Sammy Snickers

# JOEL'S CHOCOLATE MALE



**DNA Test Report** 

Test Date: February 16th, 2024

embk.me/joelschocolatemale

**BREED ANCESTRY** 

Poodle (Small) : 100.0%

**GENETIC STATS** 

Predicted adult weight: 8 lbs

**TEST DETAILS** 

Kit number: EM-51197842 Swab number: 31220710810024

A+A+ KYKY bb DD Mm EmE 55 1-CDDY



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#### TRAITS: COAT COLOR

TRAIT RESULT

#### E Locus (MC1R)

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

Can have a melanistic mask (E<sup>m</sup>E)

#### K Locus (CBD103)

The K Locus  $\mathbf{K}^B$  allele "overrides" the A Locus, meaning that it prevents the A Locus genotype from affecting coat color. For this reason, the  $\mathbf{K}^B$  allele is referred to as the "dominant black" allele. As a result, dogs with at least one  $\mathbf{K}^B$  allele will usually have solid black or brown coats (or red/cream coats if they are  $\mathbf{e}\mathbf{e}$  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  $\mathbf{k}^y\mathbf{k}^y$  genotype will show a coat color pattern based on the genotype they have at the A Locus. Dogs who test as  $\mathbf{K}^B\mathbf{k}^y$  may be brindle rather than black or brown.

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





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## TRAITS: COAT COLOR (CONTINUED)

TRAIT RESULT

#### Intensity Loci LINKAGE

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

Any 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 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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## TRAITS: COAT COLOR (CONTINUED)

TRAIT RESULT

#### Cocoa (HPS3)

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

Brown 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 II genotype at this locus are more likely to be mostly black with tan points on the eyebrows, muzzle, and legs as commonly seen in the Doberman Pinscher and the Rottweiler. This gene modifies the A Locus at allele, so dogs that do not express at are not influenced by this gene.

Not saddle tan patterned (II)

#### S Locus (MITF)

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

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





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## TRAITS: COAT COLOR (CONTINUED)

TRAIT RESULT

#### M Locus (PMEL)

Merle coat patterning is common to several dog breeds including the Australian Shepherd, Catahoula Leopard Dog, and Shetland Sheepdog, among many others. Merle arises from an unstable SINE insertion (which we term the "M\*" allele) that disrupts activity of the pigmentary gene PMEL, leading to mottled or patchy coat color. Dogs with an M\*m result are likely to be phenotypically merle or could be "non-expressing" merle, meaning that the merle pattern is very subtle or not at all evident in their coat. Dogs with an M\*M\* result are likely to be phenotypically merle or double merle. Dogs with an mm result have no merle alleles and are unlikely to have a merle coat pattern.

Note that Embark does not currently distinguish between the recently described cryptic, atypical, atypical+, classic, and harlequin merle alleles. Our merle test only detects the presence, but not the length of the SINE insertion. We do not recommend making breeding decisions on this result alone. Please pursue further testing for allelic distinction prior to breeding decisions.

#### One merie allele; may express merie (M\*m)

Note: This locus includes several alleles. At the time this dog was genotyped Embark we could not distinguish all of the possible alleles.

#### R Locus (USH2A) LINKAGE

The R Locus regulates the presence or absence of the roan coat color pattern. Partial duplication of the USH2A gene is strongly associated with this coat pattern. Dogs with at least one **R** allele will likely have roaning on otherwise uniformly unpigmented white areas. Roan appears in white areas controlled by the S Locus but not in other white or cream areas created by other loci, such as the E Locus with **ee** along with Dilute Red Pigmentation by I Locus (for example, in Samoyeds). Mechanisms for controlling the extent of roaning are currently unknown, and roaning can appear in a uniform or non-uniform pattern. Further, non-uniform roaning may appear as ticked, and not obviously roan. The roan pattern can appear with or without ticking.

Likely no impact on coat pattern (rr)

#### H Locus (Harlequin)

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

No harlequin alleles (hh)





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#### TRAITS: OTHER COAT TRAITS

TRAIT RESULT

#### Furnishings (RSPO2) LINKAGE

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

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

#### Coat Length (FGF5)

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

Likely long coat (TT)

#### Shedding (MC5R)

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

Likely light shedding (TT)

#### 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)





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## TRAITS: OTHER COAT TRAITS (CONTINUED)

TRAIT RESULT

#### 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** variant on to their offspring.

Very unlikely to be hairless (NN)

#### 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)





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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 (AC)

#### 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) 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**

The T allele is associated with smaller 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.	Smaller (AA)	
Body Size (STC2)  The A allele is associated with smaller body size.	Intermediate (TA)	
Body Size (GHR - E191K)  The A allele is associated with smaller body size.	Smaller (AA)	
Body Size (GHR - P177L)	Intermediate (CT)	

# r light hair likely yellow or tan (Intermediate Red mentation)

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

# it is a linkage test?

ubloci	Genetic Result
ntensity_red_pigment_chr2	Red/Cream
ntensity_red_pigment_KITLG	Red/Red
ntensity_red_pigment_chr18	Red/Cream
ntensity_red_pigment_MFSD12	Red/Cream
ntensity_red_pigment_chr21	Red/Cream

## tions

ney et al 2021, Weich et al 2020, Hedan et al 2019



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#### **HEALTH REPORT**

#### How to interpret Joel's Chocolate Male's genetic health results:

If Joel's Chocolate Male 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 Joel's Chocolate Male 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 1 result that you should learn about.

Increased risk results (1)
Intervertebral Disc Disease (Type I)

Olear results

Breed-relevant (5)

Other (249)





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## **BREED-RELEVANT RESULTS**

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

0	Intervertebral Disc Disease (Type I) (FGF4 retrogene - CFA12)	ncreased risk
$\odot$	GM2 Gangliosidosis (HEXB, Poodle Variant)	Clear
0	Neonatal Encephalopathy with Seizures, NEWS (ATF2)	Clear
$\odot$	Osteochondrodysplasia (SLC13A1, Poodle Variant)	Clear
$\odot$	Progressive Retinal Atrophy, prcd (PRCD Exon 1)	Clear
0	Von Willebrand Disease Type I, Type I vWD (VWF)	Clear

