



# "SAPPHIRE"

## SAPPHIRE GEM



DNA Test Report

Test Date: January 16th, 2020

[embk.me/sapphiregem](https://embk.me/sapphiregem)

## BREED MIX



## GENETIC STATS

Predicted adult weight: **57 lbs**

Genetic age: **31 human years**

Based on the date of birth you provided

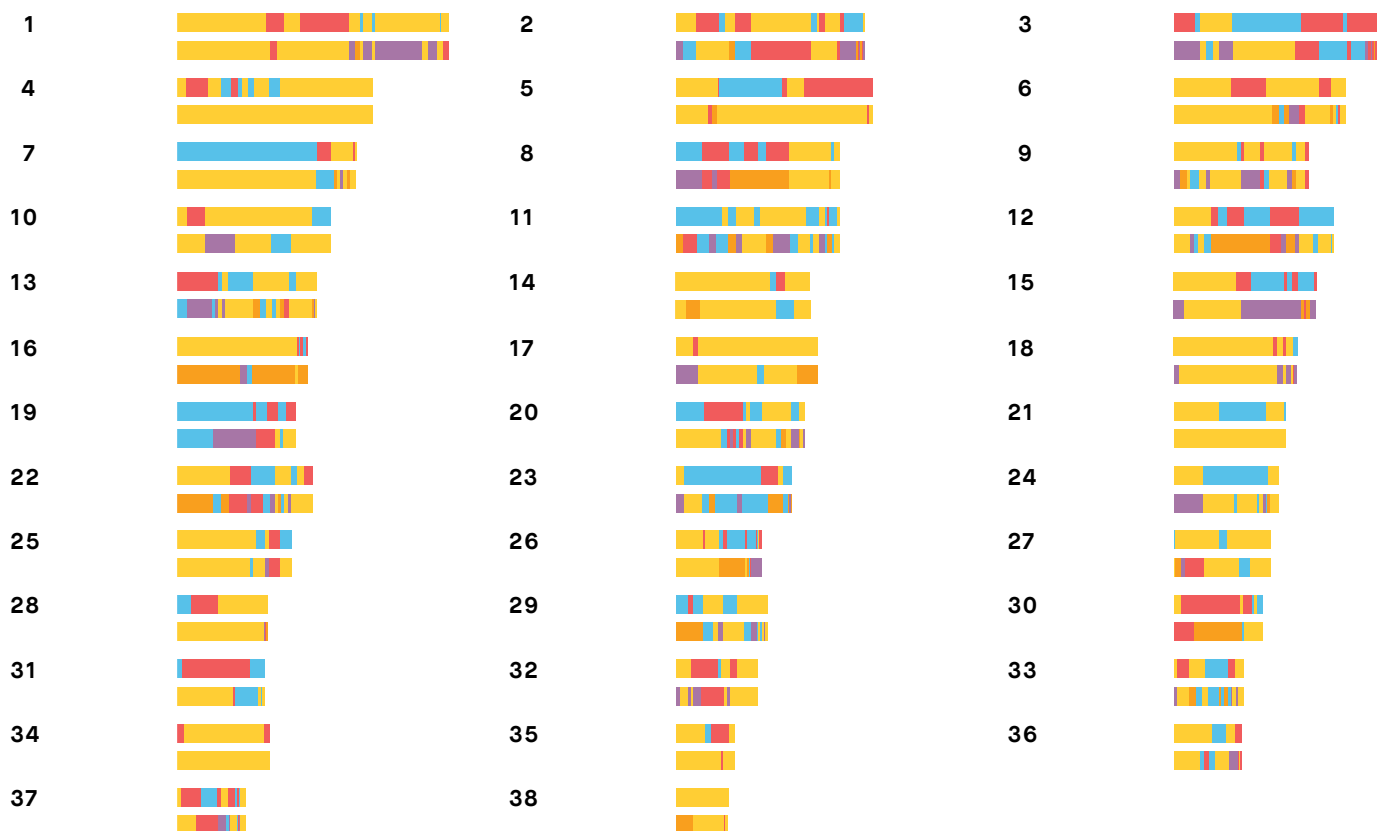
## TEST DETAILS

Kit number: EM-1579900

Swab number: 31001811417110

## BREED MIX BY CHROMOSOME

Our advanced test identifies from where Sapphire inherited every part of the chromosome pairs in her genome.





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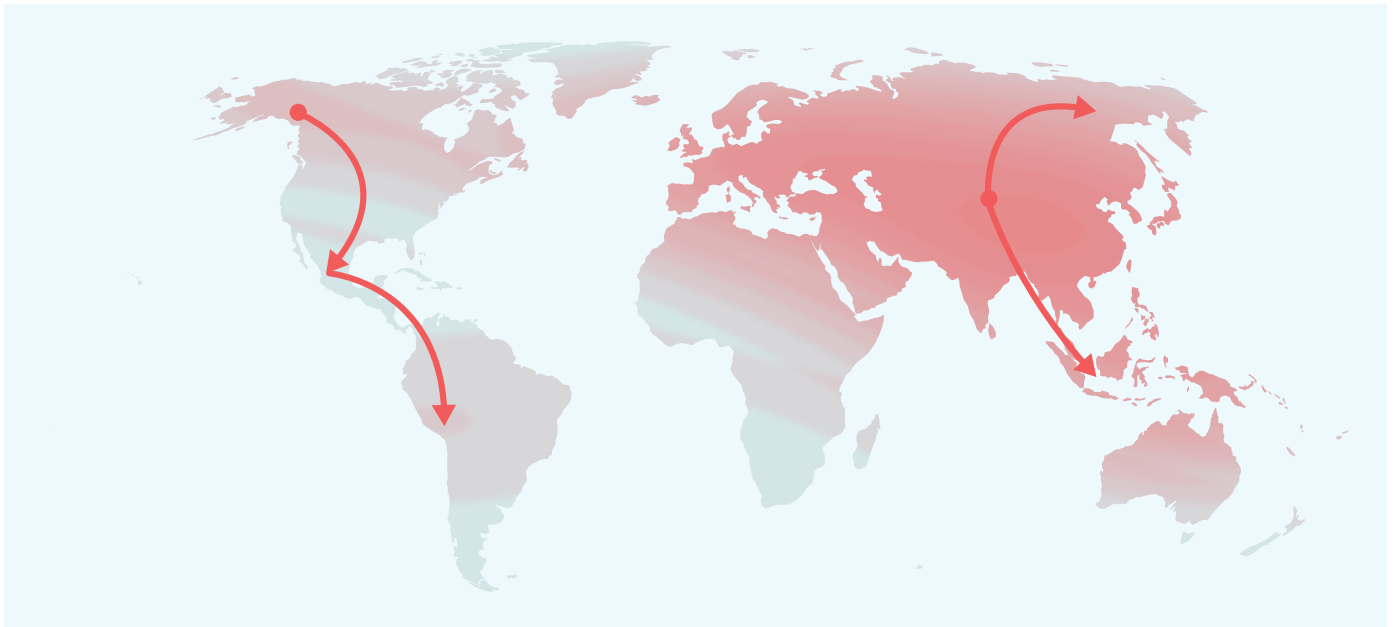


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## MATERNAL LINE



Through Sapphire's mitochondrial DNA we can trace her mother's ancestry back to where dogs and people first became friends. This map helps you visualize the routes that her ancestors took to your home. Their story is described below the map.

### HAPLOGROUP: A2

A2 is a very ancient maternal line. Most likely it was one of the major female lines that contributed to the very first domesticated dogs in Central Asia about 15,000 years ago. Some of the line stayed in Central Asia to the present day, and frequently appear as Tibetan Mastiffs and Akitas. Those that escaped the mountains of Central Asia sought out other cold spots, and are now found among Alaskan Malamutes and Siberian Huskies. This lineage is also occasionally found in several common Western breeds, such as German Shepherds and Labrador Retrievers. Curiously, all New Guinea Singing Dogs descend from this line. These are an ancient and very interesting breed found in the mountains of Papua New Guinea. Unfortunately, they are now endangered. They are closely related to the Australian dingo, so you could say its cousins are dingos! This line is also common in village dogs in Southeast and East Asia. Unlike many other lineages, A2 did not spread across the whole world, probably because it did not have the opportunity to hitch its wagon to European

### HAPLOTYPE: A29a

Part of the A2 haplogroup, this haplotype occurs most commonly in Siberian Huskies, Alaskan Malamutes, Labrador Retrievers, and village dogs from Alaska.





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## TRAITS: COAT COLOR

### TRAIT

### RESULT

#### E Locus (MC1R)

The E Locus determines if and where a dog can produce dark (black or brown) hair. Dogs with two copies of the recessive **e** allele do not produce dark hairs at all, and will be "red" over their entire body. The shade of red, which can range from a deep copper to yellow/gold to cream, is dependent on other genetic factors including the Intensity loci. In addition to determining if a dog can develop dark hairs at all, the E Locus can give a dog a black "mask" or "widow's peak," unless the dog has overriding coat color genetic factors.

Dogs with one or two copies of the **Em** allele usually have a melanistic mask (dark facial hair as commonly seen in the German Shepherd and Pug). Dogs with no copies of **Em** but one or two copies of the **Eg** allele usually have a melanistic "widow's peak" (dark forehead hair as commonly seen in the Afghan Hound and Borzoi, where it is called either "grizzle" or "domino").

**No dark mask or grizzle (EE)**

#### K Locus (CBD103)

The K Locus **K<sup>B</sup>** allele "overrides" the A Locus, meaning that it prevents the A Locus genotype from affecting coat color. For this reason, the **K<sup>B</sup>** allele is referred to as the "dominant black" allele. As a result, dogs with at least one **K<sup>B</sup>** allele will usually have solid black or brown coats (or red/cream coats if they are **ee** at the E Locus) regardless of their genotype at the A Locus, although several other genes could impact the dog's coat and cause other patterns, such as white spotting. Dogs with the **k<sup>Y</sup>k<sup>Y</sup>** genotype will show a coat color pattern based on the genotype they have at the A Locus. Dogs who test as **K<sup>B</sup>k<sup>Y</sup>** may be brindle rather than black or brown.

**More likely to have a patterned haircoat (k<sup>Y</sup>k<sup>Y</sup>)**



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## TRAITS: COAT COLOR (CONTINUED)

### TRAIT

### RESULT

#### Intensity Loci LINKAGE

Areas of a dog's coat where dark (black or brown) pigment is not expressed either contain red/yellow pigment, or no pigment at all. Five locations across five chromosomes explain approximately 70% of red pigmentation "intensity" variation across all dogs. Dogs with a result of **Intense Red Pigmentation** will likely have deep red hair like an Irish Setter or "apricot" hair like some Poodles, dogs with a result of **Intermediate Red Pigmentation** will likely have tan or yellow hair like a Soft-Coated Wheaten Terrier, and dogs with **Dilute Red Pigmentation** will likely have cream or white hair like a Samoyed. Because the mutations we test may not directly cause differences in red pigmentation intensity, we consider this to be a linkage test.

**Any light hair likely white or cream (Dilute Red Pigmentation)**

#### A Locus (ASIP)

The A Locus controls switching between black and red pigment in hair cells, but it will only be expressed in dogs that are not **ee** at the E Locus and are **k<sup>y</sup>k<sup>y</sup>** at the K Locus. Sable (also called "Fawn") dogs have a mostly or entirely red coat with some interspersed black hairs. Agouti (also called "Wolf Sable") dogs have red hairs with black tips, mostly on their head and back. Black and tan dogs are mostly black or brown with lighter patches on their cheeks, eyebrows, chest, and legs. Recessive black dogs have solid-colored black or brown coats.

**Black/Brown and tan coat color pattern (a<sup>tan</sup>a)**

#### D Locus (MLPH)

The D locus result that we report is determined by two different genetic variants that can work together to cause diluted pigmentation. These are the common **d** allele, also known as "**d1**", and a less common allele known as "**d2**". Dogs with two **d** alleles, regardless of which variant, will have all black pigment lightened ("diluted") to gray, or brown pigment lightened to lighter brown in their hair, skin, and sometimes eyes. There are many breed-specific names for these dilute colors, such as "blue", "charcoal", "fawn", "silver", and "Isabella". Note that in certain breeds, dilute dogs have a higher incidence of Color Dilution Alopecia. Dogs with one **d** allele will not be dilute, but can pass the **d** allele on to their puppies. To view your dog's **d1** and **d2** test results, click the "SEE DETAILS" link in the upper right hand corner of the "Base Coat Color" section of the Traits page, and then click the "VIEW SUBLOCUS RESULTS" link at the bottom of the page.

**Dark areas of hair and skin are not lightened (DD)**



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## TRAITS: COAT COLOR (CONTINUED)

### TRAIT

### RESULT

#### B Locus (TYRP1)

Dogs with two copies of the **b** allele produce brown pigment instead of black in both their hair and skin. Dogs with one copy of the **b** allele will produce black pigment, but can pass the **b** allele on to their puppies. E Locus **ee** dogs that carry two **b** alleles will have red or cream coats, but have brown noses, eye rims, and footpads (sometimes referred to as "Dudley Nose" in Labrador Retrievers). "Liver" or "chocolate" is the preferred color term for brown in most breeds; in the Doberman Pinscher it is referred to as "red".

**Black or gray hair and skin (BB)**

#### Saddle Tan (RALY)

The "Saddle Tan" pattern causes the black hairs to recede into a "saddle" shape on the back, leaving a tan face, legs, and belly, as a dog ages. The Saddle Tan pattern is characteristic of breeds like the Corgi, Beagle, and German Shepherd. Dogs that have the **II** genotype at this locus are more likely to be mostly black with tan points on the eyebrows, muzzle, and legs as commonly seen in the Doberman Pinscher and the Rottweiler. This gene modifies the A Locus **a<sup>t</sup>** allele, so dogs that do not express **a<sup>t</sup>** are not influenced by this gene.

**Likely saddle tan patterned (NI)**

#### S Locus (MITF)

The S Locus determines white spotting and pigment distribution. MITF controls where pigment is produced, and an insertion in the MITF gene causes a loss of pigment in the coat and skin, resulting in white hair and/or pink skin. Dogs with two copies of this variant will likely have breed-dependent white patterning, with a nearly white, parti, or piebald coat. Dogs with one copy of this variant will have more limited white spotting and may be considered flash, parti or piebald. This MITF variant does not explain all white spotting patterns in dogs and other variants are currently being researched. Some dogs may have small amounts of white on the paws, chest, face, or tail regardless of their S Locus genotype.

**Likely solid colored, but may have small amounts of white (Ssp)**



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## TRAITS: COAT COLOR (CONTINUED)

TRAIT	RESULT
<b>M Locus (PMEL)</b>	
<p>Merle coat patterning is common to several dog breeds including the Australian Shepherd, Catahoula Leopard Dog, and Shetland Sheepdog, among many others. Merle arises from an unstable SINE insertion (which we term the "M*" allele) that disrupts activity of the pigmentary gene PMEL, leading to mottled or patchy coat color. Dogs with an <b>M*m</b> result are likely to be phenotypically merle or could be "phantom" merle, that is, they have a merle allele that does not affect coat color. Dogs with an <b>M*M*</b> result are likely to be phenotypically merle or double merle. Dogs with an <b>mm</b> result have no merle alleles and are unlikely to have a merle coat pattern.</p>	<b>No merle alleles (mm)</b>

Note that Embark does not currently distinguish between the recently described cryptic, atypical, atypical+, classic, and harlequin merle alleles. Our merle test only detects the presence, but not the length of the SINE insertion. We do not recommend making breeding decisions on this result alone. Please pursue further testing for allelic distinction prior to breeding decisions.

<b>H Locus (Harlequin)</b>	
<p>This pattern is recognized in Great Danes and causes dogs to have a white coat with patches of darker pigment. A dog with an <b>Hh</b> result will be harlequin if they are also <b>M*m</b> or <b>M*M*</b> at the M Locus and are not <b>ee</b> at the E locus. Dogs with a result of <b>hh</b> will not be harlequin. This trait is thought to be homozygous lethal; a living dog with an <b>HH</b> genotype has never been found.</p>	<b>No harlequin alleles (hh)</b>



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## TRAITS: OTHER COAT TRAITS

TRAIT	RESULT
<b>Furnishings (RSP02) LINKAGE</b>	
Dogs with one or two copies of the <b>F</b> allele have "furnishings": the mustache, beard, and eyebrows characteristic of breeds like the Schnauzer, Scottish Terrier, and Wire Haired Dachshund. A dog with two <b>I</b> alleles will not have furnishings, which is sometimes called an "improper coat" in breeds where furnishings are part of the breed standard. The mutation is a genetic insertion which we measure indirectly using a linkage test highly correlated with the insertion.	<b>Likely unfurnished (no mustache, beard, and/or eyebrows) (II)</b>
<b>Coat Length (FGF5)</b>	
The FGF5 gene is known to affect hair length in many different species, including cats, dogs, mice, and humans. In dogs, the <b>T</b> allele confers a long, silky haircoat as observed in the Yorkshire Terrier and the Long Haired Whippet. The ancestral <b>G</b> allele causes a shorter coat as seen in the Boxer or the American Staffordshire Terrier. In certain breeds (such as Corgi), the long haircoat is described as "fluff."	<b>Likely short or mid-length coat (GG)</b>
<b>Shedding (MC5R)</b>	
Dogs with at least one copy of the ancestral <b>C</b> allele, like many Labradors and German Shepherd Dogs, are heavy or seasonal shedders, while those with two copies of the <b>T</b> allele, including many Boxers, Shih Tzus and Chihuahuas, tend to be lighter shedders. Dogs with furnished/wire-haired coats caused by RSP02 (the furnishings gene) tend to be low shedders regardless of their genotype at this gene.	<b>Likely heavy/seasonal shedding (CC)</b>
<b>Coat Texture (KRT71)</b>	
Dogs with a long coat and at least one copy of the <b>T</b> allele have a wavy or curly coat characteristic of Poodles and Bichon Frises. Dogs with two copies of the ancestral <b>C</b> allele are likely to have a straight coat, but there are other factors that can cause a curly coat, for example if they at least one <b>F</b> allele for the Furnishings (RSP02) gene then they are likely to have a curly coat. Dogs with short coats may carry one or two copies of the <b>T</b> allele but still have straight coats.	<b>Likely straight coat (CC)</b>



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## TRAITS: OTHER COAT TRAITS (CONTINUED)

### TRAIT

### RESULT

#### Hairlessness (FOXI3) LINKAGE

A duplication in the FOXI3 gene causes hairlessness over most of the body as well as changes in tooth shape and number. This mutation occurs in Peruvian Inca Orchid, Xoloitzcuintli (Mexican Hairless), and Chinese Crested (other hairless breeds have different mutations). Dogs with the **NDup** genotype are likely to be hairless while dogs with the **NN** genotype are likely to have a normal coat. The **DupDup** genotype has never been observed, suggesting that dogs with that genotype cannot survive to birth. Please note that this is a linkage test, so it may not be as predictive as direct tests of the mutation in some lines.

**Very unlikely to be hairless (NN)**

#### Hairlessness (SGK3)

Hairlessness in the American Hairless Terrier arises from a mutation in the SGK3 gene. Dogs with the **ND** genotype are likely to be hairless while dogs with the **NN** genotype are likely to have a normal coat.

**Very unlikely to be hairless (NN)**

#### Oculocutaneous Albinism Type 2 (SLC45A2) LINKAGE

Dogs with two copies **DD** of this deletion in the SLC45A2 gene have oculocutaneous albinism type 2 (OCA2), also known as Doberman Z Factor Albinism, a recessive condition characterized by severely reduced or absent pigment in the eyes, skin, and hair. Affected dogs sometimes suffer from vision problems due to lack of eye pigment (which helps direct and absorb ambient light) and are prone to sunburn. Dogs with a single copy of the deletion **ND** will not be affected but can pass the mutation on to their offspring. This particular mutation can be traced back to a single white Doberman Pinscher born in 1976, and it has only been observed in dogs descended from this individual. Please note that this is a linkage test, so it may not be as predictive as direct tests of the mutation in some lines.

**Likely not albino (NN)**





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## TRAITS: OTHER BODY FEATURES

TRAIT	RESULT
<b>Muzzle Length (BMP3)</b>  Dogs in medium-length muzzle (mesocephalic) breeds like Staffordshire Terriers and Labradors, and long muzzle (dolichocephalic) breeds like Whippet and Collie have one, or more commonly two, copies of the ancestral <b>C</b> allele. Dogs in many short-length muzzle (brachycephalic) breeds such as the English Bulldog, Pug, and Pekingese have two copies of the derived <b>A</b> allele. At least five different genes affect muzzle length in dogs, with BMP3 being the only one with a known causal mutation. For example, the skull shape of some breeds, including the dolichocephalic Scottish Terrier or the brachycephalic Japanese Chin, appear to be caused by other genes. Thus, dogs may have short or long muzzles due to other genetic factors that are not yet known to science.	<b>Likely medium or long muzzle (CC)</b>
<b>Tail Length (T)</b>  Whereas most dogs have two <b>C</b> alleles and a long tail, dogs with one <b>G</b> allele are likely to have a bobtail, which is an unusually short or absent tail. This mutation causes natural bobtail in many breeds including the Pembroke Welsh Corgi, the Australian Shepherd, and the Brittany Spaniel. Dogs with <b>GG</b> genotypes have not been observed, suggesting that dogs with the <b>GG</b> genotype do not survive to birth. Please note that this mutation does not explain every natural bobtail! While certain lineages of Boston Terrier, English Bulldog, Rottweiler, Miniature Schnauzer, Cavalier King Charles Spaniel, and Parson Russell Terrier, and Dobermans are born with a natural bobtail, these breeds do not have this mutation. This suggests that other unknown genetic mutations can also lead to a natural bobtail.	<b>Likely normal-length tail (CC)</b>
<b>Hind Dewclaws (LMBR1)</b>  Common in certain breeds such as the Saint Bernard, hind dewclaws are extra, nonfunctional digits located midway between a dog's paw and hock. Dogs with at least one copy of the <b>T</b> allele have about a 50% chance of having hind dewclaws. Note that other (currently unknown to science) mutations can also cause hind dewclaws, so some <b>TT</b> or <b>TC</b> dogs will have hind dewclaws.	<b>Unlikely to have hind dew claws (CC)</b>



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## TRAITS: OTHER BODY FEATURES (CONTINUED)

### TRAIT

### RESULT

#### Blue Eye Color (ALX4) LINKAGE

Embark researchers discovered this large duplication associated with blue eyes in Arctic breeds like Siberian Husky as well as tri-colored (non-merle) Australian Shepherds. Dogs with at least one copy of the duplication (**Dup**) are more likely to have at least one blue eye. Some dogs with the duplication may have only one blue eye (complete heterochromia) or may not have blue eyes at all; nevertheless, they can still pass the duplication and the trait to their offspring. **NN** dogs do not carry this duplication, but may have blue eyes due to other factors, such as merle. Please note that this is a linkage test, so it may not be as predictive as direct tests of the mutation in some lines.

**Less likely to have blue eyes (NN)**

#### Back Muscling & Bulk, Large Breed (ACSL4)

The **T** allele is associated with heavy muscling along the back and trunk in characteristically "bulky" large-breed dogs including the Saint Bernard, Bernese Mountain Dog, Greater Swiss Mountain Dog, and Rottweiler. The "bulky" **T** allele is absent from leaner shaped large breed dogs like the Great Dane, Irish Wolfhound, and Scottish Deerhound, which are fixed for the ancestral **C** allele. Note that this mutation does not seem to affect muscling in small or even mid-sized dog breeds with notable back muscling, including the American Staffordshire Terrier, Boston Terrier, and the English Bulldog.

**Likely normal muscling (CC)**



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### TRAITS: BODY SIZE

#### TRAIT

#### RESULT

##### Body Size (IGF1)

The **I** allele is associated with smaller body size.

**Larger (NN)**

##### Body Size (IGFR1)

The **A** allele is associated with smaller body size.

**Larger (GG)**

##### Body Size (STC2)

The **A** allele is associated with smaller body size.

**Larger (TT)**

##### Body Size (GHR - E191K)

The **A** allele is associated with smaller body size.

**Larger (GG)**

##### Body Size (GHR - P177L)

The **T** allele is associated with smaller body size.

**Larger (CC)**



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## TRAITS: PERFORMANCE

### TRAIT

### RESULT

#### Altitude Adaptation (EPAS1)

This mutation causes dogs to be especially tolerant of low oxygen environments (hypoxia), such as those found at high elevations. Dogs with at least one **A** allele are less susceptible to "altitude sickness." This mutation was originally identified in breeds from high altitude areas such as the Tibetan Mastiff.

**Normal altitude  
tolerance (GG)**

#### Appetite (POMC) LINKAGE

This mutation in the POMC gene is found primarily in Labrador and Flat Coated Retrievers. Compared to dogs with no copies of the mutation (**NN**), dogs with one (**ND**) or two (**DD**) copies of the mutation are more likely to have high food motivation, which can cause them to eat excessively, have higher body fat percentage, and be more prone to obesity. Read more about the genetics of POMC, and learn how you can contribute to research, in our blog post (<https://embarkvet.com/resources/blog/pomc-dogs/>). We measure this result using a linkage test.

**Normal food  
motivation (NN)**



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## CLINICAL TOOLS

These clinical genetic tools can inform clinical decisions and diagnoses. These tools do not predict increased risk for disease.

### Alanine Aminotransferase Activity (GPT)

 Sapphire's baseline ALT level is Low Normal

#### Why is this important to your vet?

Sapphire has one copy of a variant associated with reduced ALT activity as measured on veterinary blood chemistry panels. Please inform your veterinarian that Sapphire has this genotype, as ALT is often used as an indicator of liver health and Sapphire is likely to have a lower than average resting ALT activity. As such, an increase in Sapphire's ALT activity could be evidence of liver damage, even if it is within normal limits by standard ALT reference ranges.

#### What is Alanine Aminotransferase Activity?

Alanine aminotransferase (ALT) is a clinical tool that can be used by veterinarians to better monitor liver health. This result is not associated with liver disease. ALT is one of several values veterinarians measure on routine blood work to evaluate the liver. It is a naturally occurring enzyme located in liver cells that helps break down protein. When the liver is damaged or inflamed, ALT is released into the bloodstream.

#### How vets diagnose this condition

Genetic testing is the only way to provide your veterinarian with this clinical tool.

#### How this condition is treated

Veterinarians may recommend blood work to establish a baseline ALT value for healthy dogs with one or two copies of this variant.



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## HEALTH REPORT

### How to interpret Sapphire's genetic health results:

If Sapphire inherited any of the variants that we tested, they will be listed at the top of the Health Report section, along with a description of how to interpret this result. We also include all of the variants that we tested Sapphire for that we did not detect the risk variant for.

### A genetic test is not a diagnosis

This genetic test does not diagnose a disease. Please talk to your vet about your dog's genetic results, or if you think that your pet may have a health condition or disease.



### Good news!

Sapphire is not at increased risk for the genetic health conditions that Embark tests.

**Breed-Relevant Genetic Conditions**

**15 variants not detected**



**Additional Genetic Conditions**

**176 variants not detected**





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## BREED-RELEVANT CONDITIONS TESTED



**Sapphire did not have the variants that we tested for, that are relevant to her breeds:**

- ✓ MDR1 Drug Sensitivity (MDR1)
- ✓ Factor VII Deficiency (F7 Exon 5)
- ✓ Factor VIII Deficiency, Hemophilia A (F8 Exon 11, Shepherd Variant 1)
- ✓ Factor VIII Deficiency, Hemophilia A (F8 Exon 1, Shepherd Variant 2)
- ✓ Canine Leukocyte Adhesion Deficiency Type III, CLADIII (FERMT3)
- ✓ Platelet factor X receptor deficiency, Scott Syndrome (TMEM16F)
- ✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)
- ✓ Achromatopsia (CNGA3 Exon 7 German Shepherd Variant)
- ✓ Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU (SLC2A9)
- ✓ X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia (EDA Intron 8)
- ✓ Renal Cystadenocarcinoma and Nodular Dermatofibrosis, RCND (FLCN Exon 7)
- ✓ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3)
- ✓ GM1 Gangliosidosis (GLB1 Exon 15 Alaskan Husky Variant)
- ✓ Degenerative Myelopathy, DM (SOD1A)
- ✓ Polyneuropathy, NDRG1 Malamute Variant (NDRG1 Exon 4)



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## ADDITIONAL CONDITIONS TESTED



Sapphire did not have the variants that we tested for, in the following conditions that the potential effect on dogs with Sapphire's breeds may not yet be known.

- ✓ P2Y12 Receptor Platelet Disorder (P2Y12)
- ✓ Factor IX Deficiency, Hemophilia B (F9 Exon 7, Terrier Variant)
- ✓ Factor IX Deficiency, Hemophilia B (F9 Exon 7, Rhodesian Ridgeback Variant)
- ✓ Factor VIII Deficiency, Hemophilia A (F8 Exon 10, Boxer Variant)
- ✓ Thrombopathia (RASGRP1 Exon 5, Basset Hound Variant)
- ✓ Thrombopathia (RASGRP1 Exon 8)
- ✓ Thrombopathia (RASGRP1 Exon 5, American Eskimo Dog Variant)
- ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4)
- ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7)
- ✓ Von Willebrand Disease Type I (VWF)
- ✓ Von Willebrand Disease Type II, Type II vWD (VWF)
- ✓ Canine Leukocyte Adhesion Deficiency Type I, CLADI (ITGB2)
- ✓ Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant)
- ✓ Canine Elliptocytosis (SPTB Exon 30)
- ✓ Glanzmann's Thrombasthenia Type I (ITGA2B Exon 12)
- ✓ May-Hegglin Anomaly (MYH9)
- ✓ Prekallikrein Deficiency (KLKB1 Exon 8)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 5)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Labrador Variant)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Pug Variant)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Beagle Variant)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 10)
- ✓ Trapped Neutrophil Syndrome (VPS13B)







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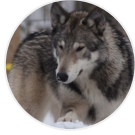
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## ADDITIONAL CONDITIONS TESTED

- ✓ Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)
- ✓ Congenital Hypothyroidism (TPO, Rat, Toy, Hairless Terrier Variant)
- ✓ Complement 3 Deficiency, C3 Deficiency (C3)
- ✓ Severe Combined Immunodeficiency (PRKDC)
- ✓ Severe Combined Immunodeficiency (RAG1)
- ✓ X-linked Severe Combined Immunodeficiency (IL2RG Variant 1)
- ✓ X-linked Severe Combined Immunodeficiency (IL2RG Variant 2)
- ✓ Progressive Retinal Atrophy, rcd1 (PDE6B Exon 21 Irish Setter Variant)
- ✓ Progressive Retinal Atrophy, rcd3 (PDE6A)
- ✓ Progressive Retinal Atrophy, CNGA (CNGA1 Exon 9)
- ✓ Progressive Retinal Atrophy, prcd (PRCD Exon 1)
- ✓ Progressive Retinal Atrophy (CNGB1)
- ✓ Progressive Retinal Atrophy (SAG)
- ✓ Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)
- ✓ Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)
- ✓ Progressive Retinal Atrophy, crd1 (PDE6B)
- ✓ Progressive Retinal Atrophy - crd4/cord1 (RPGRIP1)
- ✓ Progressive Retinal Atrophy, PRA3 (FAM161A)
- ✓ Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1)
- ✓ Day blindness, Cone Degeneration, Achromatopsia (CNGB3 Exon 6)
- ✓ Achromatopsia (CNGA3 Exon 7 Labrador Retriever Variant)
- ✓ Autosomal Dominant Progressive Retinal Atrophy (RHO)
- ✓ Canine Multifocal Retinopathy (BEST1 Exon 2)
- ✓ Canine Multifocal Retinopathy (BEST1 Exon 5)
- ✓ Canine Multifocal Retinopathy (BEST1 Exon 10 Deletion)





# "SAPPHIRE"

## SAPPHIRE GEM



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## ADDITIONAL CONDITIONS TESTED

- ✓ Canine Multifocal Retinopathy (BEST1 Exon 10 SNP)
- ✓ Glaucoma (ADAMTS10 Exon 9)
- ✓ Glaucoma (ADAMTS10 Exon 17)
- ✓ Glaucoma (ADAMTS17 Exon 11)
- ✓ Glaucoma (ADAMTS17 Exon 2)
- ✓ Goniodysgenesis and Glaucoma (OLFM3)
- ✓ Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Shepherd Variant)
- ✓ Primary Lens Luxation (ADAMTS17)
- ✓ Congenital Stationary Night Blindness (RPE65)
- ✓ Macular Corneal Dystrophy, MCD (CHST6)
- ✓ 2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis (APRT)
- ✓ Cystinuria Type I-A (SLC3A1)
- ✓ Cystinuria Type II-A (SLC3A1)
- ✓ Cystinuria Type II-B (SLC7A9)
- ✓ Polycystic Kidney Disease, PKD (PKD1)
- ✓ Primary Hyperoxaluria (AGXT)
- ✓ Protein Losing Nephropathy, PLN (NPHS1)
- ✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)
- ✓ Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 3)
- ✓ Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3)
- ✓ Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, Dry Eye Curly Coat Syndrome, CKCSID (FAM83H Exon 5)
- ✓ Canine Fucosidosis (FUCA1)
- ✓ Glycogen Storage Disease Type II, Pompe's Disease, GSD II (GAA)
- ✓ Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC)
- ✓ Glycogen Storage Disease Type IIIA, GSD IIIA (AGL)





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## ADDITIONAL CONDITIONS TESTED

- ✓ Mucopolysaccharidosis Type I, MPS I (IDUA)
- ✓ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 1)
- ✓ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 2)
- ✓ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5)
- ✓ Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Whippet and English Springer Spaniel Variant)
- ✓ Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Wachtelhund Variant)
- ✓ Lagotto Storage Disease (ATG4D)
- ✓ Neuronal Ceroid Lipofuscinosis 1, NCL 1 (PPT1 Exon 8)
- ✓ Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4)
- ✓ Neuronal Ceroid Lipofuscinosis 1, Cerebellar Ataxia, NCL4A (ARSG Exon 2)
- ✓ Neuronal Ceroid Lipofuscinosis 1, NCL 5 (CLN5 Border Collie Variant)
- ✓ Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7)
- ✓ Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 English Setter Variant)
- ✓ Neuronal Ceroid Lipofuscinosis (MFSD8)
- ✓ Neuronal Ceroid Lipofuscinosis (CLN8 Australian Shepherd Variant)
- ✓ Neuronal Ceroid Lipofuscinosis 10, NCL 10 (CTSD Exon 5)
- ✓ Neuronal Ceroid Lipofuscinosis (CLN5 Golden Retriever Variant)
- ✓ Adult-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Tibetan Terrier Variant)
- ✓ Late-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Australian Cattle Dog Variant)
- ✓ GM1 Gangliosidosis (GLB1 Exon 15 Shiba Inu Variant)
- ✓ GM1 Gangliosidosis (GLB1 Exon 2)
- ✓ GM2 Gangliosidosis (HEXB, Poodle Variant)
- ✓ GM2 Gangliosidosis (HEXA)
- ✓ Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5)
- ✓ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Italian Greyhound Variant)





# "SAPPHIRE"

## SAPPHIRE GEM



DNA Test Report

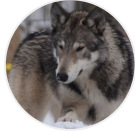
Test Date: January 16th, 2020

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## ADDITIONAL CONDITIONS TESTED

- ✓ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Parson Russell Terrier Variant)
- ✓ Persistent Mullerian Duct Syndrome, PMDS (AMHR2)
- ✓ Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MYO7A)
- ✓ Shar-Pei Autoinflammatory Disease, SPAID, Shar-Pei Fever (MTBP)
- ✓ Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy (SLC19A3)
- ✓ Alexander Disease (GFAP)
- ✓ Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration, NCCD (SPTBN2)
- ✓ Cerebellar Ataxia, Progressive Early-Onset Cerebellar Ataxia (SEL1L)
- ✓ Cerebellar Hypoplasia (VLDLR)
- ✓ Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA (CAPN1)
- ✓ Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10)
- ✓ Hereditary Ataxia (RAB24)
- ✓ Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LGI2)
- ✓ Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2)
- ✓ Hypomyelination and Tremors (FNIP2)
- ✓ Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP)
- ✓ Neuroaxonal Dystrophy, NAD (Spanish Water Dog Variant)
- ✓ Neuroaxonal Dystrophy, NAD (Rottweiler Variant)
- ✓ L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH)
- ✓ Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- ✓ Polyneuropathy, NDRG1 Greyhound Variant (NDRG1 Exon 15)
- ✓ Narcolepsy (HCRT2 Intron 6)
- ✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 15)
- ✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 4)
- ✓ Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV (RAB3GAP1, Rottweiler Variant)





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## ADDITIONAL CONDITIONS TESTED

- ✓ Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (GDNF-AS)
- ✓ Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1 (LPN1, ARHGEF10)
- ✓ Juvenile Myoclonic Epilepsy (DIRAS1)
- ✓ Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 2, LPN2 (GJA9)
- ✓ Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SeSAME/EAST Syndrome (KCNJ10)
- ✓ Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 (ATP1B2)
- ✓ Dilated Cardiomyopathy, DCM1 (PDK4)
- ✓ Dilated Cardiomyopathy, DCM2 (TTN)
- ✓ Long QT Syndrome (KCNQ1)
- ✓ Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)
- ✓ Muscular Dystrophy (DMD Pembroke Welsh Corgi Variant )
- ✓ Muscular Dystrophy (DMD Golden Retriever Variant)
- ✓ Limb Girdle Muscular Dystrophy (SGCD, Boston Terrier Variant)
- ✓ Centronuclear Myopathy (PTPLA)
- ✓ Exercise-Induced Collapse (DNM1)
- ✓ Inherited Myopathy of Great Danes (BIN1)
- ✓ Myostatin Deficiency, Bully Whippet Syndrome (MSTN)
- ✓ Myotonia Congenita (CLCN1 Exon 7)
- ✓ Myotonia Congenita (CLCN1 Exon 23)
- ✓ Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM (MTM1, Labrador Variant)
- ✓ Hypocatalasia, Acatlasemia (CAT)
- ✓ Pyruvate Dehydrogenase Deficiency (PDP1)
- ✓ Malignant Hyperthermia (RYR1)
- ✓ Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53)
- ✓ Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8)



# "SAPPHIRE"

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## ADDITIONAL CONDITIONS TESTED

- ✓ Lunde hund Syndrome (LEPREL1)
- ✓ Congenital Myasthenic Syndrome (CHAT)
- ✓ Congenital Myasthenic Syndrome (COLQ)
- ✓ Episodic Falling Syndrome (BCAN)
- ✓ Paroxysmal Dyskinesia, PxD (PGIN)
- ✓ Dystrophic Epidermolysis Bullosa (COL7A1)
- ✓ Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1)
- ✓ Ichthyosis, Epidermolytic Hyperkeratosis (KRT10)
- ✓ Ichthyosis (PNPLA1)
- ✓ Ichthyosis (SLC27A4)
- ✓ Ichthyosis (NIPAL4)
- ✓ Hereditary Footpad Hyperkeratosis (FAM83G)
- ✓ Hereditary Nasal Parakeratosis (SUV39H2)
- ✓ Musladin-Lueke Syndrome (ADAMTSL2)
- ✓ Oculocutaneous Albinism, OCA2 (Pekingese Type)
- ✓ Bald Thigh Syndrome (IGFBP5)
- ✓ Cleft Lip and/or Cleft Palate (ADAMTS20)
- ✓ Hereditary Vitamin D-Resistant Rickets (VDR)
- ✓ Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2)
- ✓ Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1)
- ✓ Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1)
- ✓ Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)
- ✓ Skeletal Dysplasia 2, SD2 (COL11A2)
- ✓ Craniomandibular Osteopathy, CMO (SLC37A2)
- ✓ Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)





# "SAPPHIRE"

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## ADDITIONAL CONDITIONS TESTED

✓ Chondrodystrophy, Norwegian Elkhound and Karelian Bear Dog Variant (ITGA10)



# "SAPPHIRE"

## SAPPHIRE GEM



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## INBREEDING AND DIVERSITY

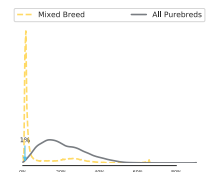
### CATEGORY

#### Coefficient Of Inbreeding

Our genetic COI measures the proportion of your dog's genome where the genes on the mother's side are identical by descent to those on the father's side.

### RESULT

1%

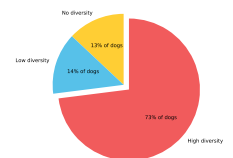


#### MHC Class II - DLA DRB1

A Dog Leukocyte Antigen (DLA) gene, DRB1 encodes a major histocompatibility complex (MHC) protein involved in the immune response. Some studies have shown associations between certain DRB1 haplotypes and autoimmune diseases such as Addison's disease (hypoadrenocorticism) in certain dog breeds, but these findings have yet to be scientifically validated.

### High Diversity

How common is this amount of diversity in mixed breed dogs:



#### MHC Class II - DLA DQA1 and DQB1

DQA1 and DQB1 are two tightly linked DLA genes that code for MHC proteins involved in the immune response. A number of studies have shown correlations of DQA-DQB1 haplotypes and certain autoimmune diseases; however, these have not yet been scientifically validated.

### No Diversity

How common is this amount of diversity in mixed breed dogs:

