



## **BREED ANCESTRY**

Maltese : 100.0%

## **GENETIC STATS**

Predicted adult weight: **9 lbs**Life stage: **Young adult** 

Based on your dog's date of birth provided.

## **TEST DETAILS**

Kit number: EM-20340612 Swab number: 31211112010571









#### **Fun Fact**

The Maltese almost became extinct in the 17th and 18th centuries as attempts were made to breed them as small as squirrels.

## **MALTESE**

The Maltese is a playful toy dog, instantly recognizable by their long and white silky coat. This compact little pooch has a long history, with an origin tracing back at least two millennia. It is believed these guys first popped up in the Mediterranean. The Maltese was popular among British royalty by the end of the 1500s, along with a prominent history among French aristocrats, ancient Egyptians and the Roman Empire. The Maltese we recognize today was developed by English breeders and first made their way to the U.S in the late 1800s. They were first recognized by the AKC as an official breed in 1888. While Maltese dogs appear gentle, they are quite energetic and agile canines that compete in many performance events. That said, this breed is well suited to being a lapdog with a daily short burst of exercise sufficient to maintain a healthy dog. Don't let their perceived innocence fool you, the Maltese is a bold and confident dog that will often challenge larger breeds. They also make a good watch dog, due to their intelligence and tendency to bark at strangers and other dogs. Their eagerness to please makes them relatively easy to train, which is necessary to ensure Maltese dogs recognize boundaries and develop into a well rounded dog. The Maltese is a glamorous dog that can be high maintenance. Their impressive coat doesn't shed heavily but does requires regular brushing. This miniature breed ranks as the 31st most popular breed by the AKC.







## **MATERNAL LINE**



Through Effie'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: A1b**

This female lineage was very likely one of the original lineages in the wolves that were first domesticated into dogs in Central Asia about 15,000 years ago. Since then, the lineage has been very successful and travelled the globe! Dogs from this group are found in ancient Bronze Age fossils in the Middle East and southern Europe. By the end of the Bronze Age, it became exceedingly common in Europe. These dogs later became many of the dogs that started some of today's most popular breeds, like German Shepherds, Pugs, Whippets, English Sheepdogs and Miniature Schnauzers. During the period of European colonization, the lineage became even more widespread as European dogs followed their owners to farflung places like South America and Oceania. It's now found in many popular breeds as well as village dogs across the world!

#### HAPLOTYPE: A361/409/611

Part of the A1b haplogroup, this haplotype occurs most frequently in German Shepherd Dogs, Poodles, and Shiloh Shepherds.







## TRAITS: COAT COLOR

TRAIT

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

Not expressed (kyky)







# 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 white or cream (Dilute Red Pigmentation)

### A Locus (ASIP)

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

Not expressed (ayay)

### 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

#### 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".

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 at allele, so dogs that do not express at are not influenced by this gene.

Not expressed (NN)

## S Locus (MITF)

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

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





# TRAITS: COAT COLOR (CONTINUED)

TRAIT

#### 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) 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)







## TRAITS: OTHER COAT TRAITS

TRAIT

#### 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 furnished (mustache, beard, and/or eyebrows) (FF)

#### 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 light shedding (CC)

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

Very unlikely to be hairless (NN)







# TRAITS: OTHER COAT TRAITS (CONTINUED)

TRAIT

## 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 wavy coat (CC)







## TRAITS: OTHER BODY FEATURES

TRAIT

#### 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.

Unlikely to have hind dew claws (CC)







# TRAITS: OTHER BODY FEATURES (CONTINUED)

TRAIT

### 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)







# **TRAITS: BODY SIZE**

| TRAIT   | RESULT            |
|---|-------------------|
| Body Size (IGF1)  The I allele is associated with smaller body size.        | Smaller (II)      |
| 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.        | Intermediate (TA) |
| Body Size (GHR - E191K)  The A allele is associated with smaller body size. | Intermediate (GA) |
| Body Size (GHR - P177L)  The T allele is associated with smaller body size. | Larger (CC)       |





## 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) 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)







## **HEALTH REPORT**

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

If Effie 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 Effie 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 255 genetic health risks we analyzed, we found 1 result that you should learn about.

O Notable results (1)

**ALT Activity** 

Clear results

**Breed-relevant** (1)

**Other** (253)





# **BREED-RELEVANT RESULTS**

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



Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC, Maltese Variant)

Clear







# **OTHER RESULTS**

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

| ALT Activity (GPT)  | Notable |
|---|---------|
| 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   |
| 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   |
| Canine Fucosidosis (FUCA1)  | Clear   |
| Canine Leukocyte Adhesion Deficiency Type I, CLAD I (ITGB2, Setter Variant)   | Clear   |
| Canine Leukocyte Adhesion Deficiency Type III, CLAD III (FERMT3, German Shepherd Variant)                               | Clear   |
| Canine Multifocal Retinopathy, cmr1 (BEST1 Exon 2)  | Clear   |
| Canine Multifocal Retinopathy, cmr2 (BEST1 Exon 5, Coton de Tulear Variant)   | Clear   |
| Canine Multifocal Retinopathy, cmr3 (BEST1 Exon 10 Deletion, Finnish and Swedish Lapphund,<br>Lapponian Herder Variant) | Clear   |





| Canine Multiple System Degeneration (SERAC1 Exon 4, Chinese Crested Variant)         | Clear |
|--|-------|
| Canine 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 |
| Cobalamin Malabsorption (CUBN Exon 8, Beagle Variant)                                | Clear |
| Cobalamin Malabsorption (CUBN Exon 53, Border Collie Variant)                        | Clear |
| Collie Eye Anomaly (NHEJ1)   | Clear |
| Complement 3 Deficiency, C3 Deficiency (C3)  | Clear |
| Congenital Cornification Disorder (NSDHL, Chihuahua Variant)                         | Clear |
| Congenital Hypothyroidism (TPO, Rat, Toy, Hairless Terrier Variant)                  | Clear |
| Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)                         | Clear |
| Congenital Hypothyroidism with Goiter (TPO Intron 13, French Bulldog Variant)        | Clear |
| Congenital Hypothyroidism with Goiter (SLC5A5, Shih Tzu Variant)                     | Clear |
| Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant)   | Clear |





| Ongenital Myasthenic Syndrome, CMS (COLQ, Labrador Retriever Variant)       | Clear |
|---|-------|
| Ongenital Myasthenic Syndrome, CMS (COLQ, Golden Retriever Variant)         | Clear |
| Congenital Myasthenic Syndrome, CMS (CHAT, Old Danish Pointing Dog Variant) | Clear |
| Congenital Myasthenic Syndrome, CMS (CHRNE, Jack Russell Terrier Variant)   | Clear |
| Congenital Stationary Night Blindness (LRIT3, Beagle Variant)               | Clear |
| Ongenital Stationary Night Blindness (RPE65, Briard Variant)                | Clear |
|   | Clear |
| Craniomandibular Osteopathy, CMO (SLC37A2 Intron 16, Basset Hound Variant)  | Clear |
| Oystinuria 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 |
| Oay Blindness (CNGB3 Deletion, Alaskan Malamute Variant)                    | Clear |
| 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 |
| Obegenerative Myelopathy, DM (SOD1A)  | Clear |
| Demyelinating Polyneuropathy (SBF2/MTRM13)                                  | Clear |





| O Dental-Skeletal-Retinal Anomaly (MIA3, Cane Corso Variant)                                   | Clear |
|--|-------|
| Oiffuse Cystic Renal Dysplasia and Hepatic Fibrosis (INPP5E Intron 9, Norwich Terrier Variant) | Clear |
| Oilated Cardiomyopathy, DCM (RBM20, Schnauzer Variant)   | Clear |
| Oilated Cardiomyopathy, DCM1 (PDK4, Doberman Pinscher Variant 1)                               | Clear |
| Oilated Cardiomyopathy, DCM2 (TTN, Doberman Pinscher Variant 2)                                | Clear |
| Oisproportionate 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 |
| 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 |
| Glanzmann's Thrombasthenia Type I (ITGA2B Exon 13, Great Pyrenees Variant)  | 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 IIIA, GSD IIIA (AGL, Curly Coated Retriever Variant)  | Clear |
| Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM, Whippet<br>and English Springer Spaniel Variant) | Clear |
| Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM, Wachtelhund Variant)                             | Clear |
| GM1 Gangliosidosis (GLB1 Exon 2, Portuguese Water Dog Variant)  | Clear |
| GM1 Gangliosidosis (GLB1 Exon 15, Shiba Inu Variant)  | Clear |
|   | Clear |
| GM2 Gangliosidosis (HEXA, Japanese Chin Variant)  | Clear |
|   | Clear |
|   | 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 |
|  | Clear |
| Hemophilia B (F9 Exon 7, Terrier Variant)  | Clear |
| Hemophilia B (F9 Exon 7, Rhodesian Ridgeback 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 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 |
| ✓ Inflammatory Myopathy (SLC25A12)   | Clear |
| Inherited Myopathy of Great Danes (BIN1)   | Clear |
| Inherited Selected Cobalamin Malabsorption with Proteinuria (CUBN, Komondor Variant)       | Clear |
|  | 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 |
| Juvenile Epilepsy (LGI2)   | 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 |
| 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 |
| 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 |
| 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<br>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 |
| Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)  | Clear |
| Muscular Dystrophy (DMD, Golden 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 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 |
| 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 |
| Oculocutaneous Albinism, OCA (SLC45A2 Exon 6, Bullmastiff Variant)   | Clear |
| Oculocutaneous Albinism, OCA (SLC45A2, Small Breed Variant)  | Clear |
| Oculoskeletal Dysplasia 2 (COL9A2, Samoyed Variant)  | Clear |
| Osteochondrodysplasia (SLC13A1, Poodle Variant)  | Clear |
| Osteogenesis Imperfecta (COL1A2, Beagle Variant)   | Clear |
| Osteogenesis Imperfecta (SERPINH1, Dachshund Variant)  | Clear |





| Osteogenesis Imperfecta (COL1A1, Golden Retriever Variant)  | Clear |
|---|-------|
| P2Y12 Receptor Platelet Disorder (P2Y12)  | Clear |
| 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 |
| Platelet Factor X Receptor Deficiency, Scott Syndrome (TMEM16F)                                   | Clear |
| O Polycystic 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 (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 |
|--|-------|
| Progressive Retinal Atrophy (IFT122 Exon 26, Lapponian Herder 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 |





| 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 |





| 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 I, Type I ∨WD (VWF)   | Clear |
|   | Clear |
| ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)                   | Clear |
| On 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 |





# **OTHER RESULTS**

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

Clear







## **HEALTH REPORT**



Notable result

### **ALT Activity**

Effie Trinket inherited both copies of the variant we tested for Alanine Aminotransferase Activity

### Why is this important to your vet?

Effie has two copies of a variant in the GPT gene and is likely to have a lower than average baseline ALT activity. ALT is a commonly used measure of liver health on routine veterinary blood chemistry panels. As such, your veterinarian may want to watch for changes in Effie's ALT activity above their current, healthy, ALT activity. As an increase above Effie's baseline 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.





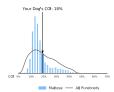
## INBREEDING AND DIVERSITY

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.



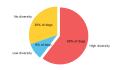


### MHC Class II - DLA DRB1

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**

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:

