### **PINE CREEK GRACIE**



DNA Test Report Test Date: December 10th, 2024 embk.me/pinecreekgracie

### **BREED ANCESTRY**

Bernese Mountain Dog: 100.0%

### **GENETIC STATS**

Predicted adult weight: 94 lbs

Life stage: Puppy

Based on your dog's date of birth provided.

### **TEST DETAILS**

Kit number: EM-53825426 Swab number: 31220710609856

Registration: American Kennel Club

(AKC)



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Fun Fact
Berners can haul up to 1,000 pounds 10 times their weight!

### **BERNESE MOUNTAIN DOG**

The Bernese Mountain Dog, commonly referred to as a 'Berner', is a versatile working dog that is both visually pleasing and a loyal companion. The Bernese Mountain Dog was bred to herd cattle, pull carts and be a watchdog in the Swiss farmlands. The ancient 'Molosser' breed is considered the main contributor to Mastiff-type dogs, which include the Berner. It is likely that the Molosser bred with farm dogs from the Swiss Alps in the first century B.C., developing a number of Swiss Sennenhund ("mountain dog") breeds, including the Berner Sennenhund. It is thought that the Berner continued working on these Swiss farmlands for over 2,000 years, before their primary purpose switched from herding cattle to appearing as a show dog in the early 20th century. They were first classified as the Bernese Mountain Dog at this time by the Swiss Kennel Club. Following World War I, in which the breed nearly became extinct, Berners were exported to America before being accepted by the AKC as an official breed in 1937. Breed development faltered somewhat during World War II before Berners became an established and popular breed in the mid to late 20th century. This easygoing breed likes to be around their owners, where their calm and intelligent nature makes them a beloved family dog. Berners exhibit their working dog instincts in their willingness to learn and relative ease to be trained. Their heritage also often results in being protective and sometimes shy towards new people and dogs. Early socialization training allows the Bernese Mountain Dog to learn to overcome initial caution around new things. This breed is a large dog, weighing around 100 pounds, and likes to keep busy, so it is important training is conducted while young and manageable. While they are well-tempered dogs, they are slow to mature and often exhibit puppy behavior for a number of years before reaching full maturity. Due to their beautiful and thick double coat, Berners tend to shed generously, requiring frequent brushing to keep under control. Unfortunately, owing to their size and limited gene pool, Bernese Mountain Dogs are prone to health problems and have a life expectancy of between 6-8 years. Nonetheless, this lovable dog

Registration:



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### **MATERNAL LINE**



Through Gracie'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: A1e

This female lineage likely stems from some of the original Central Asian wolves that were domesticated into modern dogs starting about 15,000 years ago. It seemed to be a fairly rare dog line for most of dog history until the past 300 years, when the lineage seemed to "explode" out and spread quickly. What really separates this group from the pack is its presence in Alaskan village dogs and Samoyeds. It is possible that this was an indigenous lineage brought to the Americas from Siberia when people were first starting to make that trip themselves! We see this lineage pop up in overwhelming numbers of Irish Wolfhounds, and it also occurs frequently in popular large breeds like Bernese Mountain Dogs, Saint Bernards and Great Danes. Shetland Sheepdogs are also common members of this maternal line, and we see it a lot in Boxers, too. Though it may be all mixed up with European dogs thanks to recent breeding events, its origins in the Americas makes it a very exciting lineage for sure!

#### **HAPLOTYPE: A228**

Part of the large A1e haplogroup, we have spotted this haplotype in village dogs in the Democratic Republic of the Congo and in the Dominican Republic. Among breeds, we see it frequently in big dogs like Saint Bernards, Leonbergers, and Great Danes. However, we also see it in small breeds including wire Fox Terriers and Rat Terriers. That's a pretty wide size range!



### PINE CREEK GRACIE



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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** variant do not produce dark hairs and will express a red pigment called pheomelanin over their entire body. The shade of red, which can range from a deep copper to white, depends on other genetic factors, including the Intensity loci. In addition to determining if a dog can develop dark hairs, the E Locus can give a dog a black "mask" or "widow's peak" unless the dog has overriding coat color genetic factors.

No dark mask or grizzle (EE)

Dogs with one or two copies of the E<sup>m</sup> variant may have a melanistic mask (dark facial hair as commonly seen in the German Shepherd Dog and Pug). In the absence of E<sup>m</sup>, dogs with the E<sup>g</sup> variant can have a "grizzle" phenotype (darker color on the head and top with a melanistic "widow's peak" and a lighter underside, commonly seen in the Afghan Hound and Borzoi and also referred to as "domino"). In the absence of both E<sup>m</sup> and E variants, dogs with the E<sup>a</sup> or E<sup>h</sup> variants can express the grizzle phenotype. Additionally, a dog with any combination of two of the E<sup>g</sup>, E<sup>a</sup>, or E<sup>h</sup> variants (example: E<sup>g</sup>E<sup>a</sup>) is also expected to express the grizzle phenotype.

#### K Locus (CBD103)

The K Locus  $K^B$  allele "overrides" the A Locus, meaning that it prevents the A Locus genotype from affecting coat color. For this reason, the  $K^B$  allele is referred to as the "dominant black" allele. As a result, dogs with at least one  $K^B$  allele will usually have solid black or brown coats (or red/cream coats if they are  $E^B$  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  $E^J$  genotype will show a coat color pattern based on the genotype they have at the A Locus. Dogs who test as  $E^J$  may be brindle rather than black or brown.

More likely to have a patterned haircoat  $(k^{y}k^{y})$ 





### PINE CREEK GRACIE



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## TRAITS: COAT COLOR (CONTINUED)

TRAIT RESULT

#### Intensity Loci

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 yellow or tan (Intermediate 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>t</sup>a<sup>t</sup>)

#### D Locus (MLPH)

The D locus result that we report is determined by three different genetic variants that can work together to cause diluted pigmentation. These are the common **d** allele, also known as "**d1**", and the less common alleles known as "**d2**" and "**d3**". 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.

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





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## TRAITS: COAT COLOR (CONTINUED)

TRAIT RESULT

#### Cocoa (HPS3)

Dogs with the **coco** genotype will produce dark brown pigment instead of black in both their hair and skin. Dogs with the **Nco** genotype will produce black pigment, but can pass the **co** allele on to their puppies. Dogs that have the **coco** genotype as well as the **bb** genotype at the B locus are generally a lighter brown than dogs that have the **Bb** or **BB** genotypes at the B locus.

No co alleles, not expressed (NN)

#### **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.

Not saddle tan patterned (II)

#### 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 to have little to no white in coat (SS)

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### PINE CREEK GRACIE



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## TRAITS: COAT COLOR (CONTINUED)

TRAIT RESULT

#### M Locus (PMEL)

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 M\*m result are likely to be phenotypically merle or could be "non-expressing" merle, meaning that the merle pattern is very subtle or not at all evident in their coat. Dogs with an M\*M\* result are likely to be phenotypically merle or double merle. Dogs with an mm result have no merle alleles and are unlikely to have a merle coat pattern.

No merle alleles (mm)

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.

#### R Locus (USH2A)

The R Locus regulates the presence or absence of the roan coat color pattern. Partial duplication of the USH2A gene is strongly associated with this coat pattern. Dogs with at least one **R** allele will likely have roaning on otherwise uniformly unpigmented white areas. Roan appears in white areas controlled by the S Locus but not in other white or cream areas created by other loci, such as the E Locus with **ee** along with Dilute Red Pigmentation by I Locus (for example, in Samoyeds). Mechanisms for controlling the extent of roaning are currently unknown, and roaning can appear in a uniform or non-uniform pattern. Further, non-uniform roaning may appear as ticked, and not obviously roan. The roan pattern can appear with or without ticking.

Likely no impact on coat pattern (rr)

#### H Locus (Harlequin)

This pattern is recognized in Great Danes and causes dogs to have a white coat with patches of darker pigment. A dog with an **Hh** result will be harlequin if they are also **M\*m** or **M\*M\*** at the M Locus and are not **ee** at the E locus. Dogs with a result of **hh** will not be harlequin. This trait is thought to be homozygous lethal; a living dog with an **HH** genotype has never been found.

No harlequin alleles (hh)

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## TRAITS: COAT COLOR (CONTINUED)

TRAIT RESULT

#### **Panda White Spotting**

Panda White Spotting originated in a line of German Shepherd Dogs and causes a mostly symmetrical white spotting of the head and/or body. This is a dominant variant of the KIT gene, which has a role in pigmentation.

Not expected to display Panda pattern (NN)

Dogs with one copy of the I allele will exhibit this white spotting. Dogs with two copies of the I allele have never been observed, as two copies of the variant is suspected to be lethal to the developing embryo. Dogs with the NN result will not exhibit white spotting due to this variant.

## **PINE CREEK GRACIE**



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### TRAITS: OTHER COAT TRAITS

TRAIT RESULT

#### Furnishings (RSPO2)

Dogs with one or two copies of the **F** allele have "furnishings": the mustache, beard, and eyebrows characteristic of breeds like the Schnauzer, Scottish Terrier, and Wire Haired Dachshund. A dog with two **I** 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.

Likely unfurnished (no mustache, beard, and/or eyebrows) (II)





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## TRAITS: OTHER COAT TRAITS (CONTINUED)

TRAIT RESULT

#### Coat Length (FGF5)

The FGF5 gene affects hair length in many species, including cats, dogs, mice, and humans. In dogs, an **Lh** allele confers a long, silky hair coat across many breeds, including Yorkshire Terriers, Cocker Spaniels, and Golden Retrievers, while the **Sh** allele causes a shorter coat, as seen in the Boxer or the American Staffordshire Terrier. In certain breeds, such as the Pembroke Welsh Corgi and French Bulldog, the long haircoat is described as "fluffy". The coat length determined by FGF5, as reported by us, is influenced by four genetic variants that work together to promote long hair.

The most common of these is the **Lh1** variant (G/T, CanFam3.1, chr32, g.4509367) and the less common ones are **Lh2** (C/T, CanFam3.1, chr32, g.4528639), **Lh3** (16bp deletion, CanFam3.1, chr32, g.4528616), and **Lh4** (GG insertion, CanFam3.1, chr32, g.4528621). The FGF5\_Lh1 variant is found across many dog breeds. The less common alleles, FGF5\_Lh2, have been found in the Akita, Samoyed, and Siberian Husky, FGF5\_Lh3 have been found in the Eurasier, and FGF5\_Lh4 have been found in the Afghan Hound, Eurasier, and French Bulldog.

Likely long coat (LhLh)

The **Lh** alleles have a recessive mode of inheritance, meaning that two copies of the **Lh** alleles are required to have long hair. The presence of two Lh alleles at any of these FGF5 loci is expected to result in long hair. One copy each of **Lh1** and **Lh2** have been found in Samoyeds, one copy each of **Lh1** and **Lh3** have been found in Eurasiers, and one copy each of **Lh1** and **Lh4** have been found in the Afghan Hounds and Eurasiers.

Interestingly, the Lh3 variant, a 16 base pair deletion, encompasses the Lh4 variant (GG insertion). The presence of one or two copies of Lh3 influences the outcome at the Lh4 locus. When two copies of Lh3 are present, there will be no reportable result for the FGF5\_Lh4 locus. With one copy of Lh3, Lh4 can have either one copy of the variant allele or the normal allele. The overall FGF5 result remains unaffected by this.

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## TRAITS: OTHER COAT TRAITS (CONTINUED)

TRAIT RESULT

#### Shedding (MC5R)

Dogs with at least one copy of the ancestral **C** allele, like many Labradors and German Shepherd Dogs, are heavy or seasonal shedders, while those with two copies of the **T** allele, including many Boxers, Shih Tzus and Chihuahuas, tend to be lighter shedders. Dogs with furnished/wire-haired coats caused by RSPO2 (the furnishings gene) tend to be low shedders regardless of their genotype at this gene.

Likely heavy/seasonal shedding (CT)

#### Coat Texture (KRT71)

Dogs with a long coat and at least one copy of the **T** allele have a wavy or curly coat characteristic of Poodles and Bichon Frises. Dogs with two copies of the ancestral **C** 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 **F** allele for the Furnishings (RSPO2) gene then they are likely to have a curly coat. Dogs with short coats may carry one or two copies of the **T** allele but still have straight coats.

Likely straight coat (CC)

#### Hairlessness (FOXI3)

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 **DD** result are likely to be hairless. Dogs with the **ND** genotype will have a normal coat, but can pass the **D** variant on to their offspring.

Very unlikely to be hairless (NN)





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# TRAITS: OTHER COAT TRAITS (CONTINUED)

TRAIT RESULT

#### Oculocutaneous Albinism Type 2 (SLC45A2)

Dogs with two copies **DD** of this deletion in the SLC45A2 gene have oculocutaneous albinism (OCA), 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

#### Muzzle Length (BMP3)

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  $\mathbf{C}$  allele. Dogs in many short-length muzzle (brachycephalic) breeds such as the English Bulldog, Pug, and Pekingese have two copies of the derived  $\mathbf{A}$  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.

Likely medium or long muzzle (CC)

#### Tail Length (T)

Whereas most dogs have two **C** alleles and a long tail, dogs with one **G** 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 **GG** genotypes have not been observed, suggesting that dogs with the **GG** 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.

Likely normal-length tail (CC)

#### Hind Dewclaws (LMBR1)

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 **T** 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 **CC** or **TC** dogs will have hind dewclaws.

Likely to have hind dew claws (CT)

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## TRAITS: OTHER BODY FEATURES (CONTINUED)

TRAIT RESULT

#### Chondrodysplasia (Chr. 18 FGF4 Retrogene)

Dogs with one or two copies of the I allele will exhibit a short-legged trait known as chondrodysplasia (CDPA). CDPA is a breed-defining characteristic of many breeds exhibiting the "short-legged, long-bodied" appearance known as disproportionate dwarfism, including the corgi, dachshund and basset hound. The impact of the I allele on leg length is additive. Therefore, dogs with the II result display the largest reduction in leg length. Dogs with the NI genotype will have an intermediate leg length, while dogs with the NN result will not exhibit leg shortening due to this variant. Breeds that display disproportionate dwarfism also frequently inherit a genetic variant known as the chondrodystrophy (CDDY) variant. The CDDY variant also shortens legs (in a less significant amount than CDPA) but, secondarily, increases the risk of Type I Intervertebral Disc Disease (IVDD). Test results for CDDY are listed in this dog's health testing results under "Intervertebral Disc Disease (Type I)". In contrast, the CDPA variant has NOT been shown to increase the risk of IVDD.

Not indicative of chondrodysplasia (normal leg length) (NN)

#### **Blue Eye Color (ALX4)**

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 heavy muscling (TT)

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TRAITS: BODY SIZE

**TRAIT RESULT Body Size (IGF1)** Larger (NN) The I allele is associated with smaller body size. **Body Size (IGFR1)** Larger (GG) The A allele is associated with smaller body size. Body Size (STC2) Intermediate (TA) The A allele is associated with smaller body size. Body Size (GHR - E191K) Larger (GG) The A allele is associated with smaller body size. Body Size (GHR - P177L) Larger (CC) The  ${\bf T}$  allele is associated with smaller body size.

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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  $\bf A$  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)

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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### **HEALTH REPORT**

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

If Gracie 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 Gracie 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.

#### **Summary**

Of the 274 genetic health risks we analyzed, we found 1 result that you should learn about.

Notable results (1)

**Copper Toxicosis (Attenuating)** 

Clear results

Breed-relevant (2)

Other (270)

Registration: American Kennel Club

Hembark

(AKC)

### **PINE CREEK GRACIE**



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### **BREED-RELEVANT RESULTS**

Research studies indicate that these results are more relevant to dogs like Gracie, and may influence her chances of developing certain health conditions.

 ✓ Degenerative Myelopathy, DM (SOD1A)
 Clear

 ✓ Von Willebrand Disease Type I, Type I vWD (VWF)
 Clear



### **PINE CREEK GRACIE**



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### **OTHER RESULTS**

Research has not yet linked these conditions to dogs with similar breeds to Gracie. Review any increased risk or notable results to understand her potential risk and recommendations.

Copper Toxicosis (Attenuating) (ATP7A, Labrador Retriever)	Notable
② 2-DHA Kidney & Bladder Stones (APRT)	Clear
Acral Mutilation Syndrome (GDNF-AS, Spaniel and Pointer Variant)	Clear
Alaskan Husky Encephalopathy (SLC19A3)	Clear
Alaskan Malamute Polyneuropathy, AMPN (NDRG1 SNP)	Clear
Alexander Disease (GFAP)	Clear
ALT Activity (GPT)	Clear
Anhidrotic Ectodermal Dysplasia (EDA Intron 8)	Clear
Autosomal Dominant Progressive Retinal Atrophy (RHO)	Clear
Bald Thigh Syndrome (IGFBP5)	Clear
Bernard-Soulier Syndrome, BSS (GP9, Cocker Spaniel Variant)	Clear
Bully Whippet Syndrome (MSTN)	Clear
⊘ Canine Elliptocytosis (SPTB Exon 30)	Clear
	Clear
Canine Leukocyte Adhesion Deficiency Type I, CLAD I (ITGB2, Setter Variant)	Clear
Oanine Leukocyte Adhesion Deficiency Type III, CLAD III (FERMT3, German Shepherd Variant)	Clear
Oanine Multifocal Retinopathy, cmr1 (BEST1 Exon 2)	Clear
Oanine Multifocal Retinopathy, cmr2 (BEST1 Exon 5, Coton de Tulear Variant)	Clear

## **PINE CREEK GRACIE**



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## **OTHER RESULTS**

Canine Multifocal Retinopathy, cmr3 (BEST1 Exon 10 Deletion, Finnish and Swedish Lapphund, Lapponian Herder Variant)	Clear
Oanine Multiple System Degeneration (SERAC1 Exon 4, Chinese Crested Variant)	Clear
Oanine Multiple System Degeneration (SERAC1 Exon 15, Kerry Blue Terrier Variant)	Clear
Cardiomyopathy and Juvenile Mortality (YARS2)	Clear
Centronuclear Myopathy, CNM (PTPLA)	Clear
Cerebellar Hypoplasia (VLDLR, Eurasier Variant)	Clear
Chondrodystrophy (ITGA10, Norwegian Elkhound and Karelian Bear Dog Variant)	Clear
Cleft Lip and/or Cleft Palate (ADAMTS20, Nova Scotia Duck Tolling Retriever Variant)	Clear
○ Cleft Palate, CP1 (DLX6 intron 2, Nova Scotia Duck Tolling Retriever Variant)	Clear
Obalamin Malabsorption (CUBN Exon 8, Beagle Variant)	Clear
Obalamin Malabsorption (CUBN Exon 53, Border Collie Variant)	Clear
○ Collie Eye Anomaly (NHEJ1)	Clear
Omplement 3 Deficiency, C3 Deficiency (C3)	Clear
Ongenital Cornification Disorder (NSDHL, Chihuahua Variant)	Clear
Ongenital Dyserythropoietic Anemia and Polymyopathy (EHPB1L1, Labrador Retriever Variant)	Clear
Ongenital Hypothyroidism (TPO, Rat, Toy, Hairless Terrier Variant)	Clear
Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)	Clear
Ongenital Hypothyroidism with Goiter (TPO Intron 13, French Bulldog Variant)	Clear



## **PINE CREEK GRACIE**



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## **OTHER RESULTS**

Congenital Hypothyroidism with Goiter (SLC5A5, Shih Tzu Variant)	Clear
Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant)	Clear
Congenital Muscular Dystrophy (LAMA2, Italian Greyhound)	Clear
Ongenital Myasthenic Syndrome, CMS (COLQ, Labrador Retriever Variant)	Clear
Ongenital Myasthenic Syndrome, CMS (COLQ, Golden Retriever Variant)	Clear
Ongenital Myasthenic Syndrome, CMS (CHAT, Old Danish Pointing Dog Variant)	Clear
Ongenital Myasthenic Syndrome, CMS (CHRNE, Jack Russell Terrier Variant)	Clear
Congenital Stationary Night Blindness (LRIT3, Beagle Variant)	Clear
Congenital Stationary Night Blindness (RPE65, Briard Variant)	Clear
Opper Toxicosis (Accumulating) (ATP7B)	Clear
Opper Toxicosis (Attenuating) (RETN, Labrador Retriever)	Clear
⊘ Craniomandibular Osteopathy, CMO (SLC37A2)	Clear
⊘ Craniomandibular Osteopathy, CMO (SLC37A2 Intron 16, Basset Hound Variant)	Clear
Cystinuria Type I-A (SLC3A1, Newfoundland Variant)	Clear
Cystinuria Type II-A (SLC3A1, Australian Cattle Dog Variant)	Clear
Cystinuria Type II-B (SLC7A9, Miniature Pinscher Variant)	Clear
Oarier Disease (ATP2A2, Irish Terrier Variant)	Clear
Oay Blindness (CNGB3 Deletion, Alaskan Malamute Variant)	Clear



## **PINE CREEK GRACIE**



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## **OTHER RESULTS**

Oay Blindness (CNGA3 Exon 7, German Shepherd Variant)	Clear
Oay Blindness (CNGA3 Exon 7, Labrador Retriever Variant)	Clear
Oay Blindness (CNGB3 Exon 6, German Shorthaired Pointer Variant)	Clear
O Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MYO7A)	Clear
Demyelinating Polyneuropathy (SBF2/MTRM13)	Clear
O Dental-Skeletal-Retinal Anomaly (MIA3, Cane Corso Variant)	Clear
O Diffuse Cystic Renal Dysplasia and Hepatic Fibrosis (INPP5E Intron 9, Norwich Terrier Variant)	Clear
Dilated Cardiomyopathy, DCM (RBM20, Schnauzer Variant)	Clear
	Clear
Dilated Cardiomyopathy, DCM2 (TTN, Doberman Pinscher Variant 2)	Clear
Disproportionate Dwarfism (PRKG2, Dogo Argentino Variant)	Clear
Ory Eye Curly Coat Syndrome (FAM83H Exon 5)	Clear
Oystrophic Epidermolysis Bullosa (COL7A1, Central Asian Shepherd Dog Variant)	Clear
Oystrophic Epidermolysis Bullosa (COL7A1, Golden Retriever Variant)	Clear
Early Bilateral Deafness (LOXHD1 Exon 38, Rottweiler Variant)	Clear
Early Onset Adult Deafness, EOAD (EPS8L2 Deletion, Rhodesian Ridgeback Variant)	Clear
Early Onset Cerebellar Ataxia (SEL1L, Finnish Hound Variant)	Clear
Ehlers Danlos (ADAMTS2, Doberman Pinscher Variant)	Clear



## **PINE CREEK GRACIE**



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## **OTHER RESULTS**

Ehlers-Danlos Syndrome (EDS) (COL5A1, Labrador Retriever Variant)	Clear
Enamel Hypoplasia (ENAM Deletion, Italian Greyhound Variant)	Clear
Enamel Hypoplasia (ENAM SNP, Parson Russell Terrier Variant)	Clear
Episodic Falling Syndrome (BCAN)	Clear
Exercise-Induced Collapse, EIC (DNM1)	Clear
Factor VII Deficiency (F7 Exon 5)	Clear
Factor XI Deficiency (F11 Exon 7, Kerry Blue Terrier Variant)	Clear
Familial Nephropathy (COL4A4 Exon 3, Cocker Spaniel Variant)	Clear
Familial Nephropathy (COL4A4 Exon 30, English Springer Spaniel Variant)	Clear
Fanconi Syndrome (FAN1, Basenji Variant)	Clear
Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2, Giant Schnauzer Variant)	Clear
	Clear
Glanzmann's Thrombasthenia Type I (ITGA2B Exon 12, Otterhound Variant)	Clear
Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5, Terrier Variant)	Clear
Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC1, German Pinscher Variant)	Clear
Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC, Maltese Variant)	Clear
Glycogen Storage Disease Type IIIA, GSD IIIA (AGL, Curly Coated Retriever Variant)	Clear
Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM, Whippet and English Springer Spaniel Variant)	Clear



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## **OTHER RESULTS**

Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM, Wachtelhund Variant)	Clear
	Clear
Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)	Clear
	Clear
Goniodysgenesis and Glaucoma, Pectinate Ligament Dysplasia, PLD (OLFM3)	Clear
Hemophilia A (F8 Exon 11, German Shepherd Variant 1)	Clear
Hemophilia A (F8 Exon 1, German Shepherd Variant 2)	Clear
Hemophilia A (F8 Exon 10, Boxer Variant)	Clear
Hemophilia B (F9 Exon 7, Terrier Variant)	Clear
Hemophilia B (F9 Exon 7, Rhodesian Ridgeback Variant)	Clear
Hereditary Ataxia (PNPLA8, Australian Shepherd Variant)	Clear
Hereditary Ataxia, Cerebellar Degeneration (RAB24, Old English Sheepdog and Gordon Setter Variant)	Clear
Hereditary Cataracts (HSF4 Exon 9, Australian Shepherd Variant)	Clear
Hereditary Cataracts (FYCO1, Wirehaired Pointing Griffon Variant)	Clear



## **PINE CREEK GRACIE**



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## **OTHER RESULTS**

Hereditary Cerebellar Ataxia (SELENOP, Belgian Shepherd Variant)	Clear
Hereditary Footpad Hyperkeratosis (FAM83G, Terrier and Kromfohrlander Variant)	Clear
Hereditary Footpad Hyperkeratosis (DSG1, Rottweiler Variant)	Clear
Hereditary Nasal Parakeratosis (SUV39H2 Intron 4, Greyhound Variant)	Clear
Hereditary Nasal Parakeratosis, HNPK (SUV39H2)	Clear
Hereditary Vitamin D-Resistant Rickets (VDR)	Clear
Hypocatalasia, Acatalasemia (CAT)	Clear
Hypomyelination and Tremors (FNIP2, Weimaraner Variant)	Clear
Hypophosphatasia (ALPL Exon 9, Karelian Bear Dog Variant)	Clear
O Ichthyosis (NIPAL4, American Bulldog Variant)	Clear
O Ichthyosis (ASPRV1 Exon 2, German Shepherd Variant)	Clear
O Ichthyosis (SLC27A4, Great Dane Variant)	Clear
Olichthyosis, Epidermolytic Hyperkeratosis (KRT10, Terrier Variant)	Clear
O Ichthyosis, ICH1 (PNPLA1, Golden Retriever Variant)	Clear
O Ichthyosis, ICH2 (ABHD5, Golden Retriever Variant)	Clear
✓ Inflammatory Myopathy (SLC25A12)	Clear
Inherited Myopathy of Great Danes (BIN1)	Clear
Inherited Selected Cobalamin Malabsorption with Proteinuria (CUBN, Komondor Variant)	Clear



## **PINE CREEK GRACIE**



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## **OTHER RESULTS**

Intervertebral Disc Disease (Type I) (FGF4 retrogene - CFA12)	Clear
Intestinal Lipid Malabsorption (ACSL5, Australian Kelpie)	Clear
Junctional Epidermolysis Bullosa (LAMA3 Exon 66, Australian Cattle Dog Variant)	Clear
Junctional Epidermolysis Bullosa (LAMB3 Exon 11, Australian Shepherd Variant)	Clear
	Clear
Juvenile Laryngeal Paralysis and Polyneuropathy (RAB3GAP1, Rottweiler Variant)	Clear
Juvenile Myoclonic Epilepsy (DIRAS1)	Clear
L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH, Staffordshire Bull Terrier Variant)	Clear
	Clear
Laryngeal Paralysis (RAPGEF6, Miniature Bull Terrier Variant)	Clear
Laryngeal Paralysis and Polyneuropathy (CNTNAP1, Leonberger, Saint Bernard, and Labrador Retriever variant)	Clear
Late Onset Spinocerebellar Ataxia (CAPN1)	Clear
Late-Onset Neuronal Ceroid Lipofuscinosis, NCL 12 (ATP13A2, Australian Cattle Dog Variant)	Clear
Leonberger Polyneuropathy 1 (LPN1, ARHGEF10)	Clear
	Clear
	Clear
Leukodystrophy (TSEN54 Exon 5, Standard Schnauzer Variant)	Clear
	Clear



## **PINE CREEK GRACIE**



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# **OTHER RESULTS**

Limb Girdle Muscular Dystrophy (SGCD, Boston Terrier Variant)	Clear
	Clear
O Long QT Syndrome (KCNQ1)	Clear
Lundehund Syndrome (LEPREL1)	Clear
Macular Corneal Dystrophy, MCD (CHST6)	Clear
Malignant Hyperthermia (RYR1)	Clear
May-Hegglin Anomaly (MYH9)	Clear
Medium-Chain Acyl-CoA Dehydrogenase Deficiency, MCADD (ACADM, Cavalier King Charles Spaniel Variant)	Clear
Methemoglobinemia (CYB5R3, Pit Bull Terrier Variant)	Clear
Methemoglobinemia (CYB5R3)	Clear
Microphthalmia (RBP4 Exon 2, Soft Coated Wheaten Terrier Variant)	Clear
Mucopolysaccharidosis IIIB, Sanfilippo Syndrome Type B, MPS IIIB (NAGLU, Schipperke Variant)	Clear
Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6, Dachshund Variant)	Clear
Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6, New Zealand Huntaway Variant)	Clear
Mucopolysaccharidosis Type VI, Maroteaux-Lamy Syndrome, MPS VI (ARSB Exon 5, Miniature Pinscher Variant)	Clear
Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3, German Shepherd Variant)	Clear
Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5, Terrier Brasileiro Variant)	Clear
Multiple Drug Sensitivity (ABCB1)	Clear

## **PINE CREEK GRACIE**



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## **OTHER RESULTS**

OTHER REGGERO	
Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)	Clear
Muscular Dystrophy (DMD, Golden Retriever Variant)	Clear
Muscular Dystrophy-Dystroglycanopathy (LARGE1, Labrador Retriever Variant)	Clear
Musladin-Lueke Syndrome, MLS (ADAMTSL2)	Clear
Myasthenia Gravis-Like Syndrome (CHRNE, Heideterrier Variant)	Clear
Myotonia Congenita (CLCN1 Exon 23, Australian Cattle Dog Variant)	Clear
Myotonia Congenita (CLCN1 Exon 19, Labrador Retriever Variant)	Clear
Myotonia Congenita (CLCN1 Exon 7, Miniature Schnauzer Variant)	Clear
Narcolepsy (HCRTR2 Exon 1, Dachshund Variant)	Clear
Narcolepsy (HCRTR2 Intron 4, Doberman Pinscher Variant)	Clear
Narcolepsy (HCRTR2 Intron 6, Labrador Retriever Variant)	Clear
Nemaline Myopathy (NEB, American Bulldog Variant)	Clear
Neonatal Cerebellar Cortical Degeneration (SPTBN2, Beagle Variant)	Clear
Neonatal Encephalopathy with Seizures, NEWS (ATF2)	Clear
Neonatal Interstitial Lung Disease (LAMP3)	Clear
Neuroaxonal Dystrophy, NAD (VPS11, Rottweiler Variant)	Clear
Neuroaxonal Dystrophy, NAD (TECPR2, Spanish Water Dog Variant)	Clear
Neuronal Ceroid Lipofuscinosis 1, NCL 1 (PPT1 Exon 8, Dachshund Variant 1)	Clear



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## **OTHER RESULTS**

Neuronal Ceroid Lipofuscinosis 10, NCL 10 (CTSD Exon 5, American Bulldog Variant)	Clear
Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4, Dachshund Variant 2)	Clear
Neuronal Ceroid Lipofuscinosis 5, NCL 5 (CLN5 Exon 4 SNP, Border Collie Variant)	Clear
Neuronal Ceroid Lipofuscinosis 5, NCL 5 (CLN5 Exon 4 Deletion, Golden Retriever Variant)	Clear
Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7, Australian Shepherd Variant)	Clear
Neuronal Ceroid Lipofuscinosis 7, NCL 7 (MFSD8, Chihuahua and Chinese Crested Variant)	Clear
Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8, Australian Shepherd Variant)	Clear
Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 Exon 2, English Setter Variant)	Clear
Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 Insertion, Saluki Variant)	Clear
Neuronal Ceroid Lipofuscinosis, Cerebellar Ataxia, NCL4A (ARSG Exon 2, American Staffordshire Terrier Variant)	Clear
	Clear
Variant)	
Variant)  Oculocutaneous Albinism, OCA (SLC45A2 Exon 6, Bullmastiff Variant)	Clear
Variant)  Oculocutaneous Albinism, OCA (SLC45A2 Exon 6, Bullmastiff Variant)  Oculocutaneous Albinism, OCA (SLC45A2, Small Breed Variant)	Clear Clear
Variant)  Oculocutaneous Albinism, OCA (SLC45A2 Exon 6, Bullmastiff Variant)  Oculocutaneous Albinism, OCA (SLC45A2, Small Breed Variant)  Oculoskeletal Dysplasia 2 (COL9A2, Samoyed Variant)	Clear Clear Clear
Variant)  ✓ Oculocutaneous Albinism, OCA (SLC45A2 Exon 6, Bullmastiff Variant)  ✓ Oculocutaneous Albinism, OCA (SLC45A2, Small Breed Variant)  ✓ Oculoskeletal Dysplasia 2 (COL9A2, Samoyed Variant)  ✓ Osteochondrodysplasia (SLC13A1, Poodle Variant)	Clear Clear Clear Clear
Variant)  ✓ Oculocutaneous Albinism, OCA (SLC45A2 Exon 6, Bullmastiff Variant)  ✓ Oculocutaneous Albinism, OCA (SLC45A2, Small Breed Variant)  ✓ Oculoskeletal Dysplasia 2 (COL9A2, Samoyed Variant)  ✓ Osteochondrodysplasia (SLC13A1, Poodle Variant)  ✓ Osteogenesis Imperfecta (COL1A2, Beagle Variant)	Clear Clear Clear Clear



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## **OTHER RESULTS**

Pachyonychia Congenita (KRT16, Dogue de Bordeaux Variant)	Clear
Paroxysmal Dyskinesia, PxD (PIGN)	Clear
Persistent Mullerian Duct Syndrome, PMDS (AMHR2)	Clear
Pituitary Dwarfism (POU1F1 Intron 4, Karelian Bear Dog Variant)	Clear
	Clear
OPOlycystic Kidney Disease, PKD (PKD1)	Clear
Pompe's Disease (GAA, Finnish and Swedish Lapphund, Lapponian Herder Variant)	Clear
Prekallikrein Deficiency (KLKB1 Exon 8)	Clear
Primary Ciliary Dyskinesia, PCD (NME5, Alaskan Malamute Variant)	Clear
Primary Ciliary Dyskinesia, PCD (STK36, Australian Shepherd Variant)	Clear
Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3, Old English Sheepdog Variant)	Clear
Primary Hyperoxaluria (AGXT)	Clear
Primary Lens Luxation (ADAMTS17)	Clear
Primary Open Angle Glaucoma (ADAMTS17 Exon 11, Basset Fauve de Bretagne Variant)	Clear
Primary Open Angle Glaucoma (ADAMTS10 Exon 17, Beagle Variant)	Clear
Primary Open Angle Glaucoma (ADAMTS10 Exon 9, Norwegian Elkhound Variant)	Clear
Primary Open Angle Glaucoma and Primary Lens Luxation (ADAMTS17 Exon 2, Chinese Shar-Pei Variant)	Clear
Progressive Retinal Atrophy (SAG)	Clear



### **PINE CREEK GRACIE**



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## **OTHER RESULTS**

Progressive Retinal Atrophy (IFT122 Exon 26, Lapponian Herder Variant)	Clear
Progressive Retinal Atrophy 5, PRA5 (NECAP1 Exon 6, Giant Schnauzer Variant)	Clear
Progressive Retinal Atrophy, Bardet-Biedl Syndrome (BBS2 Exon 11, Shetland Sheepdog Variant)	Clear
Progressive Retinal Atrophy, CNGA (CNGA1 Exon 9)	Clear
Progressive Retinal Atrophy, crd1 (PDE6B, American Staffordshire Terrier Variant)	Clear
Progressive Retinal Atrophy, crd4/cord1 (RPGRIP1)	Clear
Progressive Retinal Atrophy, PRA1 (CNGB1)	Clear
Progressive Retinal Atrophy, PRA3 (FAM161A)	Clear
Progressive Retinal Atrophy, prcd (PRCD Exon 1)	Clear
Progressive Retinal Atrophy, rcd1 (PDE6B Exon 21, Irish Setter Variant)	Clear
Progressive Retinal Atrophy, rcd3 (PDE6A)	Clear
Proportionate Dwarfism (GH1 Exon 5, Chihuahua Variant)	Clear
Protein Losing Nephropathy, PLN (NPHS1)	Clear
Pyruvate Dehydrogenase Deficiency (PDP1, Spaniel Variant)	Clear
Pyruvate Kinase Deficiency (PKLR Exon 5, Basenji Variant)	Clear
Pyruvate Kinase Deficiency (PKLR Exon 7, Beagle Variant)	Clear
Pyruvate Kinase Deficiency (PKLR Exon 10, Terrier Variant)	Clear
Pyruvate Kinase Deficiency (PKLR Exon 7, Labrador Retriever Variant)	Clear



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## **OTHER RESULTS**

Pyruvate Kinase Deficiency (PKLR Exon 7, Pug Variant)	Clear
Raine Syndrome (FAM20C)	Clear
Recurrent Inflammatory Pulmonary Disease, RIPD (AKNA, Rough Collie Variant)	Clear
Renal Cystadenocarcinoma and Nodular Dermatofibrosis (FLCN Exon 7)	Clear
Retina Dysplasia and/or Optic Nerve Hypoplasia (SIX6 Exon 1, Golden Retriever Variant)	Clear
Sensory Neuropathy (FAM134B, Border Collie Variant)	Clear
Severe Combined Immunodeficiency, SCID (PRKDC, Terrier Variant)	Clear
Severe Combined Immunodeficiency, SCID (RAG1, Wetterhoun Variant)	Clear
Shaking Puppy Syndrome (PLP1, English Springer Spaniel Variant)	Clear
Shar-Pei Autoinflammatory Disease, SPAID, Shar-Pei Fever (MTBP)	Clear
Skeletal Dysplasia 2, SD2 (COL11A2, Labrador Retriever Variant)	Clear
Skin Fragility Syndrome (PKP1, Chesapeake Bay Retriever Variant)	Clear
Spinocerebellar Ataxia (SCN8A, Alpine Dachsbracke Variant)	Clear
Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10)	Clear
Spongy Degeneration with Cerebellar Ataxia 1 (KCNJ10)	Clear
Spongy Degeneration with Cerebellar Ataxia 2 (ATP1B2)	Clear
Stargardt Disease (ABCA4 Exon 28, Labrador Retriever Variant)	Clear
Succinic Semialdehyde Dehydrogenase Deficiency (ALDH5A1 Exon 7, Saluki Variant)	Clear



## **PINE CREEK GRACIE**



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## **OTHER RESULTS**

✓ Thrombopathia (RASGRP1 Exon 5, American Eskimo Dog Variant)       Clear         ✓ Thrombopathia (RASGRP1 Exon 5, Basset Hound Variant)       Clear         ✓ Thrombopathia (RASGRP1 Exon 8, Landseer Variant)       Clear         ✓ Trapped Neutrophil Syndrome, TNS (VPS13B)       Clear         ✓ Ullrich-like Congenital Muscular Dystrophy (COL6A3 Exon 10, Labrador Retriever Variant)       Clear         ✓ Ullrich-like Congenital Muscular Dystrophy (COL6A1 Exon 3, Landseer Variant)       Clear         ✓ Unilateral Deafness and Vestibular Syndrome (PTPRQ Exon 39, Doberman Pinscher)       Clear         ✓ Urate Kidney & Bladder Stones (SLC2A9)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF, Pointer Variant)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)       Clear         ✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)       Clear         ✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)       Clear         ✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)       Clear         ✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)       Clear         ✓ X-linked Severe		
✓ Thrombopathia (RASGRP1 Exon 8, Landseer Variant)       Clear         ✓ Trapped Neutrophil Syndrome, TNS (VPS13B)       Clear         ✓ Ullrich-like Congenital Muscular Dystrophy (COL6A3 Exon 10, Labrador Retriever Variant)       Clear         ✓ Ullrich-like Congenital Muscular Dystrophy (COL6A1 Exon 3, Landseer Variant)       Clear         ✓ Unilateral Deafness and Vestibular Syndrome (PTPRQ Exon 39, Doberman Pinscher)       Clear         ✓ Urate Kidney & Bladder Stones (SLC2A9)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF, Pointer Variant)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)       Clear         ✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)       Clear         ✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)       Clear         ✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)       Clear         ✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)       Clear         ✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)       Clear	Thrombopathia (RASGRP1 Exon 5, American Eskimo Dog Variant)	Clear
<ul> <li>✓ Trapped Neutrophil Syndrome, TNS (VPS13B)</li> <li>✓ Ullrich-like Congenital Muscular Dystrophy (COL6A3 Exon 10, Labrador Retriever Variant)</li> <li>✓ Ullrich-like Congenital Muscular Dystrophy (COL6A1 Exon 3, Landseer Variant)</li> <li>✓ Unilateral Deafness and Vestibular Syndrome (PTPRQ Exon 39, Doberman Pinscher)</li> <li>✓ Urate Kidney &amp; Bladder Stones (SLC2A9)</li> <li>✓ Urate Kidney &amp; Bladder Stones (SLC2A9)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF, Pointer Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Intron 16, Nederlandse Kooikerhondje Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)</li> <li>✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)</li> <li>✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>✓ X-Linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ Clear</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ Clear</li> </ul>	Thrombopathia (RASGRP1 Exon 5, Basset Hound Variant)	Clear
<ul> <li>✓ Ullrich-like Congenital Muscular Dystrophy (COL6A3 Exon 10, Labrador Retriever Variant)</li> <li>✓ Clear</li> <li>✓ Ullrich-like Congenital Muscular Dystrophy (COL6A1 Exon 3, Landseer Variant)</li> <li>✓ Unilateral Deafness and Vestibular Syndrome (PTPRQ Exon 39, Doberman Pinscher)</li> <li>✓ Clear</li> <li>✓ Urate Kidney &amp; Bladder Stones (SLC2A9)</li> <li>✓ Von Willebrand Disease Type II, Type II vWD (VWF, Pointer Variant)</li> <li>✓ Clear</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)</li> <li>✓ Clear</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Intron 16, Nederlandse Kooikerhondje Variant)</li> <li>✓ Clear</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)</li> <li>✓ Clear</li> <li>✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)</li> <li>✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>✓ Clear</li> <li>✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>✓ X-Linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ Clear</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ Clear</li> </ul>	Thrombopathia (RASGRP1 Exon 8, Landseer Variant)	Clear
<ul> <li>Ullrich-like Congenital Muscular Dystrophy (COL6A1 Exon 3, Landseer Variant)</li> <li>Clear</li> <li>Unilateral Deafness and Vestibular Syndrome (PTPRQ Exon 39, Doberman Pinscher)</li> <li>Urate Kidney &amp; Bladder Stones (SLC2A9)</li> <li>Clear</li> <li>Von Willebrand Disease Type II, Type II vWD (VWF, Pointer Variant)</li> <li>Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)</li> <li>Clear</li> <li>Von Willebrand Disease Type III, Type III vWD (VWF Intron 16, Nederlandse Kooikerhondje Variant)</li> <li>Clear</li> <li>Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)</li> <li>Clear</li> <li>X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)</li> <li>X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>Clear</li> <li>X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> <li>Clear</li> </ul>		Clear
<ul> <li>✓ Unilateral Deafness and Vestibular Syndrome (PTPRQ Exon 39, Doberman Pinscher)</li> <li>✓ Urate Kidney &amp; Bladder Stones (SLC2A9)</li> <li>✓ Von Willebrand Disease Type II, Type II vWD (VWF, Pointer Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Intron 16, Nederlandse Kooikerhondje Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)</li> <li>✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)</li> <li>✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ Clear</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> <li>✓ Clear</li> </ul>	Ullrich-like Congenital Muscular Dystrophy (COL6A3 Exon 10, Labrador Retriever Variant)	Clear
<ul> <li>✓ Urate Kidney &amp; Bladder Stones (SLC2A9)</li> <li>✓ Von Willebrand Disease Type II, Type II vWD (VWF, Pointer Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Intron 16, Nederlandse Kooikerhondje Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)</li> <li>✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)</li> <li>✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ Clear</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> <li>✓ Clear</li> </ul>	Ullrich-like Congenital Muscular Dystrophy (COL6A1 Exon 3, Landseer Variant)	Clear
✓ Von Willebrand Disease Type III, Type III vWD (VWF, Pointer Variant)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF Intron 16, Nederlandse Kooikerhondje Variant)       Clear         ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)       Clear         ✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)       Clear         ✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)       Clear         ✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)       Clear         ✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)       Clear         ✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)       Clear	Unilateral Deafness and Vestibular Syndrome (PTPRQ Exon 39, Doberman Pinscher)	Clear
<ul> <li>✓ Von Willebrand Disease Type III, Type III vWD (vWF Exon 4, Terrier Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (vWF Intron 16, Nederlandse Kooikerhondje Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (vWF Exon 7, Shetland Sheepdog Variant)</li> <li>✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)</li> <li>✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> </ul>	Urate Kidney & Bladder Stones (SLC2A9)	Clear
<ul> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Intron 16, Nederlandse Kooikerhondje Variant)</li> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)</li> <li>✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)</li> <li>✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> </ul>		Clear
<ul> <li>✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)</li> <li>✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)</li> <li>✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> </ul>		Clear
<ul> <li>✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)</li> <li>✓ X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>✓ X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> </ul>	✓ Von Willebrand Disease Type III, Type III vWD (VWF Intron 16, Nederlandse Kooikerhondje Variant)	Clear
<ul> <li>X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)</li> <li>X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)</li> <li>X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> <li>Clear</li> </ul>	On Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)	Clear
<ul> <li></li></ul>	X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)	Clear
<ul> <li>X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)</li> <li>X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> </ul> Clear	X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)	Clear
<ul> <li>X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)</li> </ul>	X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)	Clear
	X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)	Clear
Xanthine Urolithiasis (XDH, Mixed Breed Variant)  Clear	X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)	Clear
	Xanthine Urolithiasis (XDH, Mixed Breed Variant)	Clear



### **PINE CREEK GRACIE**



**DNA Test Report** Test Date: December 10th, 2024 embk.me/pinecreekgracie

## **OTHER RESULTS**

β-Mannosidosis (MANBA Exon 16, Mixed-Breed Variant)

Clear

Mast Cell Tumor No result



### PINE CREEK GRACIE



**DNA Test Report** Test Date: December 10th, 2024 embk.me/pinecreekgracie

### **HEALTH REPORT**



Notable result

#### Copper Toxicosis (Attenuating)

Pine Creek Gracie inherited one copy of the variant we tested for Copper Toxicosis (Attenuating)

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

Gracie has a genotype at the ATP7A gene that modifies and may help mitigate some of the symptoms from dogs with variants at ATP7B (https://my.embarkvet.com/members/results/health/condition/140102?i=34). This variant is not associated with an increased risk of any disease. As this variant resides on the X- chromosome, male dogs with one copy of the variant are better protected from copper accumulation due to the ATP7B variant than female dogs with one copy of the variant.

#### What is Copper Toxicosis (Attenuating)?

The ATP7A variant is considered beneficial and may be best described as a helpful modifier of the harmful copper toxicosis variant ATP7B (https://my.embarkvet.com/members/results/health/condition/140102?i=34). The ATP7A variant may help mitigate some of the symptoms of dogs with variants at ATP7B. Dogs with the ATP7A variant have not been observed to have any beneficial or harmful complications if they have two copies of the normal ATP7B variant.

#### When signs & symptoms develop in affected dogs

A variant in this gene may delay or have no effect on the onset of clinical signs of copper toxicosis in dogs with the ATP7B (https://my.embarkvet.com/members/results/health/condition/140102?i=34) variant. If your dog has the ATP7B variant, please read more about the age of onset on the ATP7B page.

#### How vets diagnose this condition

No diagnostics are required for this variant. If your dog has the ATP7B

(https://my.embarkvet.com/members/results/health/condition/140102?i=34) variant, please read what diagnostics may be considered on the ATP7B page.

#### How this condition is treated

No treatment is required for this variant. If your dog has the ATP7B

(https://my.embarkvet.com/members/results/health/condition/140102?i=34) variant, please read the available treatment on the ATP7B page.

#### Actions to take if your dog is affected

· No actions are required for dogs with this variant. If your dog has the ATP7B (https://my.embarkvet.com/members/results/health/condition/140102?i=34) variant, please read what actions you can take on the ATP7B page.





### PINE CREEK GRACIE



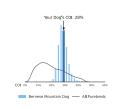
DNA Test Report Test Date: December 10th, 2024 embk.me/pinecreekgracie

### INBREEDING AND DIVERSITY

CATEGORY RESULT

#### **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.



#### 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**

28%

How common is this amount of diversity in purebreds:

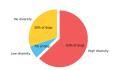


#### MHC Class II - DLA DQA1 and DQB1

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.

### **High Diversity**

How common is this amount of diversity in purebreds:



Registration: American Kennel Club

(AKC)