### AMERICAN KENNEL CLUB NUMBER SOPHIE OF SVP TS54016807 SEX CAVALIER KING CHARLES SPANIEL

**BLENHEIM** SIRE SAWYER FORD TS40620801 09-20 (OFA24G AKC DNA #V935829) DAM RAINBOW OF SVP TS45563904 10-21

BREEDER LARRY HOCHSTETLER

OWNER

NAME

BREED

COLOR

WILLARD HELMUTH 579 N CR 100 E ARTHUR IL 61911-6265

FEMALE DATE OF BIRTH **JANUARY 6, 2022**  900215006395049



CERTIFICATE ISSUED SEPTEMBER 26, 2023 This certificate invalidates all previous certificates issued.

If a date appears after the name and number of the sire and dam, it indicates the issue of the Stud Book Register in which the sire or dam is published.

For Transfer Instructions, see back of Certificate.

This Certificate issued with the right to correct or revoke by the American Kennel Club.

**REGISTRATION CERTIFICATE** NEW CONFERENCE CONFERENCE CONFERENCE



**DNA Test Report** 

embk.me/sophie5166

### **BREED ANCESTRY**

Cavalier King Charles Spaniel : 100.0%

### **GENETIC STATS**

Predicted adult weight: 19 lbs

### **TEST DETAILS**

Kit number: EM-50165650 Swab number: 31220712002973





### Fun Fact

The breed experienced two large bursts in popularity. The first is when Queen Victoria revived the dying breed. The second was when Charlotte, a popular character from the popular show Sex and the City adopted one on TV. Test Date: November 22nd, 2023

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### **CAVALIER KING CHARLES SPANIEL**

The Cavalier King Charles Spaniel is one of the most popular dog breeds in the United States, and with good reason. Their affectionate personalities combined with their need to be close to their humans make them a lovely breed of choice for families. They tend to get along well with children and peaceably with other dogs and animals in the home (though as the breed used to be used for hunting, caution around small animals should be exercised). The Cavalier has an interesting history -- their ancestors were dogs of the British monarchy, but over time, the breed began to die out as dogs with shorter muzzles were favored in the 1800s. They were crossed with Pugs and some other breeds to change their appearance. However, Roswell Eldridge sought out King Charles Spaniels that had longer muzzles, and recreated the Cavalier as it used to be from those dogs.







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## MATERNAL LINE



Through Sophie'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: A1a

A1a is the most common maternal lineage among Western dogs. This lineage traveled from the site of dog domestication in Central Asia to Europe along with an early dog expansion perhaps 10,000 years ago. It hung around in European village dogs for many millennia. Then, about 300 years ago, some of the prized females in the line were chosen as the founding dogs for several dog breeds. That set in motion a huge expansion of this lineage. It's now the maternal lineage of the overwhelming majority of Mastiffs, Labrador Retrievers and Gordon Setters. About half of Boxers and less than half of Shar-Pei dogs descend from the A1a line. It is also common across the world among village dogs, a legacy of European colonialism.

### HAPLOTYPE: A224

Part of the large A1a haplogroup, this haplotype is found in village dogs in Peru, Fiji, and Namibia. Among breeds, we see this haplotype most frequently in Cavalier King Charles Spaniels, Mastiffs, and Boston Terriers.





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RESULT

### 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 (k<sup>y</sup>k<sup>y</sup>)





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RESULT

### TRAITS: COAT COLOR (CONTINUED)

#### TRAIT

#### 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 **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.

#### 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 (a<sup>t</sup>a<sup>t</sup>)

Not expressed (DD)



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RESULT

## 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.No co alleles, notDogs with the Nco genotype will produce black pigment, but can pass the co allele on to their puppies.expressed (NN)Dogs that have the coco genotype as well as the bb genotype at the B locus are generally a lighter brownthan dogs that have the Bb or BB genotypes at the B locus.

### 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 **a**<sup>t</sup> allele, so dogs that do not express **a**<sup>t</sup> are not influenced by this gene.

### 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 flash, parti, piebald, or extreme white (spsp)

Not expressed (NI)





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No merle alleles (mm)

### DNA Test Report

### 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. 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.

#### 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)



#### RESULT





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RESULT

### TRAITS: OTHER COAT TRAITS

### TRAIT

### Furnishings (RSPO2) LINKAGE

Dogs with one or two copies of the F allele have "furnishings": the mustache, beard, and eyebrowsLikely furnishedcharacteristic of breeds like the Schnauzer, Scottish Terrier, and Wire Haired Dachshund. A dog with two I(mustache, beard,alleles will not have furnishings, which is sometimes called an "improper coat" in breeds whereand/or eyebrows) (FI)furnishings are part of the breed standard. The mutation is a genetic insertion which we measureindirectly using a linkage test highly correlated with the insertion.

### 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, areLikely light sheddingheavy or seasonal shedders, while those with two copies of the T allele, including many Boxers, Shih Tzus(TT)and Chihuahuas, tend to be lighter shedders. Dogs with furnished/wire-haired coats caused by RSP02(the furnishings gene) tend to be low shedders regardless of their genotype at this gene.

#### 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)





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RESULT

## 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.

### 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.





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RESULT

### 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 **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.

### 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 normal-length tail (CC)

Likely to have hind dew claws (TT)







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RESULT

## 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" largebreed 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)





DNA Test Report	Test Date: November 22nd, 2023	embk.me/sophie5166
TRAITS: BODY SIZE		
TRAIT		RESULT
<b>Body Size (IGF1)</b> The I allele is associated with smalle	er body size.	Smaller (II)
<b>Body Size (IGFR1)</b> The <b>A</b> allele is associated with small	er body size.	Larger (GG)
Body Size (STC2) The <b>A</b> allele is associated with small	er body size.	Smaller (AA)
<b>Body Size (GHR - E191K)</b> The <b>A</b> allele is associated with smalle	er body size.	Smaller (AA)
<b>Body Size (GHR - P177L)</b> The <b>T</b> allele is associated with smalle	er body size.	Smaller (TT)

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RESULT

### TRAITS: PERFORMANCE

TRAIT

### 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.

### 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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**DNA Test Report** 

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### **HEALTH REPORT**

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

If Sophie 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 Sophie 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 256 genetic health risks we analyzed, we found 3 results that you should learn about.

Increased risk results (1)

Intervertebral Disc Disease (Type I)

Notable results (2)

Degenerative Myelopathy, DM

**Episodic Falling Syndrome** 

**Clear results** 

Breed-relevant (2)

**Other** (250)







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**BREED-RELEVANT RESULTS** 

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

Intervertebral Disc Disease (Type I) (FGF4 retrogene - CFA12)	Increased risk
Episodic Falling Syndrome (BCAN)	Notable
Ory Eye Curly Coat Syndrome (FAM83H Exon 5)	Clear
Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)	Clear





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### **OTHER RESULTS**

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

Degenerative Myelopathy, DM (SOD1A)	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
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
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



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## **OTHER RESULTS**

Canine Multifocal Retinopathy, cmr3 (BEST1 Exon 10 Deletion, Finnish and Swedish Lapphund, 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



Clear

**DNA Test Report** Test Date: November 22nd, 2023 embk.me/sophie5166 **OTHER RESULTS** Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant) Clear Congenital Myasthenic Syndrome, CMS (COLQ, Labrador Retriever Variant) Clear  $\oslash$ Congenital Myasthenic Syndrome, CMS (COLQ, Golden Retriever Variant) Clear  $(\checkmark)$ Congenital Myasthenic Syndrome, CMS (CHAT, Old Danish Pointing Dog Variant) Clear  $\bigcirc$ Congenital Myasthenic Syndrome, CMS (CHRNE, Jack Russell Terrier Variant) Clear  $(\checkmark)$ Congenital Stationary Night Blindness (LRIT3, Beagle Variant) Clear  $\oslash$ Congenital Stationary Night Blindness (RPE65, Briard Variant)  $\langle \rangle$ Clear Craniomandibular Osteopathy, CMO (SLC37A2) Clear  $\langle \rangle$ Craniomandibular Osteopathy, CMO (SLC37A2 Intron 16, Basset Hound Variant) Clear  $\langle \rangle$  $\oslash$ Cystinuria Type I-A (SLC3A1, Newfoundland Variant) Clear Cystinuria Type II-A (SLC3A1, Australian Cattle Dog Variant)  $\oslash$ Clear  $\oslash$ Cystinuria Type II-B (SLC7A9, Miniature Pinscher Variant) Clear Day Blindness (CNGB3 Deletion, Alaskan Malamute Variant) Clear  $\oslash$  $\oslash$ Day Blindness (CNGA3 Exon 7, German Shepherd Variant) Clear Day Blindness (CNGA3 Exon 7, Labrador Retriever Variant) Clear  $\oslash$ Day Blindness (CNGB3 Exon 6, German Shorthaired Pointer Variant) Clear  $(\checkmark)$ Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MYO7A) Clear  $\oslash$ 

Rembark

Demyelinating Polyneuropathy (SBF2/MTRM13)

 $\oslash$ 

DNA



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## **OTHER RESULTS**

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
O Dilated Cardiomyopathy, DCM1 (PDK4, Doberman Pinscher Variant 1)	Clear
Dilated Cardiomyopathy, DCM2 (TTN, Doberman Pinscher Variant 2)	Clear
Disproportionate Dwarfism (PRKG2, Dogo Argentino Variant)	Clear
O Dystrophic Epidermolysis Bullosa (COL7A1, Central Asian Shepherd Dog Variant)	Clear
O Dystrophic 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
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



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## **OTHER RESULTS**

Complied Nephropothy (COL 444 Even 20 English Springer Speniol Verient)	Clear
Familial Nephropathy (COL4A4 Exon 30, English Springer Spaniel Variant)	Clear
🧭 Fanconi Syndrome (FAN1, Basenji Variant)	Clear
Setal-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 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
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
GM1 Gangliosidosis (GLB1 Exon 15, Alaskan Husky Variant)	Clear
<ul> <li>GM1 Gangliosidosis (GLB1 Exon 15, Alaskan Husky Variant)</li> <li>GM2 Gangliosidosis (HEXA, Japanese Chin Variant)</li> </ul>	Clear Clear
GM2 Gangliosidosis (HEXA, Japanese Chin Variant)	Clear
<ul> <li>GM2 Gangliosidosis (HEXA, Japanese Chin Variant)</li> <li>GM2 Gangliosidosis (HEXB, Poodle Variant)</li> </ul>	Clear Clear



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### **OTHER RESULTS**

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, 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
Ichthyosis (NIPAL4, American Bulldog Variant)	Clear
Ichthyosis (ASPRV1 Exon 2, German Shepherd Variant)	Clear
Ichthyosis (SLC27A4, Great Dane Variant)	Clear



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Clear

Clear

Clear

DNA	Test Report	Test Date: November 22nd, 2023
ОТ	HER RESULTS	
$\oslash$	Ichthyosis, Epidermolytic Hyperkeratosis (K	RT10, Terrier Variant)
$\oslash$	Ichthyosis, ICH1 (PNPLA1, Golden Retriever	Variant)
$\oslash$	Inflammatory Myopathy (SLC25A12)	

Inherited Myopathy of Great Danes (BIN1)	Clear
Inherited Selected Cobalamin Malabsorption with Proteinuria (CUBN, Komondor Variant)	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
C L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH, Staffordshire Bull Terrier Variant)	Clear
C Lagotto Storage Disease (ATG4D)	Clear
Laryngeal Paralysis (RAPGEF6, Miniature Bull Terrier Variant)	Clear
Conset Spinocerebellar Ataxia (CAPN1)	Clear
S Late-Onset Neuronal Ceroid Lipofuscinosis, NCL 12 (ATP13A2, Australian Cattle Dog Variant)	Clear
Leonberger Polyneuropathy 1 (LPN1, ARHGEF10)	Clear
Control Leonberger Polyneuropathy 2 (GJA9)	Clear



**DNA Test Report** 

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## **OTHER RESULTS**

Lethal Acrodermatitis, LAD (MKLN1)	Clear
Leukodystrophy (TSEN54 Exon 5, Standard Schnauzer Variant)	Clear
Contractional Ligneous Membranitis, LM (PLG)	Clear
Control Limb Girdle Muscular Dystrophy (SGCD, Boston Terrier Variant)	Clear
C Limb-Girdle Muscular Dystrophy 2D (SGCA Exon 3, Miniature Dachshund Variant)	Clear
Comp QT Syndrome (KCNQ1)	Clear
Undehund 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 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



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## **OTHER RESULTS**

Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5, Terrier Brasileiro Variant)	Clear
Multiple Drug Sensitivity (ABCB1)	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



**DNA Test Report** Test Date: November 22nd, 2023 embk.me/sophie5166 **OTHER RESULTS** Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4, Dachshund Variant 2) Clear  $(\checkmark)$ Neuronal Ceroid Lipofuscinosis 5, NCL 5 (CLN5 Exon 4 SNP, Border Collie Variant) Clear  $\oslash$ Neuronal Ceroid Lipofuscinosis 5, NCL 5 (CLN5 Exon 4 Deletion, Golden Retriever Variant) Clear  $(\checkmark)$ Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7, Australian Shepherd Variant) Clear  $\bigcirc$ Neuronal Ceroid Lipofuscinosis 7, NCL7 (MFSD8, Chihuahua and Chinese Crested Variant) Clear (~) Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8, Australian Shepherd Variant) Clear  $(\label{eq:started})$ Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 Exon 2, English Setter Variant)  $(\label{eq:started})$ Clear Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 Insertion, Saluki Variant) Clear  $( \land )$ Neuronal Ceroid Lipofuscinosis, Cerebellar Ataxia, NCL4A (ARSG Exon 2, American Staffordshire Terrier Clear  $\langle \rangle$ Variant) Oculocutaneous Albinism, OCA (SLC45A2 Exon 6, Bullmastiff Variant) Clear  $\oslash$ Oculocutaneous Albinism, OCA (SLC45A2, Small Breed Variant) Clear  $( \label{eq:started} )$ Oculoskeletal Dysplasia 2 (COL9A2, Samoyed Variant) Clear  $\oslash$ Osteochondrodysplasia (SLC13A1, Poodle Variant)  $(\checkmark)$ Clear Osteogenesis Imperfecta (COL1A2, Beagle Variant) Clear  $(\checkmark)$ Osteogenesis Imperfecta (SERPINH1, Dachshund Variant) Clear  $\langle \rangle$ 

Osteogenesis Imperfecta (OL1A1, Golden Retriever Variant)
 Osteogenesis Imperfecta (COL1A1, Golden Retriever Variant)
 P2Y12 Receptor Platelet Disorder (P2Y12)
 Clear
 Pachyonychia Congenita (KRT16, Dogue de Bordeaux Variant)
 Clear



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## **OTHER RESULTS**

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
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
<ul> <li>Primary Open Angle Glaucoma and Primary Lens Luxation (ADAMTS17 Exon 2, Chinese Shar-Pei Variant)</li> </ul>	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



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## **OTHER RESULTS**

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



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OTHER RESULTS		
Renal Cystadenocarcinoma and Nodula	r Dermatofibrosis (FLCN Exon 7)	Clear
Retina Dysplasia and/or Optic Nerve Hy	poplasia (SIX6 Exon 1, Golden Retriever Variant)	Clear
Sensory Neuropathy (FAM134B, Border	Collie Variant)	Clear
Severe Combined Immunodeficiency, S	CID (PRKDC, Terrier Variant)	Clear
Severe Combined Immunodeficiency, S	CID (RAG1, Wetterhoun Variant)	Clear
Shaking Puppy Syndrome (PLP1, Englis)	h Springer Spaniel Variant)	Clear
Shar-Pei Autoinflammatory Disease, SP	AID, Shar-Pei Fever (MTBP)	Clear
Skeletal Dysplasia 2, SD2 (COL11A2, Lab	orador Retriever Variant)	Clear
Skin Fragility Syndrome (PKP1, Chesape	eake Bay Retriever Variant)	Clear
Spinocerebellar Ataxia (SCN8A, Alpine I	Dachsbracke Variant)	Clear
Spinocerebellar Ataxia with Myokymia a	and/or Seizures (KCNJ10)	Clear
Spongy Degeneration with Cerebellar A	taxia 1 (KCNJ10)	Clear
Spongy Degeneration with Cerebellar A	taxia 2 (ATP1B2)	Clear
Stargardt Disease (ABCA4 Exon 28, Lab	rador Retriever Variant)	Clear
Succinic Semialdehyde Dehydrogenase	e Deficiency (ALDH5A1 Exon 7, Saluki Variant)	Clear
O Thrombopathia (RASGRP1 Exon 5, Amer	ican Eskimo Dog Variant)	Clear
O Thrombopathia (RASGRP1 Exon 5, Bass	et Hound Variant)	Clear
O Thrombopathia (RASGRP1 Exon 8, Lands	seer Variant)	Clear



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## **OTHER RESULTS**

Trapped Neutrophil Syndrome, TNS (VPS13B)	Clear
O Ullrich-like Congenital Muscular Dystrophy (COL6A3 Exon 10, Labrador Retriever Variant)	Clear
Ullrich-like Congenital Muscular Dystrophy (COL6A1 Exon 3, Landseer Variant)	Clear
O Unilateral Deafness and Vestibular Syndrome (PTPRQ Exon 39, Doberman Pinscher)	Clear
Urate Kidney & Bladder Stones (SLC2A9)	Clear
Von Willebrand Disease Type I, Type I vWD (VWF)	Clear
Von Willebrand Disease Type II, Type II 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
Xanthine Urolithiasis (XDH, Mixed Breed Variant)	Clear
β-Mannosidosis (MANBA Exon 16, Mixed-Breed Variant)	Clear
Mast Cell Tumor	No result

**DNA Test Report** 

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## **HEALTH REPORT**

Increased risk result

### Intervertebral Disc Disease (Type I)

Sophie inherited both copies of the variant we tested for Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD Sophie is at increased risk for Type I IVDD

### How to interpret this result

Sophie has two copies of an FGF4 retrogene on chromosome 12. In some breeds such as Beagles, Cocker Spaniels, and Dachshunds (among others) this variant is found in nearly all dogs. While those breeds are known to have an elevated risk of IVDD, many dogs in those breeds never develop IVDD. For mixed breed dogs and purebreds of other breeds where this variant is not as common, risk for Type I IVDD is greater for individuals with this variant than for similar dogs.

### What is Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD?

Type I Intervertebral Disc Disease (IVDD) is a back/spine issue that refers to a health condition affecting the discs that act as cushions between vertebrae. With Type I IVDD, affected dogs can have a disc event where it ruptures or herniates towards the spinal cord. This pressure on the spinal cord causes neurologic signs which can range from a wobbly gait to impairment of movement. Chondrodystrophy (CDDY) refers to the relative proportion between a dog's legs and body, wherein the legs are shorter and the body longer. There are multiple different variants that can cause a markedly chondrodystrophic appearance as observed in Dachshunds and Corgis. However, this particular variant is the only one known to also increase the risk for IVDD.

### When signs & symptoms develop in affected dogs

Signs of CDDY are recognized in puppies as it affects body shape. IVDD is usually first recognized in adult dogs, with breed specific differences in age of onset.

### Signs & symptoms

Research indicates that dogs with one or two copies of this variant have a similar risk of developing IVDD. However, there are some breeds (e.g. Beagles and Cocker Spaniels, among others) where this variant has been passed down to nearly all dogs of the breed and most do not show overt clinical signs of the disorder. This suggests that there are other genetic and environmental factors (such as weight, mobility, and family history) that contribute to an individual dog's risk of developing clinical IVDD. Signs of IVDD include neck or back pain, a change in your dog's walking pattern (including dragging of the hind limbs), and paralysis. These signs can be mild to severe, and if your dog starts exhibiting these signs, you should schedule an appointment with your veterinarian for a diagnosis.

### How vets diagnose this condition

For CDDY, dogs with one copy of this variant may have mild proportional differences in their leg length. Dogs with two copies of this variant will often have visually longer bodies and shorter legs. For IVDD, a neurological exam will be performed on any dog showing suspicious signs. Based on the result of this exam, radiographs to detect the presence of calcified discs or advanced imaging (MRI/CT) to detect a disc rupture may be recommended.

### How this condition is treated

IVDD is treated differently based on the severity of the disease. Mild cases often respond to medical management which includes

Fembark

**DNA Test Report** 

### **HEALTH REPORT**

Notable result

### Degenerative Myelopathy, DM

Sophie inherited one copy of the variant we tested for Degenerative Myelopathy, DM

### What does this result mean?

This variant should not impact Sophie's health. This variant is inherited in an autosomal recessive manner, meaning that a dog needs two copies of the variant to show signs of this condition. Sophie is unlikely to develop this condition due to this variant because she only has one copy of the variant.

### Impact on Breeding

Your dog carries this variant and will pass it on to ~50% of her offspring. You can email breeders@embarkvet.com to discuss with a genetic counselor how the genotype results should be applied to a breeding program.

### What is Degenerative Myelopathy, DM?

The dog equivalent of Amyotrophic Lateral Sclerosis, or Lou Gehrig's disease, DM is a progressive degenerative disorder of the spinal cord. Because the nerves that control the hind limbs are the first to degenerate, the most common clinical signs are back muscle wasting and gait abnormalities.

### When signs & symptoms develop in affected dogs

Affected dogs do not usually show signs of DM until they are at least 8 years old.

### How vets diagnose this condition

Definitive diagnosis requires microscopic analysis of the spinal cord after death. However, veterinarians use clues such as genetic testing, breed, age, and other diagnostics to determine if DM is the most likely cause of your dog's clinical signs.

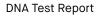
### How this condition is treated

As dogs are seniors at the time of onset, the treatment for DM is aimed towards increasing their comfort through a combination of lifestyle changes, medication, and physical therapy.

### Actions to take if your dog is affected

• Giving your dog the best quality of life for as long as possible is all you can do after receiving this diagnosis.







embk.me/sophie5166

## **HEALTH REPORT**

Notable result

### **Episodic Falling Syndrome**

Sophie inherited one copy of the variant we tested for Episodic Falling Syndrome

### What does this result mean?

This variant should not impact Sophie's health. This variant is inherited in an autosomal recessive manner, meaning that a dog needs two copies of the variant to show signs of this condition. Sophie is unlikely to develop this condition due to this variant because she only has one copy of the variant.

### Impact on Breeding

Your dog carries this variant and will pass it on to ~50% of her offspring. You can email breeders@embarkvet.com to discuss with a genetic counselor how the genotype results should be applied to a breeding program.

### What is Episodic Falling Syndrome?

This disease causes episodes of spastic muscle contraction in response to stress, excitement, or exercise. EFS is caused by deficiency of a protein called brevican, which has a role in controlling the speed and rate at which specific neurons in the brain and spinal cord fire. Loss of brevican leads to abnormal bursts of neuronal activity, leading to the downstream effect of spastic muscle contraction.

### When signs & symptoms develop in affected dogs

Signs first appear in puppies.

### How vets diagnose this condition

Genetic testing, blood work, neurological tests, and clinical signs can be used to diagnose this condition.

### How this condition is treated

Affected dogs usually recover within an hour of an episode, though the stiff limbs and gait may persist for several hours. However, they may overheat during an episode due to the uncontrollable muscle contractions, which could be life threatening. Medications are available to help control symptoms.

### Actions to take if your dog is affected

• Minimizing exposure to typical triggers may help reduce clinical signs. Please follow the recommendations from your veterinarian.







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RESULT

### INBREEDING AND DIVERSITY

CATEGORY

### **Coefficient Of Inbreeding**

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

### 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.

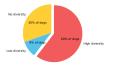
### MHC Class II - DLA DQA1 and DQB1

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.

Your Dog's COI 29%

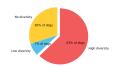
#### **High Diversity**

How common is this amount of diversity in purebreds:



### **High Diversity**

How common is this amount of diversity in purebreds:





### 29%