



“FOREST”

OUR APPALACHIAN EVERGREEN FOREST



DNA Test Report

Test Date: September 4th, 2025

embk.me/ourappalachianevergreenforest

TRAITS: COAT COLOR

TRAIT

RESULT

E Locus (MC1R)

The E Locus determines if and where a dog can produce dark (black or brown) hair. Dogs with two copies of the recessive **e** variant do not produce dark hairs and will express a red pigment called pheomelanin over their entire body. The shade of red, which can range from a deep copper to white, depends on other genetic factors, including the Intensity loci. In addition to determining if a dog can develop dark hairs, the E Locus can give a dog a black “mask” or “widow’s peak” unless the dog has overriding coat color genetic factors.

Can have a melanistic mask (E_me or Ee)

Dogs with one or two copies of the **E_m** variant may have a melanistic mask (dark facial hair as commonly seen in the German Shepherd Dog and Pug). In the absence of **E_m**, dogs with the **E_g** variant can have a “grizzle” phenotype (darker color on the head and top with a melanistic “widow’s peak” and a lighter underside, commonly seen in the Afghan Hound and Borzoi and also referred to as “domino”). In the absence of both **E_m** and **E** variants, dogs with the **E_a** or **E_h** variants can express the grizzle phenotype. Additionally, a dog with any combination of two of the **E_g**, **E_a**, or **E_h** variants (example: **E_gE_a**) is also expected to express the grizzle phenotype.

K Locus (CBD103)

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

More likely to have a patterned haircoat (kyky)

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TRAIT

RESULT

Intensity Loci

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

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

A Locus (ASIP)

The A Locus controls switching between black and red pigment in hair cells, but it will only be expressed in dogs that are not **ee** at the E Locus and are **kyky** 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 at)

D Locus (MLPH)

The D locus result that we report is determined by three different genetic variants that can work together to cause diluted pigmentation. These are the common **d** allele, also known as "**d1**", and the less common alleles known as "**d2**" and "**d3**". Dogs with two **d** alleles, regardless of which variant, will have all black pigment lightened ("diluted") to gray, or brown pigment lightened to lighter brown in their hair, skin, and sometimes eyes. There are many breed-specific names for these dilute colors, such as "blue", "charcoal", "fawn", "silver", and "Isabella". Note that in certain breeds, dilute dogs have a higher incidence of Color Dilution Alopecia. Dogs with one **d** allele will not be dilute, but can pass the **d** allele on to their puppies.

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

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TRAIT

RESULT

Cocoa (HPS3)

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

No co alleles, not expressed (NN)

B Locus (TYRP1)

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

Black or gray hair and skin (BB)

Saddle Tan (RALY)

The "Saddle Tan" pattern causes the black hairs to recede into a "saddle" shape on the back, leaving a tan face, legs, and belly, as a dog ages. The Saddle Tan pattern is characteristic of breeds like the Corgi, Beagle, and German Shepherd. Dogs that have the **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** allele, so dogs that do not express **a** are not influenced by this gene.

Not 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 flash, parti, piebald, or extreme white (spsp)

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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)

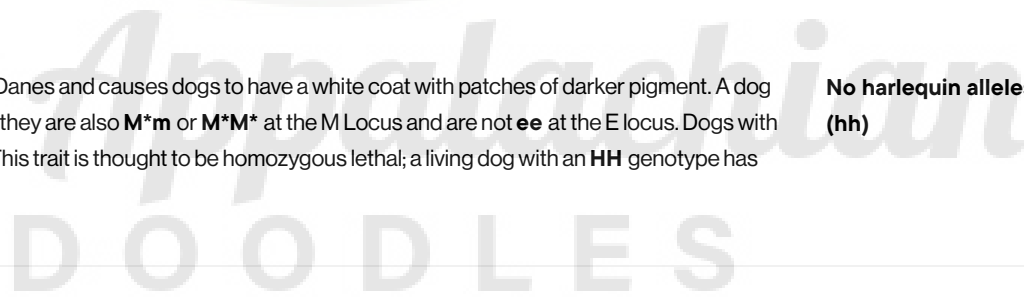
The R Locus regulates the presence or absence of the roan coat color pattern. Partial duplication of the USH2A gene is strongly associated with this coat pattern. Dogs with at least one **R** allele will likely have roaning on otherwise uniformly unpigmented white areas. Roan appears in white areas controlled 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, nonuniform roaning may appear as ticked, and not obviously roan. The roan pattern can appear with or without ticking.

Likely no impact on coat pattern (rr)

H Locus (Harlequin)

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

No harlequin alleles (hh)



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TRAIT

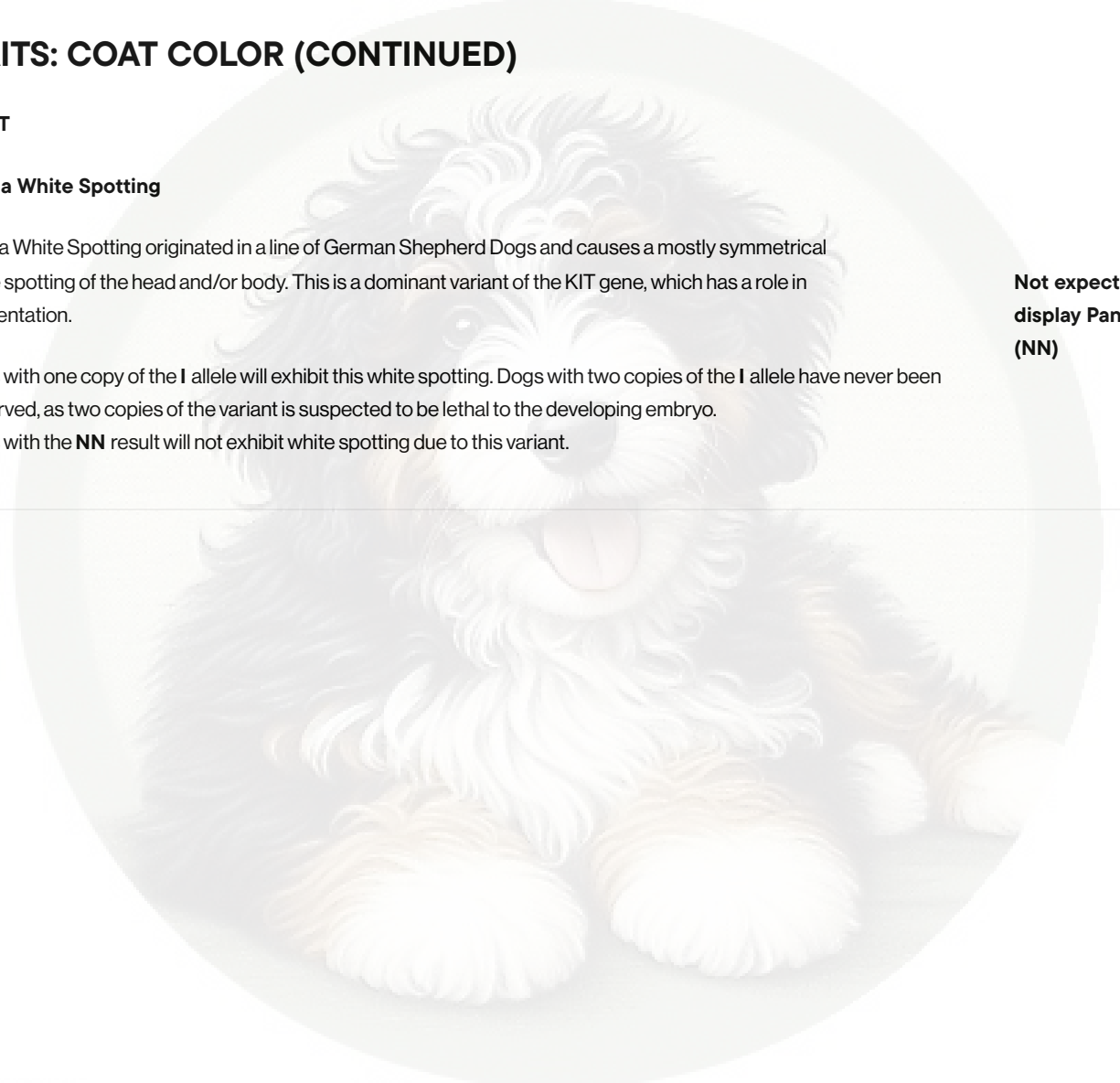
RESULT

Panda White Spotting

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

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

Not expected to display Panda pattern (NN)



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TRAITS: OTHER COAT TRAITS

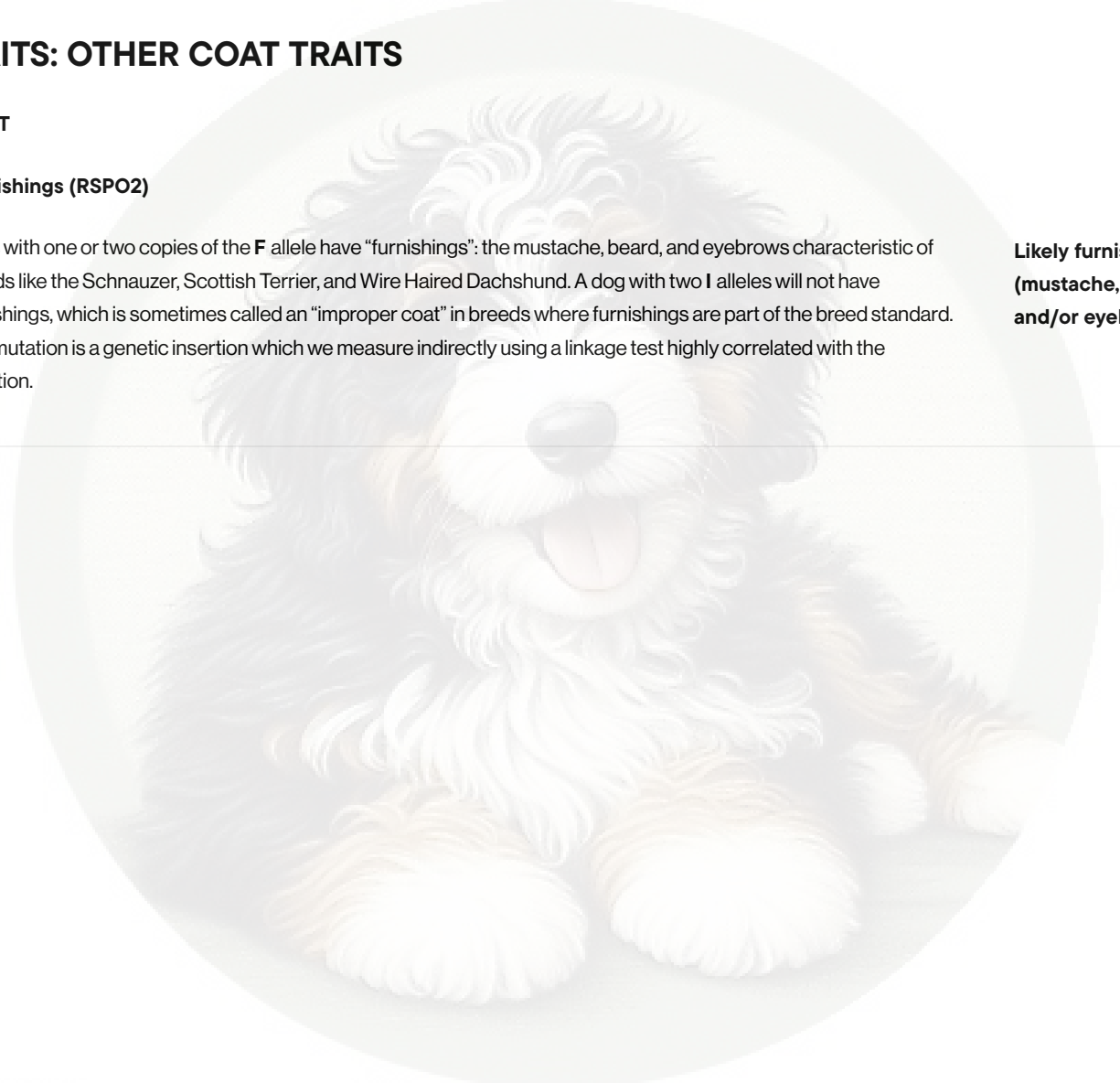
TRAIT

RESULT

Furnishings (RSPO2)

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

**Likely furnished
(mustache, beard,
and/or eyebrows) (FF)**



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TRAITS: OTHER COAT TRAITS (CONTINUED)

TRAIT

RESULT

Coat Length (FGF5)

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

The most common of these is the **Lh1** variant (G/T, CanFam3.1, chr32, g.4509367) and the less common ones are **Lh2** (C/T, CanFam3.1, chr32, g.4528639), **Lh3** (16bp deletion, CanFam3.1, chr32, g.4528616), and **Lh4** (GG insertion, CanFam3.1, chr32, g.4528621). The FGF5_Lh1 variant is found across many dog breeds. The less common alleles, FGF5_Lh2, have been found in the Akita, Samoyed, and Siberian Husky, FGF5_Lh3 have been found in the Eurasier, and FGF5_Lh4 have been found in the Afghan Hound, Eurasier, and French Bulldog.

Likely long coat (LhLh)

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

Interestingly, the Lh3 variant, a 16 base pair deletion, encompasses the Lh4 variant (GG insertion). The presence of one or two copies of Lh3 influences the outcome at the Lh4 locus. When two copies of Lh3 are present, there will be no reportable result for the FGF5_Lh4 locus. With one copy of Lh3, Lh4 can have either one copy of the variant allele or the normal allele. The overall FGF5 result remains unaffected by this.

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TRAITS: OTHER COAT TRAITS (CONTINUED)

TRAIT

RESULT

Shedding (MC5R)

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

Likely light shedding (CC)

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 curly coat (TT)

Hairlessness (FOXI3)

A duplication in the FOXI3 gene causes hairlessness over most of the body as well as changes in tooth shape and number. This mutation occurs in Peruvian Inca Orchid, Xoloitzcuintli (Mexican Hairless), and Chinese Crested (other hairless breeds have different mutations). Dogs with the **NDup** genotype are likely to be hairless while dogs with the **NN** genotype are likely to have a normal coat. The **DupDup** genotype has never been observed, suggesting that dogs with that genotype cannot survive to birth. Please note that this is a linkage test, so it may not be as predictive as direct tests of the mutation in some lines.

Very unlikely to be hairless (NN)

Hairlessness (SGK3)

Hairlessness in the American Hairless Terrier arises from a mutation in the SGK3 gene. Dogs with the **DD** result are likely to be hairless. Dogs with the **ND** genotype will have a normal coat, but can pass the **D** variant on to their offspring.

Very unlikely to be hairless (NN)

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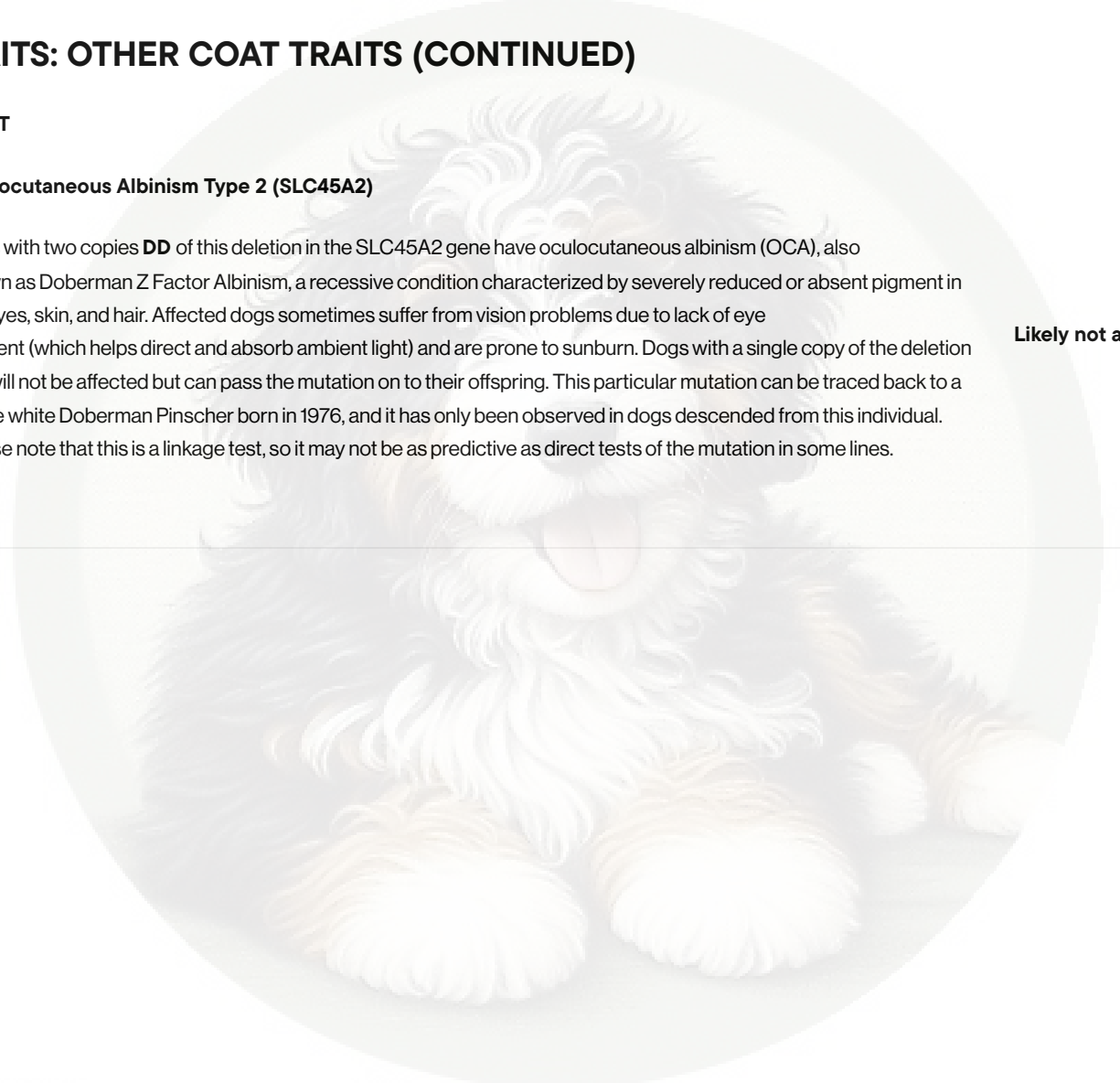
TRAIT

RESULT

Oculocutaneous Albinism Type 2 (SLC45A2)

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

Likely not albino (NN)



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TRAITS: OTHER BODY FEATURES

TRAIT

RESULT

Muzzle Length (BMP3)

Dogs in medium-length muzzle (mesocephalic) breeds like Staffordshire Terriers and Labradors, and long muzzle (dolichocephalic) breeds like Whippet and Collie have one, or more commonly two, copies of the ancestral **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

Chondrodysplasia (Chr. 18 FGF4 Retrogene)

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

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

Blue Eye Color (ALX4)

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

Less likely to have blue eyes (NN)

Back Muscling & Bulk, Large Breed (ACSL4)

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

Likely normal muscling (CC)

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TRAITS: BODY SIZE

TRAIT

RESULT

Body Size (IGF1)

The I allele is associated with smaller body size.

Smaller (II)

Body Size (IGFR1)

The A allele is associated with smaller body size.

Larger (GG)

Body Size (STC2)

The A allele is associated with smaller body size.

Larger (TT)

Body Size (GHR - E191K)

The A allele is associated with smaller body size.

Larger (GG)

Body Size (GHR - P177L)

The T allele is associated with smaller body size.

Larger (CC)

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TRAITS: PERFORMANCE

TRAIT

RESULT

Altitude Adaptation (EPAS1)

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

Normal altitude tolerance (GG)

Appetite (POMC)

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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