

# "BOSTON"

## BOSTON



DNA Test Report

Test Date: March 2nd, 2022

embk.me/boston160

### BREED ANCESTRY

 Dachshund : 100.0%

### GENETIC STATS

Predicted adult weight: **15 lbs**

Life stage: **Young adult**

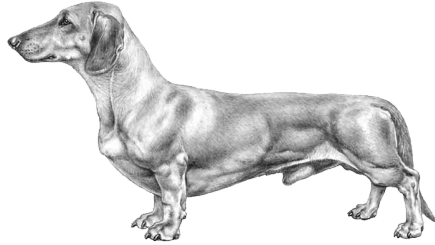
Based on your dog's date of birth provided.

### TEST DETAILS

Kit number: EM-80417154

Swab number: 31210751909681

### DACHSHUND



The Dachshund was bred originally in Germany to flush out Badgers and other den animals in the 15th century. The breed, originally known as the Teckel, was refined by German Foresters to have the elongated shape that is advantageous for fitting into tight animal burrows. Dachshunds are often viewed as a symbol for Germany. For example, a Dachshund named Waldi was the first official mascot of the 1972 Summer Olympics held in Munich. Dachshunds are one of the most popular breeds in the United States, ranking 13th in AKC's most popular breeds. The Dachshund's personality is described as energetic, clever, and persistent to the point of stubbornness.

#### Alternative Names

Dachshund (Miniature), Dachshund (Standard)

#### Fun Fact

The name Dachshund is derived from "Dachs Krieger" meaning "Badger Warrior", who knew your Dachshund has such a fearsome name!

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## MATERNAL LINE



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

### HAPLOGROUP: A1d

This female lineage can be traced back about 15,000 years to some of the original Central Asian wolves that were domesticated into modern dogs. The early females that represent this lineage were likely taken into Eurasia, where they spread rapidly. As a result, many modern breed and village dogs from the Americas, Africa, through Asia and down into Oceania belong to this group! This widespread lineage is not limited to a select few breeds, but the majority of Rottweilers, Afghan Hounds and Wirehaired Pointing Griffons belong to it. It is also the most common female lineage among Papillons, Samoyeds and Jack Russell Terriers. Considering its occurrence in breeds as diverse as Afghan Hounds and Samoyeds, some of this is likely ancient variation. But because of its presence in many modern European breeds, much of its diversity likely can be attributed to much more recent breeding.

### HAPLOTYPE: A247

Part of the large A1d haplogroup, this common haplotype occurs in village dogs all over the world. Among the 32 breeds we have sampled it in, the most common occurrences include Boxers, Labrador Retrievers, and Papillons.

Registration: American Kennel Club

(AKC) HP61363101



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### PATERNAL LINE



Through Boston's Y chromosome we can trace his father's ancestry back to where dogs and people first became friends. This map helps you visualize the routes that his ancestors took to your home. Their story is described below the map.

#### HAPLOGROUP: A2b

A2b appears to have split a few times in succession, which means that some of the Central Asian male ancestors of this lineage went their separate ways before their respective Y chromosomes made their rounds. There is not much diversity in this lineage, meaning that it has only begun to take off recently. Two iconic breeds, the Dachshund and Bloodhound, represent this lineage well. Over half of Rottweilers are A2b, as are the majority of Labrador Retrievers and Cavalier King Charles Spaniels. While A2a is restricted mostly to East Asia, this paternal line is also found among European breeds.

#### HAPLOTYPE: Hc.9

Part of the A2b haplogroup, this haplotype is found in village dogs spanning South America, Africa, and the South Pacific. Among the breeds we have spotted it in, the most frequent occurrences are in Dachshund, Bloodhound, American Eskimo Dog, and Jack Russell Terrier.

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### TRAITS: COAT COLOR

TRAIT	RESULT
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#### E Locus (MC1R)

The E Locus determines if and where a dog can produce dark (black or brown) hair. Dogs with two copies of the recessive **e** allele do not produce dark hairs at all, and will be "red" over their entire body. The shade of red, which can range from a deep copper to yellow/gold to cream, is dependent on other genetic factors including the Intensity loci. In addition to determining if a dog can develop dark hairs at all, the E Locus can give a dog a black "mask" or "widow's peak," unless the dog has overriding coat color genetic factors. Dogs with one or two copies of the **Em** allele usually have a melanistic mask (dark facial hair as commonly seen in the German Shepherd and Pug). Dogs with no copies of **Em** but one or two copies of the **Eg** allele usually have a melanistic "widow's peak" (dark forehead hair as commonly seen in the Afghan Hound and Borzoi, where it is called either "grizzle" or "domino").

No dark mask or grizzle (Ee)

#### K Locus (CBD103)

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

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

### TRAITS: COAT COLOR (CONTINUED)

TRAIT	RESULT
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#### Intensity Loci LINKAGE

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

**Any light hair likely yellow or tan (Intermediate Red Pigmentation)**

#### A Locus (ASIP)

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

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

#### D Locus (MLPH)

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

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

### TRAITS: COAT COLOR (CONTINUED)

TRAIT	RESULT
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#### Cocoa (HPS3)

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

**No co alleles, not expressed (NN)**

#### B Locus (TYRP1)

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

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

#### Saddle Tan (RALY)

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

**Likely saddle tan patterned (ll)**

#### S Locus (MITF)

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

**Likely to have little to no white in coat (SS)**

### TRAITS: COAT COLOR (CONTINUED)

TRAIT	RESULT
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#### 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	RESULT
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#### Furnishings (RSPO2) LINKAGE

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

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

#### Coat Length (FGF5)

The FGF5 gene is known to affect hair length in many different species, including cats, dogs, mice, and humans. In dogs, the **T** allele confers a long, silky haircoat as observed in the Yorkshire Terrier and the Long Haired Whippet. The ancestral **G** allele causes a shorter coat as seen in the Boxer or the American Staffordshire Terrier. In certain breeds (such as Corgi), the long haircoat is described as "fluff."

**Likely long coat (TT)**

#### Shedding (MC5R)

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

**Likely light shedding (TT)**

#### 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

#### RESULT

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

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

Likely not albino (NN)

##### Coat Texture (KRT71)

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

Likely straight coat (CC)

### TRAITS: OTHER BODY FEATURES

TRAIT	RESULT
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#### Muzzle Length (BMP3)

Dogs in medium-length muzzle (mesocephalic) breeds like Staffordshire Terriers and Labradors, and long muzzle (dolichocephalic) breeds like Whippet and Collie have one, or more commonly two, copies of the ancestral **C** allele. Dogs in many short-length muzzle (brachycephalic) breeds such as the English Bulldog, Pug, and Pekingese have two copies of the derived **A** allele. At least five different genes affect muzzle length in dogs, with BMP3 being the only one with a known causal mutation. For example, the skull shape of some breeds, including the dolichocephalic Scottish Terrier or the brachycephalic Japanese Chin, appear to be caused by other genes. Thus, dogs may have short or long muzzles due to other genetic factors that are not yet known to science.

**Likely medium or long muzzle (CC)**

#### Tail Length (T)

Whereas most dogs have two **C** alleles and a long tail, dogs with one **G** allele are likely to have a bobtail, which is an unusually short or absent tail. This mutation causes natural bobtail in many breeds including the Pembroke Welsh Corgi, the Australian Shepherd, and the Brittany Spaniel. Dogs with **GG** genotypes have not been observed, suggesting that dogs with the **GG** genotype do not survive to birth. Please note that this mutation does not explain every natural bobtail! While certain lineages of Boston Terrier, English Bulldog, Rottweiler, Miniature Schnauzer, Cavalier King Charles Spaniel, and Parson Russell Terrier, and Dobermans are born with a natural bobtail, these breeds do not have this mutation. This suggests that other unknown genetic mutations can also lead to a natural bobtail.

**Likely normal-length tail (CC)**

#### Hind Dewclaws (LMBR1)

Common in certain breeds such as the Saint Bernard, hind dewclaws are extra, nonfunctional digits located midway between a dog's paw and hock. Dogs with at least one copy of the **T** allele have about a 50% chance of having hind dewclaws. Note that other (currently unknown to science) mutations can also cause hind dewclaws, so some **CC** or **TC** dogs will have hind dewclaws.

**Unlikely to have hind dew claws (CC)**

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### TRAITS: OTHER BODY FEATURES (CONTINUED)

#### TRAIT

#### RESULT

##### Blue Eye Color (ALX4) LINKAGE

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

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

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

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

**Likely normal muscling (CC)**

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### TRAITS: BODY SIZE

TRAIT	RESULT
<b>Body Size (IGF1)</b> The I allele is associated with smaller body size.	<b>Intermediate (NI)</b>
<b>Body Size (IGFR1)</b> The A allele is associated with smaller body size.	<b>Smaller (AA)</b>
<b>Body Size (STC2)</b> The A allele is associated with smaller body size.	<b>Intermediate (TA)</b>
<b>Body Size (GHR - E191K)</b> The A allele is associated with smaller body size.	<b>Intermediate (GA)</b>
<b>Body Size (GHR - P177L)</b> The T allele is associated with smaller body size.	<b>Larger (CC)</b>

Registration:



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### TRAITS: PERFORMANCE

#### TRAIT

#### RESULT

##### Altitude Adaptation (EPAS1)

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

**Normal altitude tolerance (GG)**

##### Appetite (POMC) LINKAGE

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

**Normal food motivation (NN)**

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## HEALTH REPORT

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

If Boston 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 Boston 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 242 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)

**ALT Activity**

**Progressive Retinal Atrophy, crd4/crd1**

#### Clear results

**Breed-relevant (6)**

**Other (233)**

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### BREED-RELEVANT RESULTS

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

 Intervertebral Disc Disease (Type I) (FGF4 retrogene - CFA12)	Increased risk
 Progressive Retinal Atrophy, crd4/cord1 (RPGRIP1)	Notable
 Limb-Girdle Muscular Dystrophy 2D (SGCA Exon 3, Miniature Dachshund Variant)	Clear
 Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6, Dachshund Variant)	Clear
 Narcolepsy (HCRTR2 Exon 1, Dachshund Variant)	Clear
 Neuronal Ceroid Lipofuscinosis 1, NCL 1 (PPT1 Exon 8, Dachshund Variant 1)	Clear
 Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4, Dachshund Variant 2)	Clear
 Osteogenesis Imperfecta (SERPINH1, Dachshund Variant)	Clear

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### OTHER RESULTS

Research has not yet linked these conditions to dogs with similar breeds to Boston. Review any increased risk or notable results to understand his 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, Lapponian Herder Variant)	Clear

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### OTHER RESULTS

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 Hypothyroidism (TPO, Rat, Toy, Hairless Terrier Variant)	Clear
Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)	Clear
Congenital Hypothyroidism with Goiter (SLC5A5, Shih Tzu Variant)	Clear
Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant)	Clear
Congenital Myasthenic Syndrome, CMS (COLQ, Labrador Retriever Variant)	Clear
Congenital Myasthenic Syndrome, CMS (COLQ, Golden Retriever Variant)	Clear

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### OTHER RESULTS

 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
 Congenital Stationary Night Blindness (RPE65, Briard Variant)	Clear
 Craniomandibular Osteopathy, CMO (SLC37A2)	Clear
 Cystinuria Type I-A (SLC3A1, Newfoundland Variant)	Clear
 Cystinuria Type II-A (SLC3A1, Australian Cattle Dog Variant)	Clear
 Cystinuria Type II-B (SLC7A9, Miniature Pinscher Variant)	Clear
 Day Blindness (CNGB3 Deletion, Alaskan Malamute Variant)	Clear
 Day Blindness (CNGA3 Exon 7, German Shepherd Variant)	Clear
 Day Blindness (CNGA3 Exon 7, Labrador Retriever Variant)	Clear
 Day Blindness (CNGB3 Exon 6, German Shorthaired Pointer Variant)	Clear
 Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MYO7A)	Clear
 Degenerative Myelopathy, DM (SOD1A)	Clear
 Demyelinating Polyneuropathy (SBF2/MTRM13)	Clear
 Diffuse Cystic Renal Dysplasia and Hepatic Fibrosis (INPP5E Intron 9, Norwich Terrier Variant)	Clear
 Dilated Cardiomyopathy, DCM (RBM20, Schnauzer Variant)	Clear
 Dilated Cardiomyopathy, DCM1 (PDK4, Doberman Pinscher Variant 1)	Clear

Registration: American Kennel Club (AKC)

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# "BOSTON"

## BOSTON



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### OTHER RESULTS

- ✔ Dilated Cardiomyopathy, DCM2 (TTN, Doberman Pinscher Variant 2) Clear
- ✔ Dry Eye Curly Coat Syndrome (FAM83H Exon 5) Clear
- ✔ Dystrophic Epidermolysis Bullosa (COL7A1, Central Asian Shepherd Dog Variant) Clear
- ✔ 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
- ✔ 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

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### OTHER RESULTS

 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
 GM2 Gangliosidosis (HEXA, Japanese Chin Variant)	Clear
 GM2 Gangliosidosis (HEXB, Poodle Variant)	Clear
 Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)	Clear
 Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)	Clear
 Goniodysgenesis and Glaucoma, Pectinate Ligament Dysplasia, PLD (OLFM3)	Clear
 Hemophilia A (F8 Exon 11, German Shepherd Variant 1)	Clear
 Hemophilia A (F8 Exon 1, German Shepherd Variant 2)	Clear
 Hemophilia A (F8 Exon 10, Boxer Variant)	Clear

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### OTHER RESULTS

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 (SLC27A4, Great Dane Variant)	Clear
Ichthyosis, Epidermolytic Hyperkeratosis (KRT10, Terrier Variant)	Clear
Ichthyosis, ICH1 (PNPLA1, Golden Retriever Variant)	Clear
Inflammatory Myopathy (SLC25A12)	Clear
Inherited Myopathy of Great Danes (BIN1)	Clear

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# "BOSTON"

## BOSTON



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### OTHER RESULTS

- |                                                                                                                                |       |
|--------------------------------------------------------------------------------------------------------------------------------|-------|
| <input checked="" type="checkbox"/> Inherited Selected Cobalamin Malabsorption with Proteinuria (CUBN, Komondor Variant)       | Clear |
| <input checked="" type="checkbox"/> Junctional Epidermolysis Bullosa (LAMA3 Exon 66, Australian Cattle Dog Variant)            | Clear |
| <input checked="" type="checkbox"/> Junctional Epidermolysis Bullosa (LAMB3 Exon 11, Australian Shepherd Variant)              | Clear |
| <input checked="" type="checkbox"/> Juvenile Epilepsy (LGI2)                                                                   | Clear |
| <input checked="" type="checkbox"/> Juvenile Laryngeal Paralysis and Polyneuropathy (RAB3GAP1, Rottweiler Variant)             | Clear |
| <input checked="" type="checkbox"/> Juvenile Myoclonic Epilepsy (DIRAS1)                                                       | Clear |
| <input checked="" type="checkbox"/> L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH, Staffordshire Bull Terrier Variant)            | Clear |
| <input checked="" type="checkbox"/> Lagotto Storage Disease (ATG4D)                                                            | Clear |
| <input checked="" type="checkbox"/> Laryngeal Paralysis (RAPGEF6, Miniature Bull Terrier Variant)                              | Clear |
| <input checked="" type="checkbox"/> Late Onset Spinocerebellar Ataxia (CAPN1)                                                  | Clear |
| <input checked="" type="checkbox"/> Late-Onset Neuronal Ceroid Lipofuscinosis, NCL 12 (ATP13A2, Australian Cattle Dog Variant) | Clear |
| <input checked="" type="checkbox"/> Leonberger Polyneuropathy 1 (LPN1, ARHGEF10)                                               | Clear |
| <input checked="" type="checkbox"/> Leonberger Polyneuropathy 2 (GJA9)                                                         | Clear |
| <input checked="" type="checkbox"/> Lethal Acrodermatitis, LAD (MKLN1)                                                         | Clear |
| <input checked="" type="checkbox"/> Ligneous Membranitis, LM (PLG)                                                             | Clear |
| <input checked="" type="checkbox"/> Limb Girdle Muscular Dystrophy (SGCD, Boston Terrier Variant)                              | Clear |
| <input checked="" type="checkbox"/> Long QT Syndrome (KCNQ1)                                                                   | Clear |
| <input checked="" type="checkbox"/> Lundehund Syndrome (LEPREL1)                                                               | Clear |

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### OTHER RESULTS

Macular Corneal Dystrophy, MCD (CHST6)	Clear
Malignant Hyperthermia (RYR1)	Clear
May-Hegglin Anomaly (MYH9)	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, New Zealand Huntaway Variant)	Clear
Mucopolysaccharidosis Type VI, Maroteaux-Lamy Syndrome, MPS VI (ARSB Exon 5, Miniature Pinscher Variant)	Clear
Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3, German Shepherd Variant)	Clear
Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5, Terrier Brasileiro Variant)	Clear
Multiple Drug Sensitivity (ABCB1)	Clear
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 Intron 4, Doberman Pinscher Variant)	Clear

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### OTHER RESULTS

✔ 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 10, NCL 10 (CTSD Exon 5, American Bulldog Variant)	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, Small Breed Variant)	Clear
✔ Oculoskeletal Dysplasia 2 (COL9A2, Samoyed Variant)	Clear

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## BOSTON



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### OTHER RESULTS

- |                                                                                                                      |       |
|----------------------------------------------------------------------------------------------------------------------|-------|
| <input checked="" type="checkbox"/> Osteochondrodysplasia (SLC13A1, Poodle Variant)                                  | Clear |
| <input checked="" type="checkbox"/> Osteogenesis Imperfecta (COL1A2, Beagle Variant)                                 | Clear |
| <input checked="" type="checkbox"/> Osteogenesis Imperfecta (COL1A1, Golden Retriever Variant)                       | Clear |
| <input checked="" type="checkbox"/> P2Y12 Receptor Platelet Disorder (P2Y12)                                         | Clear |
| <input checked="" type="checkbox"/> Pachyonychia Congenita (KRT16, Dogue de Bordeaux Variant)                        | Clear |
| <input checked="" type="checkbox"/> Paroxysmal Dyskinesia, PxD (PIGN)                                                | Clear |
| <input checked="" type="checkbox"/> Persistent Mullerian Duct Syndrome, PMDS (AMHR2)                                 | Clear |
| <input checked="" type="checkbox"/> Pituitary Dwarfism (POU1F1 Intron 4, Karelian Bear Dog Variant)                  | Clear |
| <input checked="" type="checkbox"/> Platelet Factor X Receptor Deficiency, Scott Syndrome (TMEM16F)                  | Clear |
| <input checked="" type="checkbox"/> Polycystic Kidney Disease, PKD (PKD1)                                            | Clear |
| <input checked="" type="checkbox"/> Pompe's Disease (GAA, Finnish and Swedish Lapphund, Lapponian Herder Variant)    | Clear |
| <input checked="" type="checkbox"/> Prekallikrein Deficiency (KLKB1 Exon 8)                                          | Clear |
| <input checked="" type="checkbox"/> Primary Ciliary Dyskinesia, PCD (NME5, Alaskan Malamute Variant)                 | Clear |
| <input checked="" type="checkbox"/> Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3, Old English Sheepdog Variant)    | Clear |
| <input checked="" type="checkbox"/> Primary Hyperoxaluria (AGXT)                                                     | Clear |
| <input checked="" type="checkbox"/> Primary Lens Luxation (ADAMTS17)                                                 | Clear |
| <input checked="" type="checkbox"/> Primary Open Angle Glaucoma (ADAMTS17 Exon 11, Basset Fauve de Bretagne Variant) | Clear |
| <input checked="" type="checkbox"/> Primary Open Angle Glaucoma (ADAMTS10 Exon 17, Beagle Variant)                   | Clear |

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### OTHER RESULTS

✔ 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, 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

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# "BOSTON"

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### OTHER RESULTS

- ✓ 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
- ✓ 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 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

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### OTHER RESULTS

<input checked="" type="checkbox"/> Thrombopathia (RASGRP1 Exon 5, Basset Hound Variant)	Clear
<input checked="" type="checkbox"/> Thrombopathia (RASGRP1 Exon 8, Landseer Variant)	Clear
<input checked="" type="checkbox"/> Trapped Neutrophil Syndrome, TNS (VPS13B)	Clear
<input checked="" type="checkbox"/> Ullrich-like Congenital Muscular Dystrophy (COL6A3 Exon 10, Labrador Retriever Variant)	Clear
<input checked="" type="checkbox"/> Ullrich-like Congenital Muscular Dystrophy (COL6A1 Exon 3, Landseer Variant)	Clear
<input checked="" type="checkbox"/> Unilateral Deafness and Vestibular Syndrome (PTPRQ Exon 39, Doberman Pinscher)	Clear
<input checked="" type="checkbox"/> Urate Kidney & Bladder Stones (SLC2A9)	Clear
<input checked="" type="checkbox"/> Von Willebrand Disease Type I, Type I vWD (VWF)	Clear
<input checked="" type="checkbox"/> Von Willebrand Disease Type II, Type II vWD (VWF, Pointer Variant)	Clear
<input checked="" type="checkbox"/> Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)	Clear
<input checked="" type="checkbox"/> Von Willebrand Disease Type III, Type III vWD (VWF Intron 16, Nederlandse Kooikerhondje Variant)	Clear
<input checked="" type="checkbox"/> Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)	Clear
<input checked="" type="checkbox"/> X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)	Clear
<input checked="" type="checkbox"/> X-Linked Myotubular Myopathy (MTM1, Labrador Retriever Variant)	Clear
<input checked="" type="checkbox"/> X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)	Clear
<input checked="" type="checkbox"/> X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)	Clear
<input checked="" type="checkbox"/> X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)	Clear
<input checked="" type="checkbox"/> $\beta$ -Mannosidosis (MANBA Exon 16, Mixed-Breed Variant)	Clear

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## HEALTH REPORT

### Increased risk result

#### Intervertebral Disc Disease (Type I)

Boston inherited one copy of the variant we tested for Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD  
Boston is at increased risk for Type I IVDD

#### How to interpret this result

Boston has one copy 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

## HEALTH REPORT

### Notable result

#### ALT Activity

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

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

Boston 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 Boston's ALT activity above their current, healthy, ALT activity. As an increase above Boston'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.

## HEALTH REPORT

### Notable result

#### **Progressive Retinal Atrophy, crd4/crd1**

Boston inherited one copy of the variant we tested for Progressive Retinal Atrophy, crd4/crd1

#### **What does this result mean?**

This result should not impact Boston's health but it could have consequences for siblings or other related dogs if they inherited two copies of the variant. We recommend discussing this result with their owners or breeders if you are in contact.

#### **Impact on Breeding**

Your dog carries this variant and will pass it on to ~50% of his offspring.

#### **What is Progressive Retinal Atrophy, crd4/crd1?**

PRA-CRD4/crd1 is a retinal disease that causes progressive, non-painful vision loss over a 1-2 year period. The retina contains cells, called photoreceptors, that collect information about light and send signals to the brain. There are two types of photoreceptors: rods, for night vision and movement, and cones, for day vision and color. This type of PRA leads to early loss of cone cells, causing day blindness before night blindness.

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

The earliest ophthalmic signs are typically present by 6 months of age. There is a wide range in the age of when dogs become clinically affected, although the average age is approximately 5 years. Dogs as young as 6 months may be blind, while dogs as old as 10 may still have vision.

#### **How vets diagnose this condition**

Veterinarians use a focused light to examine the pupils. In affected dogs, the pupils will appear more dilated and slower to contract. Your vet may also use a lens to visualize the retina at the back of the eye to look for changes in the optic nerve or blood vessels. You may be referred to a veterinary ophthalmologist for a definitive diagnosis.

#### **How this condition is treated**

Currently, there is no definitive treatment for PRA. Supplements, including antioxidants, have been proposed for management of the disease, but have not been scientifically proven effective.

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

- Careful monitoring by your veterinarian will be required for the rest of your affected dog's life as secondary complications, including cataracts, can develop.
- With blind dogs, keeping furniture in the same location, making sure they are on a leash in unfamiliar territory, and training them to understand verbal commands are some of the ways to help them at home.



# "BOSTON"

## BOSTON



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## INBREEDING AND DIVERSITY

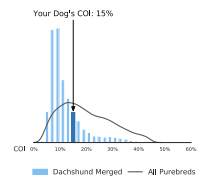
### CATEGORY

### 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.

15%

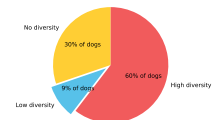


#### MHC Class II - DLA DRB1

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.

#### Low 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.

#### Low Diversity

How common is this amount of diversity in purebreds:

