ALLIE

DNA Test Report | Test Date: April 30th, 2022 | embk.me/allie641

BREED ANCESTRY

Golden Retriever : 100.0%

GENETIC STATS

Predicted adult weight: **62 lbs**
Genetic age: n/a (Date of birth unknown)

TEST DETAILS

Kit number: EM-78830466
Swab number: 31210752615479

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Fun Fact

A Golden Retriever is also pictured in the Guinness Book of World's Records for "Most tennis balls held in mouth" (with 6).

GOLDEN RETRIEVER

The Golden Retriever was developed in the early 19th century as an ideal hunting companion, able to retrieve birds on both land and water in the marshy Scottish countryside. Their friendliness and intelligence makes the both a popular family pet and an excellent working dog, well suited for being a service dog, therapy dog or for search and rescue. The third most popular breed in the US, the American and Canadian Goldens are generally lankier and darker than their British counterparts. Their wavy, feathered topcoat is water resistant, their undercoat helps them with thermoregulation and both coats have a tendency for heavy seasonal shedding. Goldens need lots of exercise (especially when younger), and their love of play and water means their owners usually get a lot of exercise too! In 2013, the 100th anniversary of Britain's Golden Retriever Club, Goldens from around the world came made the pilgrimage to the breed's birthplace in Scotland, where 222 of them posed in a single record-breaking photo. At the same time, the Golden Retriever Lifetime Study was getting started in the United States, recruiting 3,000 Golden Retrievers for a lifetime study aimed at understanding how genetics, lifestyle and environment influences healthy aging and cancer risk in Goldens.

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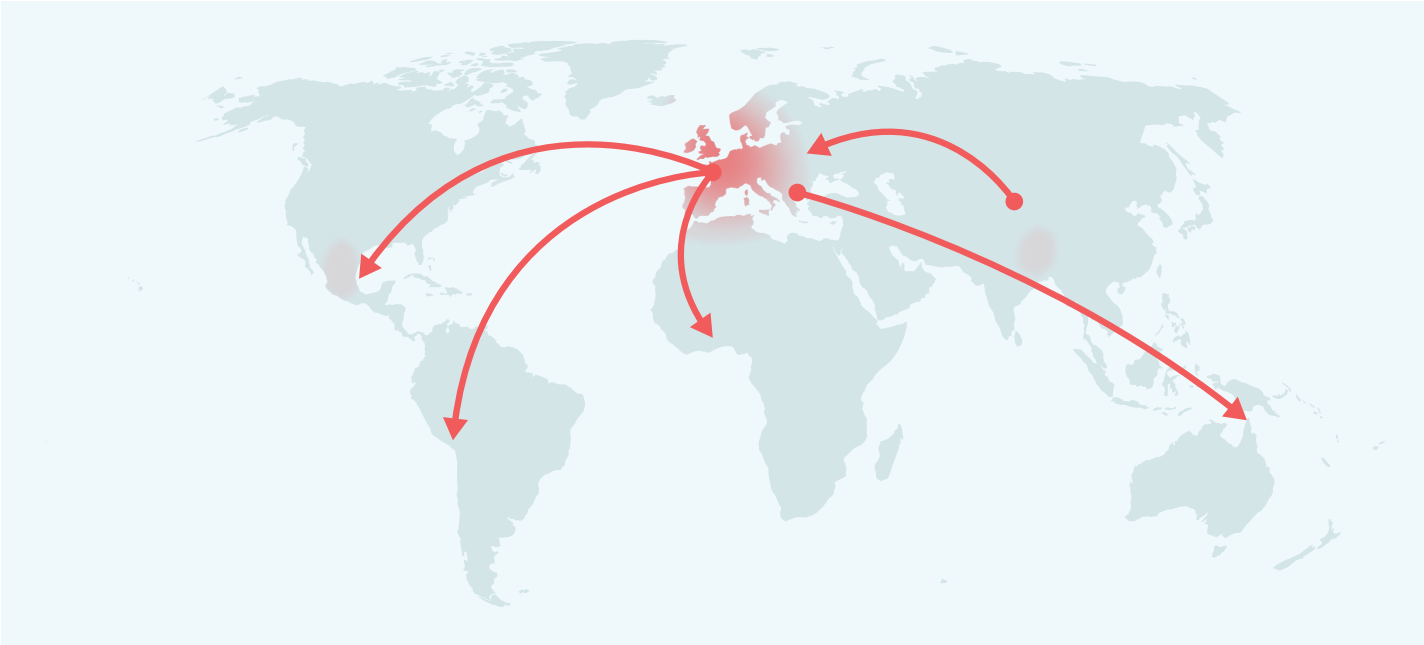
MATERNAL LINE



Through Allie'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: B1

B1 is the second most common maternal lineage in breeds of European or American origin. It is the female line of the majority of Golden Retrievers, Basset Hounds, and Shih Tzus, and about half of Beagles, Pekingese and Toy Poodles. This lineage is also somewhat common among village dogs that carry distinct ancestry from these breeds. We know this is a result of B1 dogs being common amongst the European dogs that their conquering owners brought around the world, because nowhere on earth is it a very common lineage in village dogs. It even enables us to trace the path of (human) colonization: Because most Bichons are B1 and Bichons are popular in Spanish culture, B1 is now fairly common among village dogs in Latin America.

HAPLOTYPE: B84

Part of the large B1 haplogroup, this haplotype occurs most frequently in Golden Retrievers, Beagles, and Staffordshire Terriers.

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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 hairs anywhere (ee)**
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 **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^yk^y genotype will show a coat color pattern based on the genotype they have at the A Locus. Dogs who test as K^Bk^y may be brindle rather than black or brown.	**Not expressed (K^Bk^y)**

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 pigmented 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 $\mathbf{k^y k^y}$ 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.	**Not expressed ($a^t a^t$)**
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.	**Not expressed (DD)**

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 **Bbb** 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".	**Likely black colored nose/feet (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 $\mathbf{a^t}$ allele, so dogs that do not express $\mathbf{a^t}$ are not influenced by this gene.	**Not expressed (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)**

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. 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.	**No merle alleles (mm)**
R Locus (USH2A) LINKAGE 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: OTHER COAT TRAITS

TRAIT	RESULT
Furnishings (RSPO2) LINKAGE 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)**
Coat Length (FGF5) The FGF5 gene is known to affect hair length in many different species, including cats, dogs, mice, and humans. In dogs, the **T** allele confers a long, silky haircoat as observed in the Yorkshire Terrier and the Long Haired Whippet. The ancestral **G** 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."	**Likely long coat (TT)**
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)**
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)**

TRAITS: OTHER COAT TRAITS (CONTINUED)

TRAIT	RESULT
Oculocutaneous Albinism Type 2 (SLC45A2) LINKAGE 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)**
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)**

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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 **C** allele. Dogs in many short-length muzzle (brachycephalic) breeds such as the English Bulldog, Pug, and Pekingese have two copies of the derived **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.	**Unlikely to have hind dew claws (CC)**

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)

Allie's baseline ALT level may be Low Normal

Why is this important to your vet?
Allie has one copy of a variant associated with reduced ALT activity as measured on veterinary blood chemistry panels. Please inform your veterinarian that Allie has this genotype, as ALT is often used as an indicator of liver health and Allie is likely to have a lower than average resting ALT activity. As such, an increase in Allie'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 Allie's genetic health results:
If Allie 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 Allie 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!

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

Breed-Relevant Genetic Conditions	**10 variants not detected**
Additional Genetic Conditions	**209 variants not detected**

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BREED-RELEVANT CONDITIONS TESTED

Allie did not have the variants that we tested for, that are relevant to her breed:

- Progressive Retinal Atrophy, prcd (PRCD Exon 1)
- Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)
- Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)
- Neuronal Ceroid Lipofuscinosis 5, NCL 5 (CLN5 Exon 4 Deletion, Golden Retriever Variant)
- Degenerative Myelopathy, DM (SOD1A)
- Muscular Dystrophy (DMD, Golden Retriever Variant)
- Congenital Myasthenic Syndrome, CMS (COLQ, Golden Retriever Variant)
- Dystrophic Epidermolysis Bullosa (COL7A1, Golden Retriever Variant)
- Ichthyosis, ICH1 (PNPLA1, Golden Retriever Variant)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1, Golden Retriever Variant)

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ADDITIONAL CONDITIONS TESTED

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

- MDR1 Drug Sensitivity (ABCB1)
- 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 VII Deficiency (F7 Exon 5)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 10, Boxer Variant)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 11, German Shepherd Variant 1)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 1, German Shepherd Variant 2)
- Thrombopathia (RASGRP1 Exon 5, Basset Hound Variant)
- Thrombopathia (RASGRP1 Exon 8, Landseer Variant)
- Thrombopathia (RASGRP1 Exon 5, American Eskimo Dog Variant)
- Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)
- Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)
- Von Willebrand Disease Type I, Type I vWD (VWF)
- Von Willebrand Disease Type II, Type II vWD (VWF, Pointer Variant)
- Canine Leukocyte Adhesion Deficiency Type I, CLAD I (ITGB2, Setter Variant)
- Canine Leukocyte Adhesion Deficiency Type III, CLAD III (FERMT3, German Shepherd Variant)
- Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant)
- Canine Elliptocytosis (SPTB Exon 30)
- Glanzmann's Thrombasthenia Type I (ITGA2B Exon 13, Great Pyrenees Variant)
- Glanzmann's Thrombasthenia Type I (ITGA2B Exon 12, Otterhound Variant)
- May-Hegglin Anomaly (MYH9)
- Prekallikrein Deficiency (KLKB1 Exon 8)

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ADDITIONAL CONDITIONS TESTED

- Pyruvate Kinase Deficiency (PKLR Exon 7, Pug Variant)
- Pyruvate Kinase Deficiency (PKLR Exon 7, Beagle Variant)
- Pyruvate Kinase Deficiency (PKLR Exon 10, Terrier Variant)
- Trapped Neutrophil Syndrome, TNS (VPS13B)
- Ligneous Membranitis, LM (PLG)
- Platelet Factor X Receptor Deficiency, Scott Syndrome (TMEM16F)
- Methemoglobinemia (CYB5R3)
- Bernard-Soulier Syndrome, BSS (GP9, Cocker Spaniel Variant)
- Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)
- Congenital Hypothyroidism (TPO, Rat, Toy, Hairless Terrier Variant)
- Congenital Dyshormonogenic Hypothyroidism with Goiter (SLC5A5, Shih Tzu Variant)
- Complement 3 Deficiency, C3 Deficiency (C3)
- Severe Combined Immunodeficiency, SCID (PRKDC, Terrier Variant)
- Severe Combined Immunodeficiency, SCID (RAG1, Wetterhoun Variant)
- X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)
- X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)
- 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, PRA1 (CNGB1)
- Progressive Retinal Atrophy (SAG)
- Progressive Retinal Atrophy, crd1 (PDE6B, American Staffordshire Terrier Variant)
- Progressive Retinal Atrophy, crd4/cord1 (RPGRIP1)
- X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)
- Progressive Retinal Atrophy, PRA3 (FAM161A)

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ADDITIONAL CONDITIONS TESTED

- Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1)
- Day Blindness, Cone Degeneration, Achromatopsia (CNGB3 Deletion, Alaskan Malamute Variant)
- Day Blindness, Cone Degeneration, Achromatopsia (CNGB3 Exon 6, German Shorthaired Pointer Variant)
- Achromatopsia (CNGA3 Exon 7, German Shepherd Variant)
- Achromatopsia (CNGA3 Exon 7, Labrador Retriever Variant)
- Autosomal Dominant Progressive Retinal Atrophy (RHO)
- Canine Multifocal Retinopathy, cmr1 (BEST1 Exon 2)
- Canine Multifocal Retinopathy, cmr2 (BEST1 Exon 5, Coton de Tulear Variant)
- Canine Multifocal Retinopathy, cmr3 (BEST1 Exon 10 Deletion, Finnish and Swedish Lapphund, Lapponian Herder Variant)
- Primary Open Angle Glaucoma (ADAMTS10 Exon 9, Norwegian Elkhound Variant)
- Primary Open Angle Glaucoma (ADAMTS10 Exon 17, Beagle Variant)
- Primary Open Angle Glaucoma (ADAMTS17 Exon 11, Basset Fauve de Bretagne Variant)
- Primary Open Angle Glaucoma and Primary Lens Luxation (ADAMTS17 Exon 2, Chinese Shar-Pei Variant)
- Goniodysgenesis and Glaucoma, Pectinate Ligament Dysplasia, PLD (OLFM3)
- Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9, Australian Shepherd Variant)
- Primary Lens Luxation (ADAMTS17)
- Congenital Stationary Night Blindness (RPE65, Briard Variant)
- Congenital Stationary Night Blindness (LRIT3, Beagle Variant)
- Macular Corneal Dystrophy, MCD (CHST6)
- 2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis (APRT)
- Cystinuria Type I-A (SLC3A1, Newfoundland Variant)
- Cystinuria Type II-A (SLC3A1, Australian Cattle Dog Variant)
- Cystinuria Type II-B (SLC7A9, Miniature Pinscher Variant)
- Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU (SLC2A9)
- Polycystic Kidney Disease, PKD (PKD1)

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ADDITIONAL CONDITIONS TESTED

- 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 30, English Springer Spaniel Variant)
- Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 3, Cocker Spaniel Variant)
- Fanconi Syndrome (FAN1, Basenji Variant)
- Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3, Old English Sheepdog Variant)
- Primary Ciliary Dyskinesia, PCD (NME5, Alaskan Malamute Variant)
- Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatosis, Dry Eye Curly Coat Syndrome, CKCSID (FAM83H Exon 5)
- X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia, XHED (EDA Intron 8)
- Renal Cystadenocarcinoma and Nodular Dermatofibrosis, RCND (FLCN Exon 7)
- Canine Fucosidosis (FUCA1)
- Glycogen Storage Disease Type II, Pompe's Disease, GSD II (GAA, Finnish and Swedish Lapphund, Lapponian Herder Variant)
- Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC, Maltese Variant)
- Glycogen Storage Disease Type IIIA, GSD IIIA (AGL, Curly Coated Retriever Variant)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6, Dachshund Variant)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6, New Zealand Huntaway Variant)
- Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5, Terrier Brasileiro Variant)
- Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3, German Shepherd Variant)
- 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, Dachshund Variant 1)
- Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4, Dachshund Variant 2)
- Neuronal Ceroid Lipofuscinosis, Cerebellar Ataxia, NCL4A (ARSG Exon 2, American Staffordshire Terrier Variant)

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ADDITIONAL CONDITIONS TESTED

- Neuronal Ceroid Lipofuscinosis 5, NCL 5 (CLN5 Exon 4 SNP, Border Collie Variant)
- Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7, Australian Shepherd Variant)
- Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 Exon 2, English Setter Variant)
- Neuronal Ceroid Lipofuscinosis 7, NCL 7 (MFSD8, Chihuahua and Chinese Crested Variant)
- Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8, Australian Shepherd Variant)
- Neuronal Ceroid Lipofuscinosis 10, NCL 10 (CTSD Exon 5, American Bulldog Variant)
- Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 Insertion, Saluki Variant)
- Late-Onset Neuronal Ceroid Lipofuscinosis, NCL 12 (ATP13A2, Australian Cattle Dog Variant)
- GM1 Gangliosidosis (GLB1 Exon 15, Shiba Inu Variant)
- GM1 Gangliosidosis (GLB1 Exon 15, Alaskan Husky Variant)
- GM1 Gangliosidosis (GLB1 Exon 2, Portuguese Water Dog Variant)
- GM2 Gangliosidosis (HEXB, Poodle Variant)
- GM2 Gangliosidosis (HEXA, Japanese Chin Variant)
- Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5, Terrier Variant)
- Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (ENAM Deletion, Italian Greyhound Variant)
- Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (ENAM SNP, 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)
- Neonatal Interstitial Lung Disease (LAMP3)
- Recurrent Inflammatory Pulmonary Disease, RIPD (AKNA, Rough Collie Variant)
- Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy (SLC19A3)
- Alexander Disease (GFAP)
- Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration, NCCD (SPTBN2, Beagle Variant)
- Cerebellar Ataxia, Progressive Early-Onset Cerebellar Ataxia (SEL1L, Finnish Hound Variant)

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ADDITIONAL CONDITIONS TESTED

- Cerebellar Hypoplasia (VLDLR, Eurasier Variant)
- Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA (CAPN1)
- Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10)
- Hereditary Ataxia, Cerebellar Degeneration (RAB24, Old English Sheepdog and Gordon Setter Variant)
- Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LGI2)
- Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2, Giant Schnauzer Variant)
- Hypomyelination and Tremors (FNIP2, Weimaraner Variant)
- Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP1, English Springer Spaniel Variant)
- Neuroaxonal Dystrophy, NAD (TECPR2, Spanish Water Dog Variant)
- Neuroaxonal Dystrophy, NAD (VPS11, Rottweiler Variant)
- L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH, Staffordshire Bull Terrier Variant)
- Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- Alaskan Malamute Polyneuropathy, AMPN (NDRG1 SNP)
- Narcolepsy (HCRTR2 Intron 4, Doberman Pinscher Variant)
- Narcolepsy (HCRTR2 Intron 6, Labrador Retriever Variant)
- Narcolepsy (HCRTR2 Exon 1, Dachshund Variant)
- Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 15, Kerry Blue Terrier Variant)
- Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 4, Chinese Crested Variant)
- Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV (RAB3GAP1, Rottweiler Variant)
- Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (GDNF-AS, Spaniel and Pointer Variant)
- Sensory Neuropathy (FAM134B, Border Collie Variant)
- 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)

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ADDITIONAL CONDITIONS TESTED

- Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 (ATP1B2)
- Dilated Cardiomyopathy, DCM1 (PDK4, Doberman Pinscher Variant 1)
- Dilated Cardiomyopathy, DCM2 (TTN, Doberman Pinscher Variant 2)
- Long QT Syndrome (KCNQ1)
- Cardiomyopathy and Juvenile Mortality (YARS2)
- Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)
- Limb Girdle Muscular Dystrophy (SGCD, Boston Terrier Variant)
- Ullrich-like Congenital Muscular Dystrophy (COL6A3 Exon 10, Labrador Retriever Variant)
- Centronuclear Myopathy, CNM (PTPLA)
- Exercise-Induced Collapse, EIC (DNM1)
- Inherited Myopathy of Great Danes (BIN1)
- Myostatin Deficiency, Bully Whippet Syndrome (MSTN)
- Myotonia Congenita (CLCN1 Exon 7, Miniature Schnauzer Variant)
- Myotonia Congenita (CLCN1 Exon 23, Australian Cattle Dog Variant)
- Nemaline Myopathy (NEB, American Bulldog Variant)
- Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM (MTM1, Labrador Retriever Variant)
- Inflammatory Myopathy (SLC25A12)
- Hypocatalasia, Acatalasemia (CAT)
- Pyruvate Dehydrogenase Deficiency (PDP1, Spaniel Variant)
- Malignant Hyperthermia (RYR1)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53, Border Collie Variant)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8, Beagle Variant)
- Inherited Selected Cobalamin Malabsorption with Proteinuria (CUBN, Komondor Variant)
- Lundehund Syndrome (LEPREL1)
- Congenital Myasthenic Syndrome, CMS (CHAT, Old Danish Pointing Dog Variant)

