



Test Date: December 10th, 2019

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## **BREED MIX**

Lagotto Romagnolo : 100.0%

## **GENETIC STATS**

Wolfiness: 1.2 % **MEDIUM** Predicted adult weight: **32 lbs** Genetic age: n/a (Date of birth unknown)

## **TEST DETAILS**

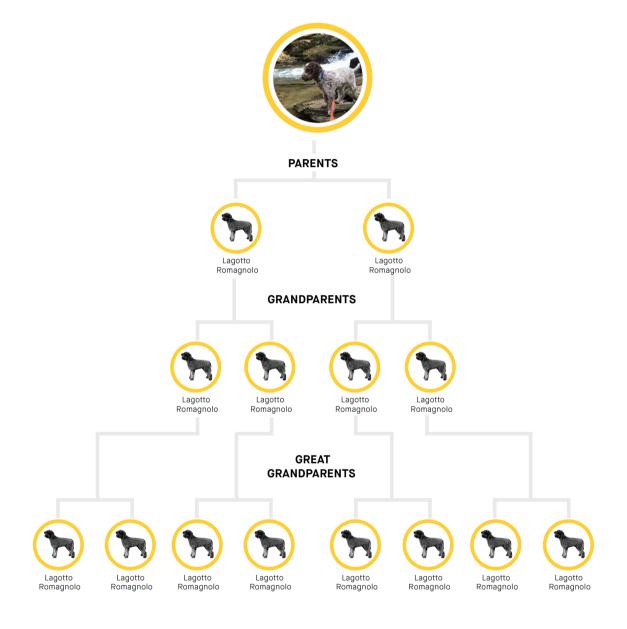
Kit number: EM-3076975 Swab number: 31019073307081



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## **FAMILY TREE**



Our algorithms predict this is the most likely family tree to explain Vito's breed mix, but this family tree may not be the only possible one.







### Fun Fact

Lagotto Romagnolos were originally bred as water retrievers, which is evident in their name: "lagotto" means little lake in Italian. Test Date: December 10th, 2019

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## LAGOTTO ROMAGNOLO

Lagotto Romagnolos are an Italian breed of dog from the Romagna region of Italy. Sporting curly hair and charming faces, they were originally bred as hunting dogs during the Medieval Period; however, today they are mostly kept as pets and as a different kind of hunting dog-Lagotto Romagnolos are expert truffle dogs. Their wonderful sense of smell makes them a great candidate for finding and unearthing truffles, rare and expensive mushrooms that are considered culinary delicacies. Lagotto Romagnolos are very much "working dogs" and likely won't be satisfied if they spend most of their time indoors. Because of this, they do best with families that are active and spend a lot of time outside. If interested in truffle hunting, prospective owners can purchase Lagotto Romagnolos that were specially trained to find truffles. If kept simply as pets, however, it is very important that Lagotto Romagnolos are given enough time outside and adequate exercise, or they can become bored and subsequently destructive. Due to this, they aren't ideal apartment dogs-though they can adapt if need be-and would generally do better in a suburban or rural environment. Mental exercise is just as important as physical exercise; Lagotto Romagnolos are very intelligent and can get bored easily. Enrolling Lagotto Romagnolos in a dog sport or obedience training is a great way to keep them occupied. Lagotto Romagnolos are very loving and loyal dogs and are a great choice for families with children and other pets. They get along well with other dogs and can do very well with cats if socialized with them from a young age. They have sweet demeanors and are relatively easy to train with proper instruction. Another great thing about Lagotto Romagnolos is that they shed very little, and they are as close to hypoallergenic as a dog can be (though no dog is completely safe for people who are severely allergic to dogs). They make great companions for families that are sensitive to allergy issues and who are looking to add an active and sweet dog to their home.

### RELATED BREEDS



Barbet Sibling breed



Spanish Water Dog Sibling breed

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## MATERNAL LINE



Through Vito'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: A1b

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

### HAPLOTYPE: A361/409/611

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







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## PATERNAL LINE



Through Vito'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: A1a

Some of the wolves that became the original dogs in Central Asia around 15,000 years ago came from this long and distinguished line of male dogs. After domestication, they followed their humans from Asia to Europe and then didn't stop there. They took root in Europe, eventually becoming the dogs that founded the Vizsla breed 1,000 years ago. The Vizsla is a Central European hunting dog, and all male Vizslas descend from this line. During the Age of Exploration, like their owners, these pooches went by the philosophy, "Have sail, will travel!" From the windy plains of Patagonia to the snug and homey towns of the American Midwest, the beaches of a Pacific paradise, and the broad expanse of the Australian outback, these dogs followed their masters to the outposts of empires. Whether through good fortune or superior genetics, dogs from the A1a lineage traveled the globe and took root across the world. Now you find village dogs from this line frolicking on Polynesian beaches, hanging out in villages across the Americas, and scavenging throughout Old World settlements.

### HAPLOTYPE: H1a.42

Part of the A1a haplogroup, the H1a.42 haplotype occurs most commonly in Airedale Terriers, Lagotto Romagnolos and American Pit Bull Terriers.

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Dilution Alopecia which causes hair loss in some patches.

**DNA Test Report** 



embk.me/vito24 TRAITS: BASE COAT COLOR TRAIT RESULT Dark or Light Fur | E (Extension) Locus | Gene: Melanocortin Receptor 1 (MC1R) | Genetic Result: Ee This gene helps determine whether a dog can produce dark (black or brown) hairs or lighter yellow or red hairs. Any result except for ee means that the dog can produce dark hairs. An ee result means that the dog does not produce dark hairs at all, and will have lighter yellow or red hairs over their entire body. Can have dark fur Did You Know? If a dog has a ee result then the fur's actual shade can range from a deep copper to yellow/gold to cream - the exact color cannot be predicted solely from this result, and will depend on other genetic factors. Brown or Black Pigment | B (Brown) Locus | Gene: Tyrosinase Related Protein 1 (TYRP1) | Genetic Result: bb This gene helps determine whether a dog produces brown or black pigments. Dogs with a **bb** result produce brown pigment instead of black in both their hair and skin, while dogs with a Bb or BB result produce black pigment. Dogs that have ee at the E (Extension) Locus and bb at this B (Brown) Locus are Brown fur and skin likely to have red or cream coats and brown noses, eye rims, and footpads, which is sometimes referred to as "Dudley Nose" in Labrador Retrievers. Did You Know? "Liver" or "chocolate" is the preferred color term for brown in most breeds; in the Doberman Pinscher it is referred to as "red". Color Dilution | D (Dilute) Locus | Gene: Melanophilin (MLPH) | Genetic Result: DD This gene helps determine whether a dog has lighter "diluted" pigment. A dog with a Dd or DD result will not be dilute. A dog with a dd result will have all their black or brown pigment lightened ("diluted") to gray or light brown, and sometimes lightens red pigment to cream. This affects their fur, skin, and sometimes Dark (non-dilute) fur and skin eye color. Did You Know? There are many breed-specific names for these dilute colors, such as "blue", "charcoal", "fawn", "silver", and "Isabella". Dilute dogs, especially in certain breeds, have a higher incidence of Color

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## TRAITS: COAT COLOR MODIFIERS

## TRAIT RESULT Hidden Patterning | K (Dominant Black) Locus | Gene: Canine Beta-Defensin 103 (CBD103) | Genetic Result: **K<sup>B</sup>K<sup>B</sup>** This gene helps determine whether the dog has a black coat. Dogs with a kyky result will show a coat color pattern based on the result they have at the A (Agouti) Locus. A K<sup>B</sup>K<sup>B</sup> or K<sup>B</sup>k<sup>y</sup> result means the dog is dominant black, which overrides the fur pattern that would otherwise be determined by the A (Agouti) Locus. These dogs will usually have solid black or brown coats, or if they have ee at the E (Extension) brown fur coat Locus then red/cream coats, regardless of their result at the A (Agouti) Locus. Dogs who test as K<sup>B</sup>k<sup>y</sup> may be brindle rather than black or brown. Did You Know? Even if a dog is "dominant black" several other genes could still impact the dog's fur and cause other patterns, such as white spotting. Body Pattern | A (Agouti) Locus | Gene: Agouti Signalling Protein (ASIP) | Genetic Result: No Call This gene is responsible for causing different coat patterns. It only affects the fur of dogs that do not have ee at the E (Extension) Locus and do have kyky at the K (Dominant Black) Locus. It controls switching between black and red pigment in hair cells, which means that it can cause a dog to have hairs that have sections of black and sections of red/cream, or hairs with different colors on different parts of the dog's No impact on coat body. Sable or Fawn dogs have a mostly or entirely red coat with some interspersed black hairs. Agouti or pattern 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.

Did You Know? The ASIP gene causes interesting coat patterns in many other species of animals as well as dogs.

### Facial Fur Pattern | E (Extension) Locus | Gene: Melanocortin Receptor 1 (MC1R) | Genetic Result: Ee

In addition to determining if a dog can develop dark fur at all, this gene 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 E<sup>m</sup> in their result will have a mask, which is dark facial fur as seen in the German Shepherd and Pug. Dogs with no E<sup>m</sup> in their result but one or two copies of E<sup>g</sup> will instead have a "widow's peak", which is dark forehead fur.

Did You Know? The widow's peak is seen in the Afghan Hound and Borzoi, where it is called either "grizzle" or "domino".



More likely to have a mostly solid black or

No dark mask or grizzle facial fur patterns





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# TRAITS: COAT COLOR MODIFIERS (CONTINUED)

### TRAIT

Saddle Tan | Gene: RALY | Genetic Result: II

The *RALY* gene is responsible for the Saddle Tan coat pattern, where a dog's black hairs recede into a "saddle" shape on the back as the dog ages, leaving a tan face, legs, and belly. This gene only impacts dogs that have **a<sup>t</sup>a<sup>t</sup>** at the A (Agouti) Locus, do not have **ee** at the E (Extension) Locus, and do not have **K<sup>B</sup>** at the K (Dominant Black) Locus. Dogs with one or two copies of the normal "N" allele are likely to have a saddle tan pattern. Dogs that with a **II** result (where "I" represents the mutant allele) 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.

**Did You Know?** The Saddle Tan pattern is characteristic of breeds like the Corgi, Beagle, and German Shepherd.

Merle | M (Merle) Locus | Gene: PMEL | Genetic Result: mm

This gene is responsible for mottled or patchy coat color in some dogs. Dogs with an **M\*m** result are likely to have merle coat patterning or be "phantom" merle (where the merle allele is not obvious in their coat). Dogs with an **M\*M\*** result are likely to have merle or double merle coat patterning. Dogs with an **mm** result are unlikely to have a merle coat pattern.

**Did You Know?** Merle coat patterning is common to several dog breeds including the Australian Shepherd, Catahoula Leopard Dog, and Shetland Sheepdog.

RESULT

No impact on coat pattern

Unlikely to have merle pattern

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# TRAITS: OTHER COAT TRAITS

TRAIT	RESULT
Furnishings LINKAGE   Gene: RSP02   Genetic Result: FF	
This gene is responsible for "furnishings", which is another name for the mustache, beard, and eyebrows that are characteristic of breeds like the Schnauzer, Scottish Terrier, and Wire Haired Dachshund. A dog with an <b>FF</b> or <b>FI</b> result is likely to have furnishings. A dog with an <b>II</b> result will not have furnishings. We measure this result using a linkage test.	Likely furnished (mustache, beard, and/or eyebrows)
<b>Did You Know?</b> In breeds that are expected to have furnishings, dogs without furnishings are the exception - this is sometimes called an "improper coat".	
Coat Length   Gene: FGF5   Genetic Result: TT	
This gene is known to affect hair/fur length in many different species, including cats, dogs, mice, and humans. In dogs, a <b>TT</b> result means the dog is likely to have a long, silky coat as seen in the Yorkshire Terrier and the Long Haired Whippet. A <b>GG</b> or <b>GT</b> result is likely to mean a shorter coat, like in the Boxer or the American Staffordshire Terrier.	Likely long coat
Did You Know? In certain breeds, such as Corgi, the long coat is described as "fluff."	
Shedding   Gene: MC5R   Genetic Result: TT	
This gene affects how much a dog sheds. Dogs with furnishings or wire-haired coats tend to be low shedders regardless of their result for this gene. In other dogs, a <b>CC</b> or <b>CT</b> result indicates heavy or seasonal shedding, like many Labradors and German Shepherd Dogs. Dogs with a <b>TT</b> result tend to be lighter shedders, like Boxers, Shih Tzus and Chihuahuas.	Likely light shedding
Coat Texture   Gene: KRT71   Genetic Result: TT	
For dogs with long fur, dogs with a <b>TT</b> or <b>CT</b> result will likely have a wavy or curly coat like the coat of Poodles and Bichon Frises. Dogs with a <b>CC</b> result will likely have a straight coat—unless the dog has a "Likely Furnished" result for the Furnishings trait, since this can also make the coat more curly.	Likely curly coat
Did You Know? Dogs with short coats may have straight coats, whatever result they have for this gene.	

Hairlessness (Xolo type) LINKAGE | Gene: FOX/3 | Genetic Result: NN





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# TRAITS: OTHER COAT TRAITS (CONTINUED)

TRAIT	RESULT
Hairlessness (Terrier type)   Gene: SGK3   Genetic Result: NN	Van unlikaluta ha
This gene is responsible for Hairlessness in the American Hairless Terrier. Dogs with the <b>ND</b> result are likely to be hairless. Dogs with the <b>NN</b> result are likely to have a normal coat.	Very unlikely to be hairless
Oculocutaneous Albinism Type 2 LINKAGE   Gene: SLC45A2   Genetic Result: NN	
This gene causes oculocutaneous albinism type 2 (OCA2), also known as Doberman Z Factor Albinism. Dogs with a <b>DD</b> result will have OCA2. Effects include severely reduced or absent pigment in the eyes, skin, and hair, and sometimes vision problems due to lack of eye pigment (which helps direct and absorb ambient light) and are prone to sunburn. Dogs with a <b>ND</b> result will not be affected, but can pass the mutation on to their offspring. We measure this result using a linkage test.	Likely not albino
<b>Did You Know?</b> 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.	





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## TRAITS: OTHER BODY FEATURES

### TRAIT RESULT Muzzle Length | Gene: BMP3 | Genetic Result: CC This gene affects muzzle length. A dog with a AC or CC result is likely to have a medium-length muzzle like a Staffordshire Terrier or Labrador, or a long muzzle like a Whippet or Collie. A dog with a AA result is likely to have a short muzzle, like an English Bulldog, Pug, or Pekingese. Likely medium or long muzzle Did You Know? At least five different genes affect snout length in dogs, with BMP3 being the only one with a known causal mutation. For example, the muzzle length of some breeds, including the long-snouted Scottish Terrier or the short-snouted Japanese Chin, appear to be caused by other genes. This means your dog may have a long or short snout due to other genetic factors. Embark is working to figure out what these might be. Tail Length | Gene: T | Genetic Result: CC This is one of the genes that can cause a short bobtail. Most dogs have a CC result and a long tail. Dogs with a CG result are likely to have a bobtail, which is an unusually short or absent tail. This can be seen in many "natural bobtail" breeds including the Pembroke Welsh Corgi, the Australian Shepherd, and the Likely normal-length Brittany Spaniel. Dogs with GG genotypes have not been observed, suggesting that dogs with such a result do not survive to birth. tail Did You Know? 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, it is not always caused by this gene. This suggests that other unknown genetic effects can also lead to a natural bobtail. Hind Dew Claws | Gene: LMBR1 | Genetic Result: CC This is one of the genes that can cause hind dew claws, which are extra, nonfunctional digits located midway between a dog's paw and hock. Dogs with a CT or TT result have about a 50% chance of having Unlikely to have hind hind dewclaws. Hind dew claws can also be caused by other, still unknown, genes. Embark is working to dew claws figure those out.

Did You Know? Hind dew claws are commonly found in certain breeds such as the Saint Bernard.

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to future discoveries!

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# TRAITS: OTHER BODY FEATURES (CONTINUED)

TRAIT	RESULT
Back Muscling & Bulk (Large Breed)   Gene: ACSL4   Genetic Result: CC	
This gene can cause 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. A dog with the <b>TT</b> result is likely to have heavy muscling. Leaner-shaped large breed dogs like the Great Dane, Irish Wolfhound, and Scottish Deerhound generally have a <b>CC</b> result. The <b>TC</b> result also indicates likely normal muscling.	Likely normal muscling
<b>Did You Know?</b> This gene does not seem to affect muscling in small or even mid-sized dog breeds with lots of back muscling, including the American Staffordshire Terrier, Boston Terrier, and the English Bulldog.	
Eye Color LINKAGE   Gene: ALX4   Genetic Result: NN	
This gene is associated with blue eyes in Arctic breeds like Siberian Husky as well as tri-colored (non- merle) Australian Shepherds. Dogs with a <b>DupDup</b> or <b>NDup</b> result are more likely to have blue eyes, although some dogs may have only one blue eye or may not have blue eyes at all; nevertheless, they can still pass blue eyes to their offspring. Dogs with a <b>NN</b> result may have blue eyes due to other factors, such as merle or white spotting. We measure this result using a linkage test.	Less likely to have blue eyes
<b>Did You Know?</b> Embark researchers discovered this gene by studying data from dogs like yours. Who knows what we will be able to discover next? Answer the questions on our research surveys to contribute	





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RAITS: BODY SIZE			
TRAIT		RESULT	
Body Size 1   Gene: IGF1   Genetic I	Result: NI		
° °	nfluence the size of a dog. A result of <b>II</b> for this gene is associated with s associated with larger body size.	Intermediate	
Body Size 2   Gene: IGFR1   Genetic	c Result: AA		
-	nfluence the size of a dog. A result of <b>AA</b> for this gene is associated with s associated with s associated with larger body size.	Smaller	
Body Size 3   Gene: STC2   Genetic	c Result: <b>TT</b>		
This is one of several genes that in smaller body size. A result of <b>TT</b> is	nfluence the size of a dog. A result of <b>AA</b> for this gene is associated with associated with associated with larger body size.	Larger	
Body Size 4   Gene: GHR - E191K   G	Genetic Result: <b>GG</b>		
	nfluence the size of a dog. A result of <b>AA</b> for this gene is associated with s associated with s associated with larger body size.	Larger	
Body Size 5   Gene: GHR - P177L   G	Genetic Result: <b>CC</b>		
-	nfluence the size of a dog. A result of <b>TT</b> for this gene is associated with s associated with larger body size.	Larger	



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TRAITS: PERFORMANCE TRAIT RESULT Altitude Adaptation | Gene: EPAS1 | Genetic Result: GG This gene causes dogs to be especially tolerant of low oxygen environments, such as those found at high Normal altitude elevations. Dogs with a AA or GA result will be less susceptible to "altitude sickness." tolerance Did You Know? This gene was originally identified in breeds from high altitude areas such as the Tibetan Mastiff. Appetite LINKAGE | Gene: POMC | Genetic Result: NN This gene influences eating behavior. An ND or DD result would predict higher food motivation compared to NN result, increasing the likelihood 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 Normal food our blog post (embarkvet.com/resources/blog/pomc-dogs). We measure this result using a linkage test. motivation Did You Know? POMC is actually short for "proopiomelanocortin," and is a large protein that is broken up into several smaller proteins that have biological activity. The smaller proteins generated from POMC control, among other things, distribution of pigment to the hair and skin cells, appetite, and energy expenditure.

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## TRAITS: GENETIC DIVERSITY

TRAIT

Inbreeding | Gene: n/a | Genetic Result: 16%

Inbreeding is a measure of how closely related this dog's parents were. The higher the number, the more closely related the parents. In general, greater inbreeding is associated with increased incidence of genetically inherited conditions.

### Immune Response 1 | Gene: DRB1 | Genetic Result: High Diversity

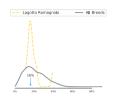
Diversity in the Major Histocompatibility Complex (MHC) region of the genome has been found in some studies to be associated with the incidence of certain autoimmune diseases. Dogs that have less diversity in the MHC region—i.e. the Dog Leukocyte Antigen (DLA) inherited from the mother is similar to the DLA inherited from the father—are considered less immunologically diverse. A High Diversity result means the dog has two highly dissimilar haplotypes. A Low Diversity result means the dog has two similar but not identical haplotypes. A No Diversity result means the dog has inherited identical haplotypes from both parents. Some studies have shown associations between certain DRB1 haplotypes and autoimmune diseases such as Cushing's disease, but these findings have yet to be scientifically validated.

### Immune Response 2 | Gene: DQA1 and DQB1 | Genetic Result: High Diversity

# Diversity in the Major Histocompatibility Complex (MHC) region of the genome has been found in some studies to be associated with the incidence of certain autoimmune diseases. Dogs that have less diversity in the MHC region—i.e. the Dog Leukocyte Antigen (DLA) inherited from the mother is similar to the DLA inherited from the father—are considered less immunologically diverse. A High Diversity result means the dog has two highly dissimilar haplotypes. A Low Diversity result means the dog has two similar but not identical haplotypes. A No Diversity result means the dog has inherited identical haplotypes from both parents. A number of studies have shown correlations of DQA-DQB1 haplotypes and certain autoimmune diseases; however, these have not yet been scientifically validated.

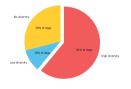
### RESULT

### 16%



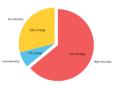
### **High Diversity**

How common is this amount of diversity in purebreds:



### **High Diversity**

How common is this amount of diversity in purebreds:



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## **CLINICAL TOOLS**

These clinical genetic traits can inform clinical decisions and diagnoses. These traits do not predict a disease state or increased risk for disease. We currently assess one clinical tool: Alanine Aminotransferase Activity.

### Alanine Aminotransferase Activity result: Normal

Vito has two normal alleles at ALT.

More information on Alanine Aminotransferase Activity:

The liver enzyme alanine aminotransferase, or ALT, is one of several values your veterinarian measures on routine blood work to gauge liver health. Dogs with one or more copies of the "A" allele are likely to have a lower baseline ALT activity ("low normal") than dogs with zero copies of the "A" allele ("normal"). This means that your veterinarian may recommend blood work to establish an individualized baseline ALT value during an annual wellness exam or before starting certain medications. You and your veterinarian would then be able to monitor your dog for any deviation from this established baseline. Please note that this mutation should never cause an increase in your dog's ALT activity and does not cause liver disease. If your dog has high ALT activity, please consult your veterinarian.

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## HEALTH

Good news! Vito did not test positive for any of the genetic conditions that Embark screens for.

It is still important to let your veterinarian know these results because they could help guide Vito's diagnosis and treatment if he gets sick in the future.

# O AT RISK

CARRIER



## **CARRIER CONDITIONS**

**CARRIER** status: This indicates the dog has inherited a recessive allele for a genetic trait or mutation. This is not enough to cause symptoms of the disease, but is important to bear in mind if the dog ever has offspring.

Carrier System: Neurologic Condition: Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LGI2)

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## BENIGN FAMILIAL JUVENILE EPILEPSY, REMITTING FOCAL EPILEPSY

(LGI2)			
Carrier			0
LGI2 (Exon 8) gene name	AA clear	AT carrier	TT at risk

Vito is a carrier for a variant in the LGI2 gene. While he or she is unlikely to exhibit signs of disease, as a carrier, he or she will pass the variant on to the next generation. If you choose to breed Vito, we highly recommend testing any potential mates for this variant. Breeding to another carrier is not recommended as this will produce a number of affected puppies.

## DESCRIPTION

A disorder of young dogs, this causes intermittent seizures that resolve with age. Affected dogs can begin experiencing seizures of varying frequency, duration, and severity, ranging from simple focal seizures where the dog is tremoring but still able to walk, eat, and respond to stimulus to complete immobilization and loss of consciousness. Puppies appear normal between episodes, though those with severe seizures can display an abnormal, uncoordinated gait after an episode. Treatment for BFJE is usually supportive; dogs typically grow out of the disease and suffer no ill effects later in life.

# CITATIONS

Seppala et al 2011 (http://www.ncbi.nlm.nih.gov/pubmed/21829378), Jokinen et al 2007 (http://www.ncbi.nlm.nih.gov/pubmed/17552452), Jokinen et al 2015 (http://www.ncbi.nlm.nih.gov/pubmed/25945683)





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## **OTHER CONDITIONS**

Good news! Vito tested clear for 2 genetic conditions that are common in his breed.

- Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU (SLC2A9)
- Lagotto Storage Disease (ATG4D)





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# FULL TEST PANEL

# Vito is also clear of 178 other genetic health conditions that Embark tests for.

To help ensure healthy breeds, every test includes analysis of our full panel of over 160 genetic health conditions.

The following pages list out all the other genetic health conditions that Vito tested clear for.



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- MDR1 Drug Sensitivity (MDR1) (Chromosome 14)
- P2Y12 Receptor Platelet Disorder (P2RY12) (Chromosome 23)
- Factor IX Deficiency, Hemophilia B (F9 Exon 7, Terrier Variant) (Chromosome X)
- Factor IX Deficiency, Hemophilia B (F9 Exon 7, Rhodesian Ridgeback Variant) (Chromosome X)
- Factor VII Deficiency (F7 Exon 5) (Chromosome 22)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 10, Boxer Variant) (Chromosome X)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 11, Shepherd Variant 1) (Chromosome X)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 1, Shepherd Variant 2) (Chromosome X)
- Thrombopathia (RASGRP2 Exon 5, Basset Hound Variant) (Chromosome 18)
- Thrombopathia (RASGRP2 Exon 8) (Chromosome 18)
- Thrombopathia (RASGRP2 Exon 5, American Eskimo Dog Variant) (Chromosome 18)
- Von Willebrand Disease Type III, Type III vWD (VWF Exon 4) (Chromosome 27)
- Von Willebrand Disease Type I (VWF) (Chromosome 27)
- Von Willebrand Disease Type II, Type II vWD (VWF) (Chromosome 27)
- Canine Leukocyte Adhesion Deficiency Type III, LAD3 (FERMT3) (Chromosome 18)
- Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cavalier King Charles Spaniel Variant) (Chromosome 24)
- Canine Elliptocytosis (SPTB Exon 30) (Chromosome 8)
- Glanzmann's Thrombasthenia Type I (ITGA2B Exon 12) (Chromosome 9)
- May-Hegglin Anomaly (MYH9) (Chromosome 10)
- Prekallikrein Deficiency (KLKB1 Exon 8) (Chromosome 16)
- Pyruvate Kinase Deficiency (PKLR Exon 5) (Chromosome 7)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Labrador Variant) (Chromosome 7)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Pug Variant) (Chromosome 7)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Beagle Variant) (Chromosome 7)
- Pyruvate Kinase Deficiency (PKLR Exon 10) (Chromosome 7)
- Trapped Neutrophil Syndrome (VPS13B) (Chromosome 13)
- Ligneous Membranitis, LM (PLG) (Chromosome 1)
- Platelet factor X receptor deficiency, Scott Syndrome (TMEM16F) (Chromosome 27)
- Methemoglobinemia CYB5R3 (Chromosome 10)
- Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant) (Chromosome 17)
- Complement 3 Deficiency, C3 Deficiency (C3) (Chromosome 20)
- Severe Combined Immunodeficiency (PRKDC) (Chromosome 29)
- Severe Combined Immunodeficiency (RAG1) (Chromosome 18)
- X-linked Severe Combined Immunodeficiency (IL2RG Variant 1) (Chromosome X)
- X-linked Severe Combined Immunodeficiency (IL2RG Variant 2) (Chromosome X)
- Progressive Retinal Atrophy, rcd1 Rod-cone dysplasia, rcd1 (PDE6B Exon 21 Irish Setter Variant) (Chromosome 3)
- Progressive Retinal Atrophy, rcd3 Rod-cone dysplasia, rcd3 (PDE6A) (Chromosome 4)
- Progressive Retinal Atrophy, CNGA (CNGA1 Exon 9) (Chromosome 13)





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- Progressive Retinal Atrophy, prcd Progressive rod-cone degeneration (PRCD Exon 1) (Chromosome 9)
- Progressive Retinal Atrophy (CNGB1) (Chromosome 2)
- Progressive Retinal Atrophy (SAG) (Chromosome 25)
- Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3) (Chromosome 37)
- Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8) (Chromosome 8)
- Progressive Retinal Atrophy, crd1 (PDE6B) (Chromosome 3)
- Progressive Retinal Atrophy, crd2 (IQCB1) (Chromosome 33)
- Progressive Retinal Atrophy crd4/cord1 (RPGRIP1) (Chromosome 15)
- Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1) (Chromosome 37)
- Achromatopsia (CNGA3 Exon 7 German Shepherd Variant) (Chromosome 10)
- Achromatopsia (CNGA3 Exon 7 Labrador Retriever Variant) (Chromosome 10)
- Autosomal Dominant Progressive Retinal Atrophy (RHO) (Chromosome 20)
- Canine Multifocal Retinopathy cmr1 (BEST1 Exon 2) (Chromosome 18)
- Canine Multifocal Retinopathy cmr2 (BEST1 Exon 5) (Chromosome 18)
- Canine Multifocal Retinopathy cmr3 (BEST1 Exon 10 Deletion) (Chromosome 18)
- Canine Multifocal Retinopathy cmr3 (BEST1 Exon 10 SNP) (Chromosome 18)
- Glaucoma Primary Open Angle Glaucoma (ADAMTS10 Exon 9) (Chromosome 20)
- Glaucoma Primary Open Angle Glaucoma (ADAMTS10 Exon 17) (Chromosome 20)
- Glaucoma Primary Open Angle Glaucoma (ADAMTS17 Exon 11) (Chromosome 3)
- Glaucoma Primary Open Angle Glaucoma (ADAMTS17 Exon 2) (Chromosome 3)
- Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Shepherd Variant) (Chromosome 5)
- Primary Lens Luxation (ADAMTS17) (Chromosome 3)
- Congenital Stationary Night Blindness (RPE65) (Chromosome 6)
- Macular Corneal Dystrophy, MCD (CHST6) (Chromosome 5)
- 2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis (APRT) (Chromosome 5)
- Cystinuria Type I-A (SLC3A1) (Chromosome 10)
- Cystinuria Type II-A (SLC3A1) (Chromosome 10)
- Cystinuria Type II-B (SLC7A9) (Chromosome 1)
- Polycystic Kidney Disease, PKD (PKD1) (Chromosome 6)
- Primary Hyperoxaluria (AGXT) (Chromosome 25)
- Protein Losing Nephropathy, PLN (NPHS1) (Chromosome 1)
- X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2) (Chromosome X)
- Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 3) (Chromosome 25)
- Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3) (Chromosome 34)
- Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatosis, Dry Eye Curly Coat Syndrome, CKCSID (FAM83H Exon 5)
  (Chromosome 13)
- X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia (EDA Intron 8) (Chromosome X)
- Renal Cystadenocarcinoma and Nodular Dermatofibrosis, RCND (FLCN Exon 7) (Chromosome 5)
- Canine Fucosidosis (FUCA1) (Chromosome 2)





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- Glycogen Storage Disease Type II, Pompe's Disease, GSD II (GAA) (Chromosome 9)
- Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC) (Chromosome 9)
- Glycogen Storage Disease Type IIIA, GSD IIIA (AGL) (Chromosome 6)
- Mucopolysaccharidosis Type I, MPS I (IDUA) (Chromosome 3)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 1) (Chromosome 9)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 2) (Chromosome 9)
- Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5) (Chromosome 6)
- Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3) (Chromosome 6)
- Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Whippet and English Springer Spaniel Variant) (Chromosome 27)
- Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Wachtelhund Variant) (Chromosome 27)
- Neuronal Ceroid Lipofuscinosis 1, NCL 1 (PPT1 Exon 8) (Chromosome 15)
- Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4) (Chromosome 21)
- Neuronal Ceroid Lipofuscinosis 1, Cerebellar Ataxia, NCL-A (ARSG Exon 2) (Chromosome 9)
- Neuronal Ceroid Lipofuscinosis 1, NCL 1 (CLN5 Border Collie Variant) (Chromosome 22)
- Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7) (Chromosome 30)
- Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 English Setter Variant) (Chromosome 37)
- Neuronal Ceroid Lipofuscinosis (MFSD8) (Chromosome 19)
- Neuronal Ceroid Lipofuscinosis (CLN8 Australian Shepherd Variant) (Chromosome 37)
- Neuronal Ceroid Lipofuscinosis 10, NCL 10 (CTSD Exon 5) (Chromosome 18)
- Neuronal Ceroid Lipofuscinosis (CLN5 Golden Retriever Variant) (Chromosome 22)
- Adult-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Tibetan Terrier Variant) (Chromosome 2)
- Late Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Australian Cattle Dog Variant) (Chromosome 2)
- GM1 Gangliosidosis (GLB1 Exon 15 Shiba Inu Variant) (Chromosome 23)
- GM1 Gangliosidosis (GLB1 Exon 15 Alaskan Husky Variant) (Chromosome 23)
- GM1 Gangliosidosis (GLB1 Exon 2) (Chromosome 23)
- GM2 Gangliosidosis (HEXB, Poodle Variant) (Chromosome 2)
- GM2 Gangliosidosis (HEXA) (Chromosome 30)
- Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5) (Chromosome 8)
- Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Italian Greyhound Variant) (Chromosome 13)
- Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Parson Russell Terrier Variant) (Chromosome 13)
- Persistent Mullerian Duct Syndrome, PMDS (AMHR2) (Chromosome 27)
- Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MYO7A) (Chromosome 21)
- Shar-Pei Autoinflammatory Disease, SPAID, Shar-Pei Fever (MTBP) (Chromosome 13)
- Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy (SLC19A3) (Chromosome 25)
- Alexander Disease (GFAP) (Chromosome 9)
- Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration, NCCD (SPTBN2) (Chromosome 18)
- Cerebellar Ataxia, Progressive Early-Onset Cerebellar Ataxia (SEL1L) (Chromosome 8)
- Cerebellar Hypoplasia (VLDLR) (Chromosome 1)





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- Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA (CAPN1) (Chromosome 18)
- Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10) (Chromosome 38)
- Degenerative Myelopathy, DM (SOD1A) (Chromosome 31)
- Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2) (Chromosome 2)
- Hypomyelination and Tremors (FNIP2) (Chromosome 15)
- Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP) (Chromosome X)
- Neuroaxonal Dystrophy, NAD (Spanish Water Dog Variant) (Chromosome 8)
- Neuroaxonal Dystrophy, NAD (Rottweiler Variant) (Chromosome 5)
- L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH) (Chromosome 0)
- Neonatal Encephalopathy with Seizures, NEWS (ATF2) (Chromosome 36)
- Polyneuropathy, NDRG1 Greyhound Variant (NDRG1 Exon 15) (Chromosome 13)
- Polyneuropathy, NDRG1 Malamute Variant (NDRG1 Exon 4) (Chromosome 13)
- Narcolepsy (HCRTR2 Intron 6) (Chromosome 12)
- Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 15) (Chromosome 1)
- Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 4) (Chromosome 1)
- Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV (RAB3GAP1, Rottweiler Variant) (Chromosome 19)
- Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (GDNF-AS) (Chromosome 4)
- Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1 (LPN1, ARHGEF10) (Chromosome 16)
- Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SeSAME/EAST Syndrome (KCNJ10) (Chromosome 38)
- Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 (ATP1B2) (Chromosome 5)
- Dilated Cardiomyopathy, DCM1 (PDK4) (Chromosome 14)
- Dilated Cardiomyopathy, DCM2 (TTN) (Chromosome 36)
- Long QT Syndrome (KCNQ1) (Chromosome 18)
- Muscular Dystrophy Cavalier King Charles Spaniel Variant 1 (Chromosome X)
- Muscular Dystrophy Muscular Dystrophy (DMD Pembroke Welsh Corgi Variant ) (Chromosome X)
- Muscular Dystrophy Muscular Dystrophy (DMD Golden Retriever Variant) (Chromosome X)
- Centronuclear Myopathy (PTPLA) (Chromosome 2)
- Exercise-Induced Collapse (DNM1) (Chromosome 9)
- Inherited Myopathy of Great Danes (BIN1) (Chromosome 19)
- Myostatin Deficiency, Bully Whippet Syndrome (MSTN) (Chromosome 37)
- Myotonia Congenita (CLCN1 Exon 7) (Chromosome 16)
- Myotonia Congenita (CLCN1 Exon 23) (Chromosome 16)
- Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM (MTM1, Labrador Variant) (Chromosome X)
- Hypocatalasia, Acatalasemia (CAT) (Chromosome 18)
- Pyruvate Dehydrogenase Deficiency (PDP1) (Chromosome 29)
- Malignant Hyperthermia (RYR1) (Chromosome 1)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53) (Chromosome 2)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8) (Chromosome 2)





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# **CLEAR CONDITIONS**

- Lundehund Syndrome (LEPREL1) (Chromosome 34)
- Congenital Myasthenic Syndrome (CHAT) (Chromosome 28)
- Congenital Myasthenic Syndrome (COLQ) (Chromosome 23)
- Episodic Falling Syndrome (BCAN) (Chromosome 7)
- Dystrophic Epidermolysis Bullosa (COL7A1) (Chromosome 20)
- Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1) (Chromosome 7)
- Ichthyosis, Epidermolytic Hyperkeratosis (KRT10) (Chromosome 9)
- Ichthyosis (PNPLA1) (Chromosome 12)
- Ichthyosis (SLC27A4) (Chromosome 9)
- Ichthyosis (NIPAL4) (Chromosome 4)
- Focal Non-Epidermolytic Palmoplantar Keratoderma, Pachyonychia Congenita (KRT16) (Chromosome 9)
- Hereditary Footpad Hyperkeratosis (FAM83G) (Chromosome 5)
- Hereditary Nasal Parakeratosis (SUV39H2) (Chromosome 2)
- Musladin-Lueke Syndrome (ADAMTSL2) (Chromosome 9)
- Bald Thigh Syndrome (IGFBP5) (Chromosome 37)
- Cleft Lip and/or Cleft Palate (ADAMTS20) (Chromosome 27)
- Hereditary Vitamin D-Resistant Rickets (VDR) (Chromosome 27)
- Oculoskeletal Dysplasia 1, Dwarfism-Retinal Dysplasia, OSD1 (COL9A3, Labrador Retriever) (Chromosome 24)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2) (Chromosome 14)
- Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1) (Chromosome 21)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1) (Chromosome 9)
- Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1) (Chromosome 14)
- Skeletal Dysplasia 2, SD2 (COL11A2) (Chromosome 12)
- Craniomandibular Osteopathy, CMO (SLC37A2) (Chromosome 5)
- Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene CFA12) (Chromosome 12)
- Chondrodystrophy, Norwegian Elkhound and Karelian Bear Dog Variant (ITGA10) (Chromosome 17)

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