

Breed: Ragdoll  
Birth date: 2022-05-02

Test date: 2024-02-05  
ID kit: FCYBWPZ

MINNA’s Profile

Pet information

|                      |               |
|----------------------|---------------|
| Registered name      | Sex           |
| MINNA                | F             |
| Owner reported breed | Date of birth |
| Ragdoll              | 2022-05-02    |

Genetic Diversity

MINNA’s Percentage of Heterozygosity  
33%

Health summary

- At Risk 0 conditions
- Carrier 0 conditions
- Clear 50 conditions

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## Genetic Diversity

### Heterozygosity

#### MINNA's Percentage of Heterozygosity

33%

MINNA's genome analysis shows an average level of genetic heterozygosity when compared with other Ragdolls.

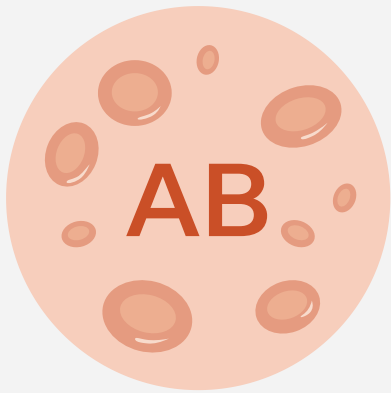
#### Typical Range for Ragdolls

32% - 37%

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Blood Type



**Blood type**  
Type AB (Very rare)

**Genotype\***  
c/b (Carrier for Blood Type B)

**Transfusion risk**  
✔ Low  
MINNA has a rare blood type. She can be transfused with Type A or Type AB blood.

**Breeding risk**  
✔ Low  
If breeding, MINNA has a low risk of blood type incompatibility with nursing kittens.

Blood variants tested\*

| Variant Tested                   | Description                    | Copies |
|----------------------------------|--------------------------------|--------|
| b variant 1                      | (Common b variant)             | 1      |
| b variant 2                      | (Discovered in Turkish breeds) | 0      |
| b variant 3                      | (Discovered in Ragdolls)       | 0      |
| c variant - Causes AB Blood Type | (Discovered in Ragdolls)       | 1      |

\*This test identifies three known 'b' variants and one known 'c' variant in the CMAH gene when determining a cat's genetic blood type. Blood Type A is inferred in reporting when less than two genetic blood variants are detected.

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## Interpreting feline blood types

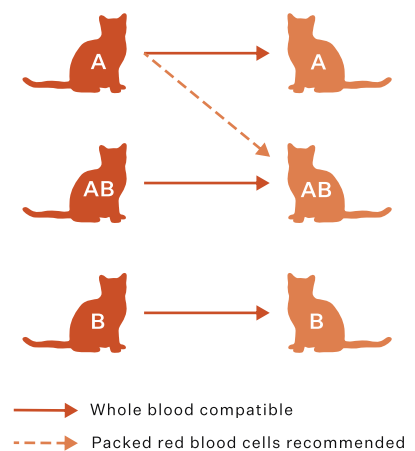
### About blood type determination

The three important feline blood types of A, B, and AB are governed primarily by variants in the CMAH gene. A cat's blood type can be determined by its genotype, which consists of two gene variants – one inherited from each parent – that should be interpreted together. When determining blood type based on genotype, the A variant associated with blood type A is most dominant while the b variants associated with blood type B are most recessive. The c variant associated with blood type AB is intermediate between the A and b variants, meaning it is recessive to the A variant but dominant to b variants. Therefore, a genotype with at least one A variant will result in blood type A. For a cat to have blood type B, the genotype must consist of two b variants. Because the c variant is intermediate, a cat with blood type AB can either have a genotype consisting of two c variants or one c variant and one b variant.

### About transfusion risk

Similar to humans, the different cat blood types will express different antigens on the surface of their red blood cells. This is significant because both type A and B cats are born with antibodies against other blood cell antigens. Notably, type B cats have high levels of antibodies against type A antigens. Cats with the rare blood type AB are most versatile as they express both red cell antigen types and, thus, can receive both type A and type AB blood transfusions.

Unlike humans, there is no cat blood type that can act as a universal blood donor. If a cat receives a non-compatible blood type during a transfusion, it may cause a severe, life-threatening reaction including fever, kidney failure, and widespread destruction of red blood cells. Prior to all transfusions, cats should be serologically typed and crossmatched to ensure compatibility.



### About breeding risk

During pregnancy, kittens are shielded from their mother's immune system. However, when kittens begin nursing, they receive some of their mother's antibodies in colostrum. Type B cats have high levels of antibodies against type A blood, so when blood type A or AB kittens are born to a blood type B mother, these antibodies, when absorbed by the newborn kitten, cause neonatal isoerythrolysis, a potentially fatal destruction of the kitten's red blood cells. Kittens of type B mothers with fathers of unknown or type A blood should be bottle fed or foster-nursed, and separated from their mother for the first 24 hours to avoid this reaction, unless blood typing performed immediately following birth shows the kitten to have a compatible blood type to the mother.

Although some blood types are less common and require additional planning when breeding, they represent normal genetic variation and should not be selected against when choosing breeding pairs.

### Current limits of this test

This test identifies 4 variants ( b variants c.269T>A, c.179G>T, c.1233delT and c variant c.346C>T) in the CMAH gene discovered in the domestic cat population and has been confirmed 99% concordant with serologic blood typing<sup>1</sup>. Mik antigens also play a role in blood type compatibility, and are not included in this test. Cats carrying undetermined, new, or undiscovered variants in CMAH or other genes may have a different blood type compatibility than that reported by this test. Accuracy of this test at predicting blood type in wildcats or wildcat hybrid breeds has not been determined.

1. Anderson H, Davison S, Lytle KM, Honkanen L, et al. Genetic epidemiology of blood type, disease and trait variants, and genome-wide genetic diversity in over 11,000 domestic cats (2022) PLOS Genetics.

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## Health conditions known in the breed

| Factor XII Deficiency (Variant 1) | Gene | Risk Variant | Copies | Inheritance | Result |
|-----------------------------------|------|--------------|--------|-------------|--------|
|                                   | F12  | Deletion     | 0      | ARa         | Clear  |

### Information about the genetic condition

Blood coagulation is a complex process involving many pathways. Factor XII, a plasma protein, classically initiates the intrinsic pathway of blood coagulation; although, there are alternative, slower ways to initiate this pathway. Factor XII Deficiency, also known as Hageman Factor Deficiency or Hageman trait, is a commonly inherited blood clotting disorder in cats. Unlike other bleeding disorders, cats deficient in Factor XII are asymptomatic and do not tend to show spontaneous bleeding or abnormal bleeding after surgery or trauma. However, affected individuals can have prolonged clotting time on the activated partial thromboplastin time (aPTT) screening test. Cats who inherit 2 copies of both Factor XII Deficiency (Variant 1) and Factor XII Deficiency (Variant 2) may show even higher aPTT values. Please note that 1 copy of Factor XII Deficiency (Variant 1) and 1 copy of Factor XII Deficiency (Variant 2) will not cause Factor XII Deficiency.

### Breeder recommendation

This condition is autosomal recessive, asymptomatic, meaning that cats with two copies of the variant will show the variant-associated condition but will not suffer disease due to this genetic cause. Current understanding is that a cat with one or two copies of the Factor XII Deficiency variant can be safely bred with a cat with zero, one or two copies of the variant. Please note: It is possible that clinical signs similar to the ones caused by the Factor XII Deficiency mutation could develop due to a different genetic or clinical cause.

| Factor XII Deficiency (Variant 2) | Gene | Risk Variant | Copies | Inheritance | Result |
|-----------------------------------|------|--------------|--------|-------------|--------|
|                                   | F12  | Deletion     | 0      | ARa         | Clear  |

### Information about the genetic condition

Blood coagulation is a complex process involving many pathways. Factor XII, a plasma protein, classically initiates the intrinsic pathway of blood coagulation; although, there are alternative, slower ways to initiate this pathway. Factor XII Deficiency, also known as Hageman Factor Deficiency or Hageman trait, is a commonly inherited blood clotting disorder in cats. Unlike other bleeding disorders, cats deficient in Factor XII are asymptomatic and do not tend to show spontaneous bleeding or abnormal bleeding after surgery or trauma. However, affected individuals can have prolonged clotting time on the activated partial thromboplastin time (aPTT) screening test. Cats who inherit 2 copies of both Factor XII Deficiency (Variant 1) and Factor XII Deficiency (Variant 2) may show even higher aPTT values. Please note that 1 copy of Factor XII Deficiency (Variant 1) and 1 copy of Factor XII Deficiency (Variant 2) will not cause Factor XII Deficiency.

### Breeder recommendation

This condition is autosomal recessive, asymptomatic, meaning that cats with two copies of the variant will show the variant-associated condition but will not suffer disease due to this genetic cause. Current understanding is that a cat with one or two copies of the Factor XII Deficiency variant can be safely bred with a cat with zero, one or two copies of the variant. Please note: It is possible that clinical signs similar to the ones caused by the Factor XII Deficiency mutation could develop due to a different genetic or clinical cause.

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## Health conditions known in the breed

| Hypertrophic Cardiomyopathy (Discovered in the Ragdoll) | Gene  | Risk Variant | Copies | Inheritance | Result |
|---------------------------------------------------------|-------|--------------|--------|-------------|--------|
|                                                         | MYBPC | C>T          | 0      | AD          | Clear  |

### Information about the genetic condition

Hypertrophic Cardiomyopathy (HCM) is the most common cardiac disease in cats worldwide. The disorder is characterized by an increase in wall thickness of the left ventricle and the interventricular septum. This causes turbulence in blood flow and increased venous pressure in the left atrium and lungs, which may or may not coincide with a cardiac murmur upon auscultation. The clinical signs most commonly noted with the disease are respiratory signs associated with congestive heart failure such as tachypnea, exercise intolerance and panting, difficulty breathing, and (rarely) coughing. Thromboembolism may also occur and affected cats have an increased risk of sudden cardiac death. Clinical and echocardiographic signs classically appear after the cat has reached breeding age, as the most common age of diagnosis for HCM is five to seven years. Specifically in Ragdoll cats, however, the disease has an early onset with an average age of diagnosis being only fifteen months. In the Ragdoll, a point mutation (R820W) in the MYBPC3 gene has been found as causative for the disease. Cats with two copies of the mutation (homozygotes) have earlier onset and increased severity of the disease compared to those with only one copy of the mutation (heterozygotes). The mode of inheritance is autosomal dominant with incomplete penetrance.

### Breeder recommendation

This disease is autosomal dominant with incomplete penetrance meaning that one copy of the variant is needed for disease signs to occur, but the relative risk of disease development in cats with two copies of the variant is significantly higher than that for cats with one copy of the variant. Due to the high frequency of the HCM variant within the Ragdoll breed, cats with one copy of the disease variant can be considered for breeding with cats tested clear for the HCM variant in order to maintain genetic diversity within the breed. Approximately half of the kittens in a litter from this breeding will have no copies of the HCM variant and half will have one copy of the HCM variant with a relatively low risk of disease development. Please note: It is possible that disease signs similar to the ones caused by the HCM variant could develop due to a different genetic or clinical cause.

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## Health conditions known in the breed

| MDR1 Medication Sensitivity | Gene  | Risk Variant | Copies | Inheritance | Result |
|-----------------------------|-------|--------------|--------|-------------|--------|
|                             | ABCB1 | Deletion     | 0      | AR          | Clear  |

### Information about the genetic condition

Cats with this variant are asymptomatic until exposed to a medication that uses the drug transport pump rendered defective by the mutation in the MDR1 (also called ABCB1) gene. Drugs known to use this P-glycoprotein pump are macrocyclic lactones including eprinomectin-containing products labeled for use in cats (antiparasitic drugs), loperamide (antidiarrheal), erythromycin (antibiotic), acepromazine (tranquilizer), butorphanol (opioid), certain drugs used in cancer treatment (vincristine, vinblastine, doxorubicin), and possibly others still to be determined. When these medications are administered, they accumulate in the brain which results in adverse reactions. Typical symptoms involve generalized neurologic dysfunction which may include mydriasis, dyspnea, tremors, hyperreactivity, ataxia or paresis. In more severe cases cats may experience seizures, coma and death. However, with appropriate supportive care by a veterinarian, most affected cats may be able to fully recover.

### Breeder recommendation

Further research is needed to determine if cats with one copy of the variant may have altered drug responses. At this time, breeding cats with one or two copies of the MDR1 Medication Sensitivity variant should be approached with caution. If a cat with one copy of the MDR1 Medication Sensitivity variant is bred with a clear cat with no copies of the MDR1 Medication Sensitivity variant, on average half of the kittens will have one copy and half will have no copies of the MDR1 Medication Sensitivity variant. If a cat with two copies of the MDR1 Medication Sensitivity variant is bred with a clear cat with no copies of the MDR1 Medication Sensitivity variant, the resulting kittens will all have one copy of the MDR1 Medication Sensitivity variant. If litters are expected to contain kittens with the MDR1 Medication Sensitivity variant, the kittens should be DNA tested as they may show signs of sensitivity to some common medications. Carrier to carrier matings are not advised as the resulting litter may contain kittens with two copies of the MDR1 Medication Sensitivity variant, which is known to cause medication sensitivity. Please note: It is possible that clinical signs similar to the ones caused by the MDR1 Medication Sensitivity variant could develop due to a different genetic or clinical cause.

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## Health conditions known in the breed

| Polycystic Kidney Disease (PKD) | Gene | Risk Variant | Copies | Inheritance | Result |
|---------------------------------|------|--------------|--------|-------------|--------|
|                                 | PKD1 | C>A          | 0      | AD          | Clear  |

### Information about the genetic condition

Polycystic Kidney Disease (PKD), also named autosomal dominant PKD, is characterized by variously sized, fluid-filled cysts in the renal cortex and medulla with hepatic and pancreatic cysts also possible. The cysts develop from birth and enlarge with age. The cysts destroy the renal parenchyma and disturb renal function, eventually causing renal failure. Affected cats present with signs of renal insufficiency such as weight loss, decreased appetite, increased drinking and urination, poor body condition, and vomiting. Biochemical labwork and ultrasonography examination are helpful tools in identifying the severity of disease within an affected individual. An autosomal dominant point mutation in the PKD1 gene has been identified as the most common genetic mutation for the disease. No homozygous cats have been identified, suggesting the mutation is a homozygous lethal mutation in utero. PKD is very common in Persian and Persian-related cats, affecting approximately 38% of Persian cats worldwide. While there is no known sex linkage to the inheritance of the mutation, research has shown male cats have a higher prevalence of the mutation.

### Breeder recommendation

This disease is autosomal dominant meaning that one copy of the mutation is needed for disease signs to occur. Use of cats with one or two copies of the disease mutation is not recommended, as there is a risk that the resulting litter will contain affected kittens. For example if a cat with one copy of the PKD mutation is bred with a clear cat with no copies of the PKD mutation, about half of the kittens will have one copy and half will have no copies of the PKD mutation. Please note: It is possible that disease signs similar to the ones caused by the PKD mutation could develop due to a different genetic or clinical cause.



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## Traits

### Coat Color

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Gene | Variant             | Copies | Result                              |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|---------------------|--------|-------------------------------------|
| <b>Charcoal (Discovered in the Bengal)</b>                                                                                                                                                                                                                                                                                                                                                                                                                          | ASIP | A <sup>Pb</sup>     | 0      | No effect                           |
| <b>Solid Color</b><br>Two copies of the Solid Color variant are needed for a cat to have solid colored hair. However, orange coloration overrides this effect, meaning that cats with partial or full orange coats can show tabby patterning in orange areas. Cats with zero or one copy of this variant are likely to have a tabby pattern due to color banding of the hairs.                                                                                      | ASIP | a                   | 1      | Banded hairs, tabby patterns likely |
| <b>Gloving (Discovered in the Birman)</b>                                                                                                                                                                                                                                                                                                                                                                                                                           | KIT  | w <sup>g</sup>      | 0      | No effect                           |
| <b>Partial and Full White</b><br>One or two copies of this variant will cause a part white or a full white appearance with blue coloration of one or both eyes possible.                                                                                                                                                                                                                                                                                            | KIT  | W or w <sup>s</sup> | 2      | Partly or fully white coat likely   |
| <b>Amber (Discovered in the Norwegian Forest Cat)</b>                                                                                                                                                                                                                                                                                                                                                                                                               | MC1R | e                   | 0      | No effect                           |
| <b>Russet (Discovered in the Burmese)</b>                                                                                                                                                                                                                                                                                                                                                                                                                           | MC1R | e <sup>r</sup>      | 0      | No effect                           |
| <b>Dilution</b><br>Two copies of the Dilution variant are required to have a lightening effect on the coat.                                                                                                                                                                                                                                                                                                                                                         | MLPH | d                   | 2      | Lightened coat color likely         |
| <b>Albinism (Discovered in Oriental breeds)</b>                                                                                                                                                                                                                                                                                                                                                                                                                     | TYR  | c <sup>a</sup>      | 0      | No effect                           |
| <b>Colorpoint (Discovered in the Burmese)</b>                                                                                                                                                                                                                                                                                                                                                                                                                       | TYR  | c <sup>b</sup>      | 0      | No effect                           |
| <b>Colorpoint (Discovered in the Siamese)</b><br>Two copies of this variant result in a colorpoint pattern, although this can be blocked by other variants. Cats with one copy of the Colorpoint (Discovered in the Burmese) variant and one copy of the Colorpoint (Discovered in the Siamese) variant will show a darker base coat color and less contrasting colorpoint pattern than cats with two copies of the Colorpoint (Discovered in the Siamese) variant. | TYR  | c <sup>s</sup>      | 2      | Siamese colorpoint pattern likely   |
| <b>Mocha (Discovered in the Burmese)</b>                                                                                                                                                                                                                                                                                                                                                                                                                            | TYR  | c <sup>m</sup>      | 0      | No effect                           |
| <b>Chocolate</b>                                                                                                                                                                                                                                                                                                                                                                                                                                                    | TYRP | b                   | 0      | No effect                           |
| <b>Cinnamon</b>                                                                                                                                                                                                                                                                                                                                                                                                                                                     | TYRP | b <sup>l</sup>      | 0      | No effect                           |

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## Coat Type

|                                                                                                                                                                                                                                                                               | Gene    | Variant          | Copies | Result                                |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------|------------------|--------|---------------------------------------|
| <b>Long Hair (Discovered in many breeds)</b>                                                                                                                                                                                                                                  | FGF5    | M4               | 0      | No effect                             |
| <b>Long Hair (Discovered in the Norwegian Forest Cat)</b>                                                                                                                                                                                                                     | FGF5    | M2               | 0      | No effect                             |
| <b>Long Hair (Discovered in the Ragdoll and Maine Coon)</b><br>Two copies of any Long Hair variant must be inherited for a cat to have a long coat. This can either be two copies of a particular variant, such as this one, or two of any combination of Long Hair variants. | FGF5    | M3               | 1      | Long coat possible, short coat likely |
| <b>Long Hair (Discovered in the Ragdoll)</b><br>Two copies of any Long Hair variant must be inherited for a cat to have a long coat. This can either be two copies of a particular variant, such as this one, or two of any combination of Long Hair variants.                | FGF5    | M1               | 1      | Long coat possible, short coat likely |
| <b>Lykoi Coat (Variant 1)</b>                                                                                                                                                                                                                                                 | HR      | hr <sup>Ca</sup> | 0      | No effect                             |
| <b>Lykoi Coat (Variant 2)</b>                                                                                                                                                                                                                                                 | HR      | hr <sup>VA</sup> | 0      | No effect                             |
| <b>Hairlessness (Discovered in the Sphynx)</b>                                                                                                                                                                                                                                | KRT71   | re <sup>hr</sup> | 0      | No effect                             |
| <b>Rexing (Discovered in the Devon Rex)</b>                                                                                                                                                                                                                                   | KRT71   | re <sup>dr</sup> | 0      | No effect                             |
| <b>Rexing (Discovered in the Cornish Rex and German Rex)</b>                                                                                                                                                                                                                  | LPAR6   | r                | 0      | No effect                             |
| <b>Glitter</b>                                                                                                                                                                                                                                                                | Pending | gl               | 0      | No effect                             |

## Tail Length

|                               | Gene | Variant  | Copies | Result    |
|-------------------------------|------|----------|--------|-----------|
| <b>Short Tail (Variant 3)</b> | HES7 | jb       | 0      | No effect |
| <b>Short Tail (Variant 1)</b> | T    | C1199del | 0      | No effect |
| <b>Short Tail (Variant 2)</b> | T    | T988del  | 0      | No effect |

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Extra Toes

|                         | Gene   | Variant | Copies | Result    |
|-------------------------|--------|---------|--------|-----------|
| Polydactyly (Variant 1) | LIMBR1 | HW      | 0      | No effect |
| Polydactyly (Variant 2) | LIMBR1 | UK1     | 0      | No effect |
| Polydactyly (Variant 3) | LIMBR1 | UK2     | 0      | No effect |

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## Other health conditions tested

| Genetic Condition                                                         | Gene    | Risk Variant | Copies | Inheritance | Result |
|---------------------------------------------------------------------------|---------|--------------|--------|-------------|--------|
| Acute Intermittent Porphyrria (Variant 1)                                 | HMBS    | Deletion     | 0      | AD          | Clear  |
| Acute Intermittent Porphyrria (Variant 2)                                 | HMBS    | G>A          | 0      | AD          | Clear  |
| Acute Intermittent Porphyrria (Variant 3)                                 | HMBS    | Insertion    | 0      | AD          | Clear  |
| Acute Intermittent Porphyrria (Variant 4)                                 | HMBS    | Deletion     | 0      | AD          | Clear  |
| Acute Intermittent Porphyrria (Variant 5)                                 | HMBS    | G>A          | 0      | AR          | Clear  |
| Autoimmune Lymphoproliferative Syndrome (Discovered in British Shorthair) | FASL    | Insertion    | 0      | AR          | Clear  |
| Burmese Head Defect (Discovered in the Burmese)                           | ALX1    | Deletion     | 0      | AD          | Clear  |
| Chediak-Higashi Syndrome (Discovered in the Persian)                      | LYST    | Insertion    | 0      | AR          | Clear  |
| Congenital Adrenal Hyperplasia                                            | CYP11B1 | G>A          | 0      | AR          | Clear  |
| Congenital Erythropoietic Porphyrria                                      | UROS    | G>A          | 0      | AR          | Clear  |
| Congenital Myasthenic Syndrome (Discovered in the Devon Rex and Sphynx)   | COLQ    | G>A          | 0      | AR          | Clear  |
| Cystinuria Type 1A                                                        | SCL3A1  | C>T          | 0      | AR          | Clear  |
| Cystinuria Type B (Variant 1)                                             | SCL7A9  | C>T          | 0      | AR          | Clear  |
| Cystinuria Type B (Variant 2)                                             | SCL7A9  | G>A          | 0      | AR          | Clear  |
| Cystinuria Type B (Variant 3)                                             | SCL7A9  | T>A          | 0      | AR          | Clear  |
| Dihydropyrimidinase Deficiency                                            | DPYS    | G>A          | 0      | AR          | Clear  |
| Earfold and Osteochondrodysplasia (Discovered in the Scottish Fold)       | TRPV4   | G>T          | 0      | AD          | Clear  |
| Familial Episodic Hypokalemic Polymyopathy (Discovered in the Burmese)    | WNK4    | C>T          | 0      | AR          | Clear  |
| Glutaric Aciduria Type II                                                 | ETFDH   | T>G          | 0      | AR          | Clear  |
| Glycogen Storage Disease (Discovered in the Norwegian Forest Cat)         | GBE1    | Insertion    | 0      | AR          | Clear  |

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## Other health conditions tested

| Genetic Condition                                                  | Gene   | Risk Variant | Copies | Inheritance | Result |
|--------------------------------------------------------------------|--------|--------------|--------|-------------|--------|
| GM1 Gangliosidosis                                                 | GLB1   | G>C          | 0      | AR          | Clear  |
| GM2 Gangliosidosis                                                 | GM2A   | Deletion     | 0      | AR          | Clear  |
| GM2 Gangliosidosis Type II (Discovered in Domestic Shorthair cats) | HEXB   | Insertion    | 0      | AR          | Clear  |
| GM2 Gangliosidosis Type II (Discovered in Japanese domestic cats)  | HEXB   | C>T          | 0      | AR          | Clear  |
| GM2 Gangliosidosis Type II (Discovered in the Burmese)             | HEXB   | Deletion     | 0      | AR          | Clear  |
| Hemophilia B (Variant 1)                                           | F9     | C>T          | 0      | XR          | Clear  |
| Hemophilia B (Variant 2)                                           | F9     | G>A          | 0      | XR          | Clear  |
| Hyperoxaluria Type II                                              | GRHPR  | G>A          | 0      | AR          | Clear  |
| Hypertrophic Cardiomyopathy (Discovered in the Maine Coon)         | MYBPC  | G>C          | 0      | AR          | Clear  |
| Hypotrichosis (Discovered in the Birman)                           | FOXN1  | Deletion     | 0      | AR          | Clear  |
| Lipoprotein Lipase Deficiency                                      | LPL    | G>A          | 0      | AR          | Clear  |
| Mucopolysaccharidosis Type I                                       | IDUA   | Deletion     | 0      | AR          | Clear  |
| Mucopolysaccharidosis Type VI                                      | ARSB   | T>C          | 0      | AR          | Clear  |
| Mucopolysaccharidosis Type VI Modifier                             | ARSB   | G>A          | 0      | MO          | Clear  |
| Mucopolysaccharidosis Type VII (Variant 1)                         | GUSB   | G>A          | 0      | AR          | Clear  |
| Mucopolysaccharidosis Type VII (Variant 2)                         | USB    | C>T          | 0      | AR          | Clear  |
| Myotonia Congenita                                                 | CLCN1  | G>T          | 0      | AR          | Clear  |
| Progressive Retinal Atrophy (Discovered in the Abyssinian)         | CEP290 | T>G          | 0      | AR          | Clear  |
| Progressive Retinal Atrophy (Discovered in the Bengal)             | KIF3B  | G>A          | 0      | AR          | Clear  |
| Progressive Retinal Atrophy (Discovered in the Persian)            | AIPL1  | C>T          | 0      | AR          | Clear  |
| Pyruvate Kinase Deficiency                                         | PKLR   | G>A          | 0      | AR          | Clear  |

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Other health conditions tested

| Genetic Condition                                      | Gene    | Risk Variant | Copies | Inheritance | Result |
|--------------------------------------------------------|---------|--------------|--------|-------------|--------|
| Sphingomyelinosis (Variant 1)                          | NPC1    | G>C          | 0      | AR          | Clear  |
| Sphingomyelinosis (Variant 2)                          | NPC2    | G>A          | 0      | AR          | Clear  |
| Spinal Muscular Atrophy (Discovered in the Maine Coon) | LIX1    | Deletion     | 0      | AR          | Clear  |
| Vitamin D-Dependent Rickets                            | CYP27B1 | G>T          | 0      | AR          | Clear  |

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## Glossary of genetic terms

### Test result definitions

**At Risk:** Based on the disorder's mode of inheritance, the cat inherited a number of genetic variant(s) which increases the cat's risk of being diagnosed with the associated disorder.

**Carrier:** The cat inherited one copy of a genetic variant when two copies are usually necessary to increase the cat's risk of being diagnosed with the associated disorder. While carriers are usually not at risk of clinical expression of the disorder, carriers of some complex variants may be associated with a low risk of developing the disorder.

**Notable:** Inheriting two copies of the genetic variant is noteworthy for specific aspects of health and breeding of the cat, but the cat should otherwise not suffer disease due to this genetic cause when in absence of other genetic variants.

**Clear:** The cat did not inherit the genetic variant(s) associated with the disorder and will not be at elevated risk of being diagnosed with the disorder due to this genotype. However, similar clinical signs could develop from different genetic or clinical causes.

**Inconclusive:** An inconclusive result indicates a confident call could not be made based on the data for that genetic variant. Health testing is performed in replicates, and on occasion the outcomes do not agree. This may occur due to an unusual sequence of DNA in the region tested, multiple cell genotypes present due to chimerism or acquired mutations, or due to quality of the DNA sample.

### Inheritance mode definitions

**Autosomal Recessive (AR):** For autosomal recessive disorders, cats with two copies of the genetic variant are at risk of developing the associated disorder. Cats with one copy of the variant are considered carriers and are usually not at risk of developing the disorder. However, carriers of some complex variants grouped in this category may be associated with a low risk of developing the disorder. Cats with one or two copies may pass the disorder-associated variant to their kittens if bred.

**Autosomal Recessive, asymptomatic (ARa):** For autosomal recessive, asymptomatic disorders, cats with two copies of the variant can exhibit certain aspects of the variant-associated disorder but otherwise, they should not suffer clinical disease as typically expected with autosomal recessive disorders. Cats with one copy of the variant are called carriers and should not exhibit any aspect of the disorder. However, cats with one or two copies may pass the disorder-associated variant to their kittens if bred.

**Autosomal Dominant (AD):** For autosomal dominant disorders, cats with one or two copies of the genetic variant are at risk of developing the associated disorder. Inheriting two copies of the variant may increase the risk of development of the disorder or cause the condition to be more severe. These cats may pass the disorder-associated variant to their kittens if bred.

**X-linked Recessive (XR):** For X-linked recessive disorders, the genetic variant is found on the X chromosome. Female cats must inherit two copies of the variant to be at risk of developing the condition, whereas male cats only need one copy to be at risk. Males and females with any copies of the variant may pass the disorder-associated variant to their kittens if bred.

**Modifier (MO):** Genetic modifiers do not cause disease on their own but can cause disease or change the onset or severity of a disorder when combined with another disorder-associated variant. For some modifier variants only one copy is required to cause an effect, for others two copies are required. Please refer to the associated variant's breeder recommendations regarding safe breeding practices for each modifier variant.