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ADDITIONAL CONDITIONS TESTED

- Congenital Myasthenic Syndrome, CMS (COLQ, Labrador Retriever Variant)
- Congenital Myasthenic Syndrome, CMS (CHRNE, Jack Russell Terrier Variant)
- Myasthenia Gravis-Like Syndrome (CHRNE, Heideterrier Variant)
- Episodic Falling Syndrome (BCAN)
- Paroxysmal Dyskinesia, PxD (PIGN)
- Demyelinating Polyneuropathy (SBF2/MTRM13)
- Laryngeal Paralysis (RAPGEF6, Miniature Bull Terrier Variant)
- Dystrophic Epidermolysis Bullosa (COL7A1, Central Asian Shepherd Dog Variant)
- Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1, Chesapeake Bay Retriever Variant)
- Ichthyosis, Epidermolytic Hyperkeratosis (KRT10, Terrier Variant)
- Ichthyosis (SLC27A4, Great Dane Variant)
- Ichthyosis (NIPAL4, American Bulldog Variant)
- Focal Non-Epidermolytic Palmoplantar Keratoderma, Pachyonychia Congenita (KRT16, Dogue de Bordeaux Variant)
- Hereditary Footpad Hyperkeratosis (FAM83G, Terrier and Kromfohrlander Variant)
- Hereditary Footpad Hyperkeratosis (DSG1, Rottweiler Variant)
- Hereditary Nasal Parakeratosis, HNPK (SUV39H2)
- Musladin-Lueke Syndrome, MLS (ADAMTSL2)
- Oculocutaneous Albinism, OCA (SLC45A2, Small Breed Variant)
- Bald Thigh Syndrome (IGFBP5)
- Lethal Acrodermatitis, LAD (MKLN1)
- Ehlers Danlos (ADAMTS2, Doberman Pinscher Variant)
- Cleft Lip and/or Cleft Palate (ADAMTS20, Nova Scotia Duck Tolling Retriever Variant)
- Hereditary Vitamin D-Resistant Rickets (VDR)
- Oculoskeletal Dysplasia 2, Dwarfism-Retinal Dysplasia 2, drd2, OSD2 (COL9A2, Samoyed Variant)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2, Beagle Variant)

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ADDITIONAL CONDITIONS TESTED

- **Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1, Dachshund Variant)**
- **Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1, Poodle Variant)**
- **Skeletal Dysplasia 2, SD2 (COL11A2, Labrador Retriever Variant)**
- **Craniomandibular Osteopathy, CMO (SLC37A2)**
- **Raine Syndrome, Canine Dental Hypomineralization Syndrome (FAM20C)**
- **Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)**
- **Chondrodystrophy (ITGA10, Norwegian Elkhound and Karelian Bear Dog Variant)**
- **Mucopolysaccharidosis IIIB, Sanfilippo Syndrome Type B, MPS IIIB (NAGLU, Schipperke Variant)**
- **Early Onset Adult Deafness, EOAD (EPS8L2 Deletion, Rhodesian Ridgeback Variant)**

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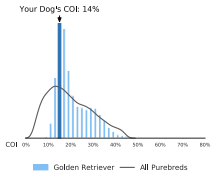
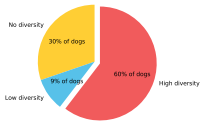
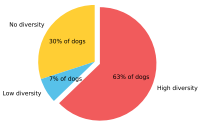
DNA Test Report | Test Date: April 30th, 2022 | embk.me/allie641

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.	**14%** 
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 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: