear
CYP27B1	C>A	Negative	Clear

Niemann-Pick disease, type C1

Niemann-Pick disease is a group of hereditary lysosomal storage diseases. Feline Niemann-Pick disease C coincides with the human type C of this disorder, and it is classified in two subtypes: C1 and C2.

Gene	Mutation	Result	
NPC1	C>G	Negative	Clear
NPC1	T>G	Negative	Clear

Health Markers

Hypokalaemic periodic paralysis

Hypokalemia refers to the state of low potassium ion (K+) levels in the blood. It often arises as a secondary problem due to other deficiencies or diseases, but it may also be a result of a primary congenital disease, such as hypokalemic period polymyopathy.

Gene	Mutation	Result	
WNK4	C>T	Negative	Clear

Autoimmune lymphoproliferative syndrome

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

Gene	Mutation	Result	
FASLG	A>ins	Negative	Clear

Pyruvate kinase deficiency of erythrocyte

Pyruvate kinase deficiency is an inherited metabolic disorder characterized by disrupted survival of erythrocytes (red blood cells).

Gene	Mutation	Result	
PKLR	G>A	Negative	Clear