research partner of
Cornell University
College of Veterinary Medicine

Embark Veterinary, Inc. | Genetic Analysis Report for the Russian Toy

About Embark

Embark Veterinary is the industry-leading canine genetics company serving breeders, veterinarians, and pet owners. Embark's DNA testing products assess over 200,000 genetic markers to offer the most accurate and comprehensive results on the market today. Embark is committed to advancing canine research and making genetic discoveries that advance the health of dogs, and which bring the company closer to achieving its mission of helping dogs live longer and healthier lives.

Research Methodology

The goal of this report is to allow breeders, breed clubs, health organizations, veterinarians, and individual dog owners to better understand the occurrence of genetic variants known to impact risk for health conditions; informed diversity measures and genotype frequencies of traits within a breed population. Importantly, key data can be further evaluated to identify trends within a breed over time.

Data referenced in this report was generated based upon dogs currently genotyped by Embark whose owners have consented to participate in research. This report does not necessarily represent the global genotype frequencies for this breed. Data reflected in this report includes:

- Frequency of detected genetic health risk variants relevant to the breed
- Frequency of genetic health variants detected within the breed that may not have been directly studied yet, to inform early detection
- Frequency of physical trait alleles
- Genetic coefficient of inbreeding (COI) statistics and population distributions

We are on a mission to improve the life and longevity of all dogs through the power of science.
The following summary is intended to provide a high level overview for each category of data requirements outlined above.
Please contact breeders@embarkvet.com with any questions or comments specific to this data.

Summary of Report Findings:

Results as of:	June 13, 2024
Sample Size:	More than 100 dogs

OFA-Registerable Health Conditions | Genetic Health Report

Health Condition			
Degenerative Myelopathy, DM	clear: 98.3%	carrier, heterozygote recessive: 1.6%	

Other Health Conditions with >1% Impacted | Genetic Health Report

Health Condition			
Bald Thigh Syndrome	clear: 99.1%	carrier, heterozygote recessive: 0.8%	
Canine Multifocal Retinopathy, cmr1	clear: 98.3%	carrier, heterozygote recessive: 1.6%	
Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (This is a linkage test)	clear: 57.3%	at risk, homozygote dominant: 8.1%	at risk, heterozygote, dominant: 34.4%
Congenital Macrothrombocytopenia	clear: 91.8%	carrier, heterozygote recessive: 8.1%	
Dilated Cardiomyopathy, DCM1	clear: 98.3%	at risk, heterozygote, dominant: 1.6%	
Dilated Cardiomyopathy, DCM2	clear: 98.3%	at risk, heterozygote, dominant: 1.6%	
Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV	clear: 96.7%	carrier, heterozygote recessive: 0.8%	
Progressive Retinal Atrophy, crd4/cord1	clear: 73.7%	carrier, heterozygote recessive: 25.4%	at risk, homozygote recessive: 0.8%
Proportionate Dwarfism	clear: 76.4%	carrier, heterozygote recessive: 23.5%	

Alanine Aminotransferase Activity* | Genetic Health Report

Health Condition			
Alanine Aminotransferase Activity	clear: 54.0%	at risk, homozygote codominant: 8.1%	at risk, heterozygote codominant: 37.7%

**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. For more information, visit [Embark's website.](#)*

Physical Traits | Genetic Traits Report

Trait Allele Frequencies	**Description**						
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 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.	AtAt: 55.6%	AyAt: 34.6%	AyAy: 7.2%	Ata: 0.8%	AwAt: 0.8%	Aya: 0.8%
B Locus (TYRP1)	Dogs with two copies of the 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".	Bb: 50.0%	bb: 29.0%	BB: 20.9%			
Body Size (GHR - E191K)	The allele is associated with smaller body size.	AA: 92.7%	GA: 7.2%				

Body Size (GHR - P177L)	The allele is associated with smaller body size.	CC: 94.3%	CT: 5.6%				
Body Size (IGF1)	The allele is associated with smaller body size.	II: 84.6%	NI: 13.7%	NN: 1.6%			
Body Size (IGFR1)	The allele is associated with smaller body size.	GG: 48.3%	GA: 40.3%	AA: 11.2%			
Body Size (STC2)	The allele is associated with smaller body size.	AA: 74.1%	TA: 25.0%	TT: 0.8%			
Coat Length (FGF5)		LhLh: 78.2%	ShLh: 17.7%	ShSh: 4.0%			
Coat Texture (KRT71)	Dogs with a long coat and at least one copy of the allele but still have straight coats.	CC: 98.3%	CT: 1.6%				
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 allele on to their puppies.	DD: 82.2%	Dd: 15.3%	dd: 0.8%			
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 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”).	EE: 76.6%	Ee: 14.5%	EmE: 5.6%	ee: 1.6%	Eme: 1.6%	
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 dogs will have hind dewclaws.	CC: 74.1%	CT: 23.3%	TT: 2.4%			
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 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.	Intermediate: 82.2%	High: 17.7%				

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 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.	CC: 87.0%	AC: 12.9%				
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 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.	rr: 99.0%	Rr: 0.9%				
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.	SS: 99.1%	Ssp: 0.8%				
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 are not influenced by this gene.	II: 52.0%	NI: 37.3%	NN: 9.7%			

Shedding (MC5R)	Dogs with at least one copy of the ancestral 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.	TT: 99.1%	CT: 0.8%				

Genetic Coefficient of Inbreeding | Genetic Diversity Report

Median COI	75th Percentile	25th Percentile
Median: 16.1%	75th Percentile: 19.6%	25th Percentile: 10.7%

Genetic coefficient of inbreeding (COI) is the most accurate method for measuring inbreeding. Unlike pedigree-based COI calculations, genetic COI directly evaluates each dog's DNA to identify the proportion that results from inbreeding. Embark's genetic COI assesses over 200,000 genetic markers and can detect generations of inbreeding that cannot be accounted for with typical pedigree documentation. Genetic COI is more precise than tests using fewer markers - ultimately providing more information to better understand a dog's genetic health and to guide strategic breeding crosses to mitigate inbreeding depression.

We recommend genetic testing of breeding dogs prior to mating to assess the expected average litter COI and to consider a preference for pairings that would produce litters with a lower COI.

Conclusion

Embark Veterinary, Inc., is committed to our mission of ending preventable disease in dogs. Preserving genetic diversity while breeding away from heritable disease is key to this mission, and every breed club and organization that joins in this effort brings us closer to this reality. Together we can accelerate the pace of discovery and develop new tests to improve canine health.

This report is one of the many resources and services provided to support breed club organizations. Partnership benefits also include discounted pricing, expert educational content, event support and more.

Please contact breeders@embarkvet.com to learn more about the information included in this report.