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# COOPER

## Veterinary Report by Embark

embarkvet.com

Test Date: December 3rd, 2022

### Customer-supplied information

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Owner Name: Mary Frances Withers

Dog Name: Cooper

Sex: Male (intact)

Date of birth: 07/25/19

Breed type: purebred

Breed: Miniature/MAS-type Australian Shepherd

Breed registration: American Kennel Club (AKC)

DN58858205

Microchip: N/A

### Genetic summary

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Genetic breed identification:

**Australian Shepherd Group**

Predicted adult weight: **35 lbs**

Calculated from 17 size genes.

Breed ancestry:

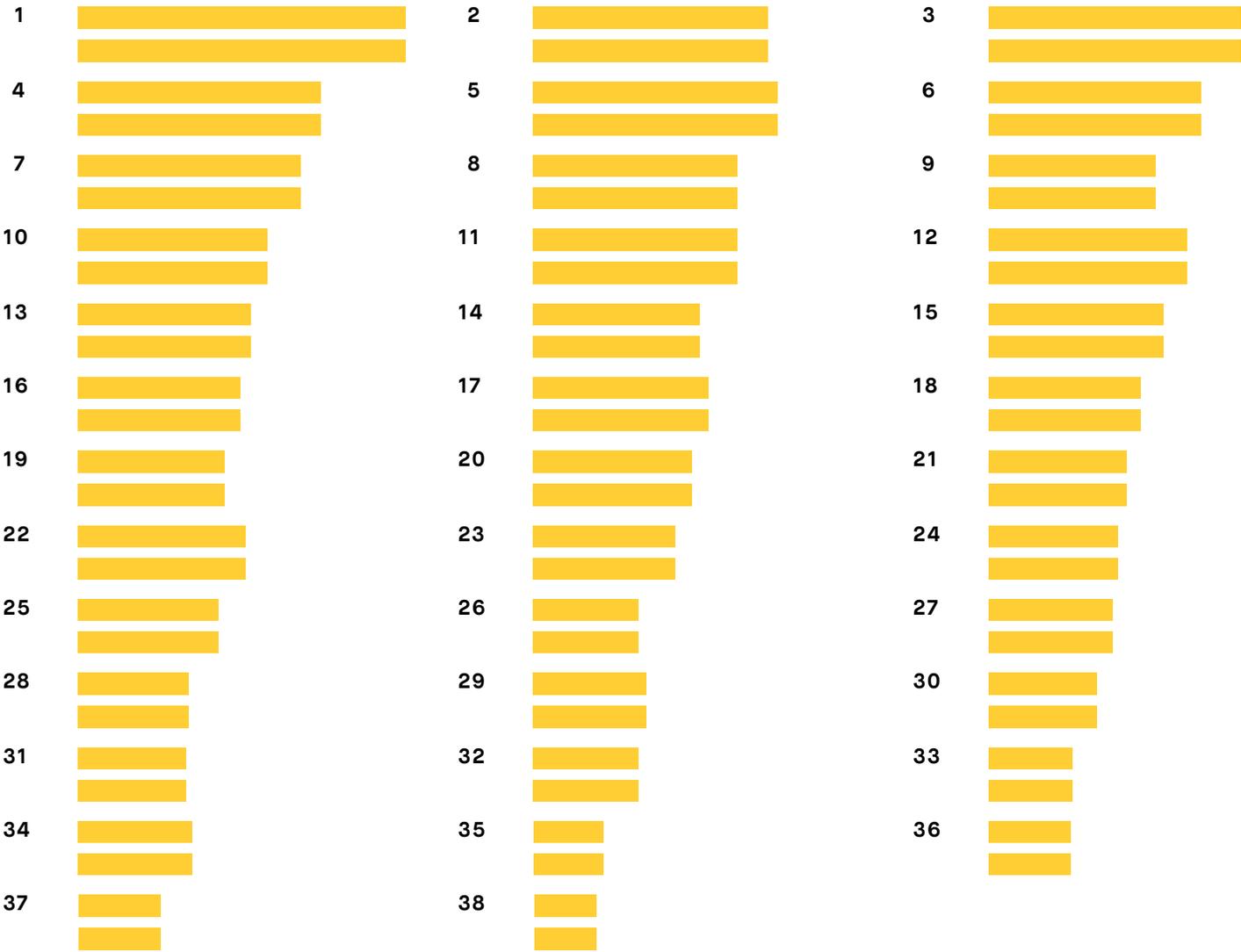
 **Miniature/MAS-type Australian Shepherd:**  
**100.0%**

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Life stage: **Young adult**

Based on date of birth provided.

# Karyogram (Chromosome painting)



# Clinical Tools

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These clinical genetic tools can inform clinical decisions and diagnoses. These tools do not predict increased risk for disease.

## **Alanine Aminotransferase Activity (GPT)**

 Cooper's baseline ALT level may be Low Normal

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

Cooper has one copy of a variant associated with reduced ALT activity as measured on veterinary blood chemistry panels. Please inform your veterinarian that Cooper has this genotype, as ALT is often used as an indicator of liver health and Cooper is likely to have a lower than average resting ALT activity. As such, an increase in Cooper's ALT activity could be evidence of liver damage, even if it is within normal limits by standard ALT reference ranges.

### **What is Alanine Aminotransferase Activity?**

Alanine aminotransferase (ALT) is a clinical tool that can be used by veterinarians to better monitor liver health. This result is not associated with liver disease. ALT is one of several values veterinarians measure on routine blood work to evaluate the liver. It is a naturally occurring enzyme located in liver cells that helps break down protein. When the liver is damaged or inflamed, ALT is released into the bloodstream.

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

Genetic testing is the only way to provide your veterinarian with this clinical tool.

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

Veterinarians may recommend blood work to establish a baseline ALT value for healthy dogs with one or two copies of this variant.

# Health Report

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

If Cooper 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 Cooper 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.



**Cooper is at increased risk for one genetic health condition.**

And inherited one variant that you should learn more about.

**MDR1 Drug Sensitivity**



**Canine Multifocal Retinopathy, cmr1**



**Breed-Relevant Genetic Conditions**

8 variants not detected



**Additional Genetic Conditions**

224 variants not detected



# Health Report

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## MDR1 Drug Sensitivity (ABCB1)

 Cooper inherited one copy of the variant we tested

Cooper is at increased risk for MDR1

### How to interpret this result

Cooper has one copy of a variant at the ABCB1 gene and is at risk for displaying adverse drug reactions. While he may not be as severely affected as a dog with two copies of the ABCB1 drug sensitivity allele, normal dosages of drugs could still have potentially severe effects on Cooper. Please inform your veterinarian that Cooper carries this variant; it is essential that they know this information before prescribing drugs.

### What is MDR1 Drug Sensitivity?

Sensitivity to certain classes of drugs, notably the parasiticide ivermectin, as well as certain gastroprotectant and anti-cancer medications, occurs in dogs with a mutation in the ABCB1 gene.

### When signs & symptoms develop in affected dogs

Symptoms arise after a dog has received an MDR1 problem drug or dosage, and can range from vomiting and diarrhea to lethargy, seizures, or coma.

### Signs & symptoms

MDR1 often presents in young adulthood, only because this is most commonly when a dog is first exposed to a problem drug like high dose ivermectin or acepromazine.

### How vets diagnose this condition

This is usually a retroactive diagnosis after a dog has an adverse reaction to a problem drug-- however, genetic testing could help you avoid a first reaction altogether.

### How this condition is treated

MDR1 is perfectly avoidable simply by avoiding the problem drugs, or problem dosages.

### Actions to take if your dog is affected

- Review the MDR1 Problem Drug List as described by Washington State University and notify your veterinarian to flag this in your dog's file!
- Farm dogs with MDR1 may also benefit if they are either kept away from herds where ivermectin is used as a routine antiparasitic, or if another form of antiparasitic is used in areas that they are working.

# Health Report

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## Canine Multifocal Retinopathy, cmr1 (BEST1 Exon 2)

 Cooper inherited one copy of the variant we tested

### What does this result mean?

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

### Impact on Breeding

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

### What is Canine Multifocal Retinopathy, cmr1?

This is a non-progressive retinal disease that, in rare cases, can lead to vision loss. Dogs with larger lesions can suffer from vision loss. CMR is fairly non-progressive; new lesions will typically stop forming by the time a dog is an adult, and some lesions will even regress with time.

### When signs & symptoms develop in affected dogs

cmr1 typically develops in puppies at 11 to 16 weeks and becomes stable by the time they are 1 year old.

### How vets diagnose this condition

CMR is typically only identified when a vet examines the back of the eye which, in dogs with CMR, reveals multiple retinal abnormalities that range from small, flat folds (called "retinal folds") to larger, irregularly edged raised lesions (called "geographic lesions").

### How this condition is treated

Currently, there is no treatment for CMR. However, CMR rarely affects vision to a significant degree, and as we stated, even the associated retinal abnormalities can regress over time.

### Actions to take if your dog is affected

- Carefully monitoring your dog's vision and seeking a veterinary ophthalmologist's opinion if you are concerned are the best ways you can help your affected dog.

# Breed-Relevant Conditions Tested

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Cooper did not have the variants that we tested for, that are relevant to his breed:

- ✔ Progressive Retinal Atrophy, prcd (PRCD Exon 1)
- ✔ Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1)
- ✔ Day Blindness, Cone Degeneration, Achromatopsia (CNGB3 Deletion, Alaskan Malamute Variant)
- ✔ Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9, Australian Shepherd Variant)
- ✔ Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU (SLC2A9)
- ✔ Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7, Australian Shepherd Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8, Australian Shepherd Variant)
- ✔ Craniomandibular Osteopathy, CMO (SLC37A2)

# Additional Conditions Tested

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 Cooper did not have the variants that we tested for, in the following conditions that the potential effect on dogs with Cooper's breed may not yet be known.

- ✔ P2Y12 Receptor Platelet Disorder (P2Y12)
- ✔ Factor IX Deficiency, Hemophilia B (F9 Exon 7, Terrier Variant)
- ✔ Factor IX Deficiency, Hemophilia B (F9 Exon 7, Rhodesian Ridgeback Variant)
- ✔ Factor VII Deficiency (F7 Exon 5)
- ✔ Factor VIII Deficiency, Hemophilia A (F8 Exon 10, Boxer Variant)
- ✔ Factor VIII Deficiency, Hemophilia A (F8 Exon 11, German Shepherd Variant 1)
- ✔ Factor VIII Deficiency, Hemophilia A (F8 Exon 1, German Shepherd Variant 2)
- ✔ Thrombopathia (RASGRP1 Exon 5, Basset Hound Variant)
- ✔ Thrombopathia (RASGRP1 Exon 8, Landseer Variant)
- ✔ Thrombopathia (RASGRP1 Exon 5, American Eskimo Dog Variant)
- ✔ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4, Terrier Variant)
- ✔ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7, Shetland Sheepdog Variant)
- ✔ Von Willebrand Disease Type I, Type I vWD (VWF)
- ✔ Von Willebrand Disease Type II, Type II vWD (VWF, Pointer Variant)
- ✔ Canine Leukocyte Adhesion Deficiency Type I, CLAD I (ITGB2, Setter Variant)
- ✔ Canine Leukocyte Adhesion Deficiency Type III, CLAD III (FERMT3, German Shepherd Variant)
- ✔ Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant)
- ✔ Canine Elliptocytosis (SPTB Exon 30)
- ✔ Glanzmann's Thrombasthenia Type I (ITGA2B Exon 13, Great Pyrenees Variant)
- ✔ Glanzmann's Thrombasthenia Type I (ITGA2B Exon 12, Otterhound Variant)

# Additional Conditions Tested

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- ✔ **May-Hegglin Anomaly (MYH9)**
- ✔ **Prekallikrein Deficiency (KLKB1 Exon 8)**
- ✔ **Pyruvate Kinase Deficiency (PKLR Exon 5, Basenji Variant)**
- ✔ **Pyruvate Kinase Deficiency (PKLR Exon 7, Labrador Retriever Variant)**
- ✔ **Pyruvate Kinase Deficiency (PKLR Exon 7, Pug Variant)**
- ✔ **Pyruvate Kinase Deficiency (PKLR Exon 7, Beagle Variant)**
- ✔ **Pyruvate Kinase Deficiency (PKLR Exon 10, Terrier Variant)**
- ✔ **Trapped Neutrophil Syndrome, TNS (VPS13B)**
- ✔ **Ligneous Membranitis, LM (PLG)**
- ✔ **Platelet Factor X Receptor Deficiency, Scott Syndrome (TMEM16F)**
- ✔ **Methemoglobinemia (CYB5R3)**
- ✔ **Bernard-Soulier Syndrome, BSS (GP9, Cocker Spaniel Variant)**
- ✔ **Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)**
- ✔ **Congenital Hypothyroidism (TPO, Rat, Toy, Hairless Terrier Variant)**
- ✔ **Congenital Dyshormonogenic Hypothyroidism with Goiter (SLC5A5, Shih Tzu Variant)**
- ✔ **Complement 3 Deficiency, C3 Deficiency (C3)**
- ✔ **Severe Combined Immunodeficiency, SCID (PRKDC, Terrier Variant)**
- ✔ **Severe Combined Immunodeficiency, SCID (RAG1, Wetterhoun Variant)**
- ✔ **X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG Exon 1, Basset Hound Variant)**
- ✔ **X-linked Severe Combined Immunodeficiency, X-SCID (IL2RG, Corgi Variant)**

# Additional Conditions Tested

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- ✔ Progressive Retinal Atrophy, rcd1 (PDE6B Exon 21, Irish Setter Variant)
- ✔ Progressive Retinal Atrophy, rcd3 (PDE6A)
- ✔ Progressive Retinal Atrophy, CNGA (CNGA1 Exon 9)
- ✔ Progressive Retinal Atrophy, PRA1 (CNGB1)
- ✔ Progressive Retinal Atrophy (SAG)
- ✔ Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)
- ✔ Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)
- ✔ Progressive Retinal Atrophy, crd1 (PDE6B, American Staffordshire Terrier Variant)
- ✔ Progressive Retinal Atrophy, crd4/cord1 (RPGRIP1)
- ✔ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)
- ✔ Progressive Retinal Atrophy, PRA3 (FAM161A)
- ✔ Day Blindness, Cone Degeneration, Achromatopsia (CNGB3 Exon 6, German Shorthaired Pointer Variant)
- ✔ Achromatopsia (CNGA3 Exon 7, German Shepherd Variant)
- ✔ Achromatopsia (CNGA3 Exon 7, Labrador Retriever Variant)
- ✔ Autosomal Dominant Progressive Retinal Atrophy (RHO)
- ✔ Canine Multifocal Retinopathy, cmr2 (BEST1 Exon 5, Coton de Tulear Variant)
- ✔ Canine Multifocal Retinopathy, cmr3 (BEST1 Exon 10 Deletion, Finnish and Swedish Lapphund, Lapponian Herder Variant)
- ✔ Primary Open Angle Glaucoma (ADAMTS10 Exon 9, Norwegian Elkhound Variant)
- ✔ Primary Open Angle Glaucoma (ADAMTS10 Exon 17, Beagle Variant)
- ✔ Primary Open Angle Glaucoma (ADAMTS17 Exon 11, Basset Fauve de Bretagne Variant)

# Additional Conditions Tested

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- ✔ Primary Open Angle Glaucoma and Primary Lens Luxation (ADAMTS17 Exon 2, Chinese Shar-Pei Variant)
- ✔ Goniodysgenesis and Glaucoma, Pectinate Ligament Dysplasia, PLD (OLFM3)
- ✔ Primary Lens Luxation (ADAMTS17)
- ✔ Congenital Stationary Night Blindness (RPE65, Briard Variant)
- ✔ Congenital Stationary Night Blindness (LRIT3, Beagle Variant)
- ✔ Macular Corneal Dystrophy, MCD (CHST6)
- ✔ 2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis (APRT)
- ✔ Cystinuria Type I-A (SLC3A1, Newfoundland Variant)
- ✔ Cystinuria Type II-A (SLC3A1, Australian Cattle Dog Variant)
- ✔ Cystinuria Type II-B (SLC7A9, Miniature Pinscher Variant)
- ✔ Polycystic Kidney Disease, PKD (PKD1)
- ✔ Primary Hyperoxaluria (AGXT)
- ✔ Protein Losing Nephropathy, PLN (NPHS1)
- ✔ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)
- ✔ Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 30, English Springer Spaniel Variant)
- ✔ Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 3, Cocker Spaniel Variant)
- ✔ Fanconi Syndrome (FAN1, Basenji Variant)
- ✔ Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3, Old English Sheepdog Variant)
- ✔ Primary Ciliary Dyskinesia, PCD (NME5, Alaskan Malamute Variant)
- ✔ Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, Dry Eye Curly Coat Syndrome, CKCSID (FAM83H Exon 5)

# Additional Conditions Tested

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- ✔ X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia, XHED (EDA Intron 8)
- ✔ Renal Cystadenocarcinoma and Nodular Dermatofibrosis, RCND (FLCN Exon 7)
- ✔ Canine Fucosidosis (FUCA1)
- ✔ Glycogen Storage Disease Type II, Pompe's Disease, GSD II (GAA, Finnish and Swedish Lapphund, Lapponian Herder Variant)
- ✔ Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC, Maltese Variant)
- ✔ Glycogen Storage Disease Type IIIA, GSD IIIA (AGL, Curly Coated Retriever Variant)
- ✔ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6, Dachshund Variant)
- ✔ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6, New Zealand Huntaway Variant)
- ✔ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5, Terrier Brasileiro Variant)
- ✔ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3, German Shepherd Variant)
- ✔ Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM, Whippet and English Springer Spaniel Variant)
- ✔ Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM, Wachtelhund Variant)
- ✔ Lagotto Storage Disease (ATG4D)
- ✔ Neuronal Ceroid Lipofuscinosis 1, NCL 1 (PPT1 Exon 8, Dachshund Variant 1)
- ✔ Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4, Dachshund Variant 2)
- ✔ Neuronal Ceroid Lipofuscinosis, Cerebellar Ataxia, NCL4A (ARSG Exon 2, American Staffordshire Terrier Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 5, NCL 5 (CLN5 Exon 4 SNP, Border Collie Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 Exon 2, English Setter Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 7, NCL 7 (MFSD8, Chihuahua and Chinese Crested Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 10, NCL 10 (CTSD Exon 5, American Bulldog Variant)

# Additional Conditions Tested

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- ✔ Neuronal Ceroid Lipofuscinosis 5, NCL 5 (CLN5 Exon 4 Deletion, Golden Retriever Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 Insertion, Saluki Variant)
- ✔ Late-Onset Neuronal Ceroid Lipofuscinosis, NCL 12 (ATP13A2, Australian Cattle Dog Variant)
- ✔ GM1 Gangliosidosis (GLB1 Exon 15, Shiba Inu Variant)
- ✔ GM1 Gangliosidosis (GLB1 Exon 15, Alaskan Husky Variant)
- ✔ GM1 Gangliosidosis (GLB1 Exon 2, Portuguese Water Dog Variant)
- ✔ GM2 Gangliosidosis (HEXB, Poodle Variant)
- ✔ GM2 Gangliosidosis (HEXA, Japanese Chin Variant)
- ✔ Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5, Terrier Variant)
- ✔ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (ENAM Deletion, Italian Greyhound Variant)
- ✔ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (ENAM SNP, Parson Russell Terrier Variant)
- ✔ Persistent Mullerian Duct Syndrome, PMDS (AMHR2)
- ✔ Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MYO7A)
- ✔ Shar-Pei Autoinflammatory Disease, SPAID, Shar-Pei Fever (MTBP)
- ✔ Neonatal Interstitial Lung Disease (LAMP3)
- ✔ Recurrent Inflammatory Pulmonary Disease, RIPD (AKNA, Rough Collie Variant)
- ✔ Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy (SLC19A3)
- ✔ Alexander Disease (GFAP)
- ✔ Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration, NCCD (SPTBN2, Beagle Variant)
- ✔ Cerebellar Ataxia, Progressive Early-Onset Cerebellar Ataxia (SEL1L, Finnish Hound Variant)

# Additional Conditions Tested

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- ✓ Cerebellar Hypoplasia (VLDLR, Eurasier Variant)
- ✓ Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA (CAPN1)
- ✓ Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10)
- ✓ Hereditary Ataxia, Cerebellar Degeneration (RAB24, Old English Sheepdog and Gordon Setter Variant)
- ✓ Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LGI2)
- ✓ Degenerative Myelopathy, DM (SOD1A)
- ✓ Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2, Giant Schnauzer Variant)
- ✓ Hypomyelination and Tremors (FNIP2, Weimaraner Variant)
- ✓ Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP1, English Springer Spaniel Variant)
- ✓ Neuroaxonal Dystrophy, NAD (TECPR2, Spanish Water Dog Variant)
- ✓ Neuroaxonal Dystrophy, NAD (VPS11, Rottweiler Variant)
- ✓ L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH, Staffordshire Bull Terrier Variant)
- ✓ Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- ✓ Alaskan Malamute Polyneuropathy, AMPN (NDRG1 SNP)
- ✓ Narcolepsy (HCRTR2 Intron 4, Doberman Pinscher Variant)
- ✓ Narcolepsy (HCRTR2 Intron 6, Labrador Retriever Variant)
- ✓ Narcolepsy (HCRTR2 Exon 1, Dachshund Variant)
- ✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 15, Kerry Blue Terrier Variant)
- ✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 4, Chinese Crested Variant)
- ✓ Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV (RAB3GAP1, Rottweiler Variant)

# Additional Conditions Tested

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- ✔ Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (GDNF-AS, Spaniel and Pointer Variant)
- ✔ Sensory Neuropathy (FAM134B, Border Collie Variant)
- ✔ Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1 (LPN1, ARHGEF10)
- ✔ Juvenile Myoclonic Epilepsy (DIRAS1)
- ✔ Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 2, LPN2 (GJA9)
- ✔ Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SeSAME/EAST Syndrome (KCNJ10)
- ✔ Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 (ATP1B2)
- ✔ Dilated Cardiomyopathy, DCM1 (PDK4, Doberman Pinscher Variant 1)
- ✔ Dilated Cardiomyopathy, DCM2 (TTN, Doberman Pinscher Variant 2)
- ✔ Dilated Cardiomyopathy, DCM (RBM20, Schnauzer Variant)
- ✔ Long QT Syndrome (KCNQ1)
- ✔ Cardiomyopathy and Juvenile Mortality (YARS2)
- ✔ Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)
- ✔ Muscular Dystrophy (DMD, Golden Retriever Variant)
- ✔ Ullrich-like Congenital Muscular Dystrophy (COL6A1 Exon 3, Landseer Variant)
- ✔ Limb Girdle Muscular Dystrophy (SGCD, Boston Terrier Variant)
- ✔ Ullrich-like Congenital Muscular Dystrophy (COL6A3 Exon 10, Labrador Retriever Variant)
- ✔ Centronuclear Myopathy, CNM (PTPLA)
- ✔ Exercise-Induced Collapse, EIC (DNM1)
- ✔ Inherited Myopathy of Great Danes (BIN1)

# Additional Conditions Tested

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- ✓ **Myostatin Deficiency, Bully Whippet Syndrome (MSTN)**
- ✓ **Myotonia Congenita (CLCN1 Exon 7, Miniature Schnauzer Variant)**
- ✓ **Myotonia Congenita (CLCN1 Exon 23, Australian Cattle Dog Variant)**
- ✓ **Nemaline Myopathy (NEB, American Bulldog Variant)**
- ✓ **Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM (MTM1, Labrador Retriever Variant)**
- ✓ **Inflammatory Myopathy (SLC25A12)**
- ✓ **Hypocatalasia, Acatlasemia (CAT)**
- ✓ **Pyruvate Dehydrogenase Deficiency (PDP1, Spaniel Variant)**
- ✓ **Malignant Hyperthermia (RYR1)**
- ✓ **Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53, Border Collie Variant)**
- ✓ **Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8, Beagle Variant)**
- ✓ **Inherited Selected Cobalamin Malabsorption with Proteinuria (CUBN, Komondor Variant)**
- ✓ **Lundehund Syndrome (LEPREL1)**
- ✓ **Congenital Myasthenic Syndrome, CMS (CHAT, Old Danish Pointing Dog Variant)**
- ✓ **Congenital Myasthenic Syndrome, CMS (COLQ, Labrador Retriever Variant)**
- ✓ **Congenital Myasthenic Syndrome, CMS (CHRNE, Jack Russell Terrier Variant)**
- ✓ **Congenital Myasthenic Syndrome, CMS (COLQ, Golden Retriever Variant)**
- ✓ **Myasthenia Gravis-Like Syndrome (CHRNE, Heideterrier Variant)**
- ✓ **Episodic Falling Syndrome (BCAN)**
- ✓ **Paroxysmal Dyskinesia, PxD (PIGN)**

# Additional Conditions Tested

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- ✔ Demyelinating Polyneuropathy (SBF2/MTRM13)
- ✔ Laryngeal Paralysis (RAPGEF6, Miniature Bull Terrier Variant)
- ✔ Dystrophic Epidermolysis Bullosa (COL7A1, Golden Retriever Variant)
- ✔ Dystrophic Epidermolysis Bullosa (COL7A1, Central Asian Shepherd Dog Variant)
- ✔ Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1, Chesapeake Bay Retriever Variant)
- ✔ Ichthyosis, Epidermolytic Hyperkeratosis (KRT10, Terrier Variant)
- ✔ Ichthyosis, ICH1 (PNPLA1, Golden Retriever Variant)
- ✔ Ichthyosis (SLC27A4, Great Dane Variant)
- ✔ Ichthyosis (NIPAL4, American Bulldog Variant)
- ✔ Focal Non-Epidermolytic Palmoplantar Keratoderma, Pachyonychia Congenita (KRT16, Dogue de Bordeaux Variant)
- ✔ Hereditary Footpad Hyperkeratosis (FAM83G, Terrier and Kromfohrlander Variant)
- ✔ Hereditary Footpad Hyperkeratosis (DSG1, Rottweiler Variant)
- ✔ Hereditary Nasal Parakeratosis, HNPk (SUV39H2)
- ✔ Musladin-Lueke Syndrome, MLS (ADAMTSL2)
- ✔ Oculocutaneous Albinism, OCA (SLC45A2, Small Breed Variant)
- ✔ Oculocutaneous Albinism, OCA (SLC45A2 Exon 6, Bullmastiff Variant)
- ✔ Bald Thigh Syndrome (IGFBP5)
- ✔ Lethal Acrodermatitis, LAD (MKLN1)
- ✔ Ehlers Danlos (ADAMTS2, Doberman Pinscher Variant)
- ✔ Cleft Lip and/or Cleft Palate (ADAMTS20, Nova Scotia Duck Tolling Retriever Variant)

# Additional Conditions Tested

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- ✓ Hereditary Vitamin D-Resistant Rickets (VDR)
- ✓ Oculoskeletal Dysplasia 2, Dwarfism-Retinal Dysplasia 2, drd2, OSD2 (COL9A2, Samoyed Variant)
- ✓ Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2, Beagle Variant)
- ✓ Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1, Dachshund Variant)
- ✓ Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1, Golden Retriever Variant)
- ✓ Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1, Poodle Variant)
- ✓ Skeletal Dysplasia 2, SD2 (COL11A2, Labrador Retriever Variant)
- ✓ Raine Syndrome, Canine Dental Hypomineralization Syndrome (FAM20C)
- ✓ Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)
- ✓ Chondrodystrophy (ITGA10, Norwegian Elkhound and Karelian Bear Dog Variant)
- ✓ Junctional Epidermolysis Bullosa (LAMB3 Exon 11, Australian Shepherd Variant)
- ✓ Leukodystrophy (TSEN54 Exon 5, Standard Schnauzer Variant)
- ✓ Mucopolysaccharidosis IIIB, Sanfilippo Syndrome Type B, MPS IIIB (NAGLU, Schipperke Variant)
- ✓ Hereditary Nasal Parakeratosis (SUV39H2 Intron 4, Greyhound Variant)
- ✓ Retina Dysplasia and/or Optic Nerve Hypoplasia (SIX6 Exon 1, Golden Retriever Variant)
- ✓ Spinocerebellar Ataxia (SCN8A, Alpine Dachsbracke Variant)
- ✓ Progressive Retinal Atrophy (IFT122 Exon 26, Lapponian Herder Variant)
- ✓ Mucopolysaccharidosis Type VI, Maroteaux-Lamy Syndrome, MPS VI (ARSB Exon 5, Miniature Pinscher Variant)
- ✓ Pituitary Dwarfism (POU1F1 Intron 4, Karelian Bear Dog Variant)
- ✓ Succinic Semialdehyde Dehydrogenase Deficiency (ALDH5A1 Exon 7, Saluki Variant)

# Additional Conditions Tested

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- ✔ **Early Bilateral Deafness (LOXHD1 Exon 38, Rottweiler Variant)**
- ✔ **Limb-Girdle Muscular Dystrophy 2D (SGCA Exon 3, Miniature Dachshund Variant)**
- ✔ **Progressive Retinal Atrophy, Bardet-Biedl Syndrome (BBS2 Exon 11, Shetland Sheepdog Variant)**
- ✔ **Early Onset Adult Deafness, EOAD (EPS8L2 Deletion, Rhodesian Ridgeback Variant)**

# Genetic Diversity and Inbreeding

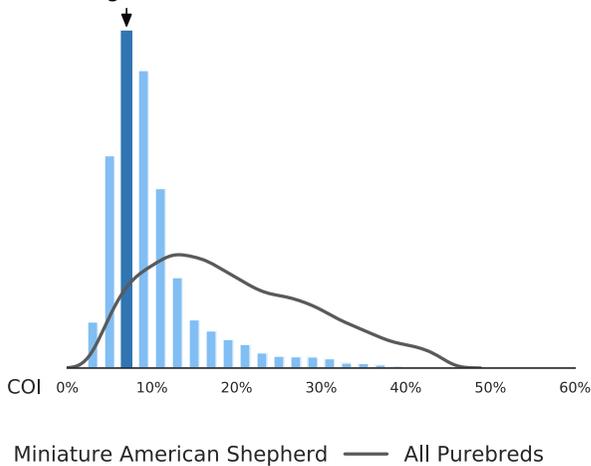
## Coefficient of Inbreeding (COI)

**Genetic Result: 6%**

Our genetic COI measures the proportion of your dog's genome (her genes) where the genes on the mother's side are identical by descent to those on the father's side. The higher your dog's coefficient of inbreeding (the percentage), the more inbred your dog is.

## Your Dog's COI

Your Dog's COI: 6%



This graph represents where your dog's inbreeding levels fall on a scale compared to both dogs with a similar breed makeup to her (the blue bars) and all purebred dogs (the grey line).

# Genetic Diversity and Inbreeding

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## More on the Science

Embark scientists, along with our research partners at Cornell University, have shown the impact of inbreeding on longevity and fertility and developed a state-of-the-art, peer-reviewed method for accurately measuring COI and predicting average COI in litters.

## Citations

Sams & Boyko 2019 "Fine-Scale Resolution of Runs of Homozygosity Reveal Patterns of Inbreeding and Substantial Overlap with Recessive Disease Genotypes in Domestic Dogs"

(<https://www.ncbi.nlm.nih.gov/pubmed/30429214>)

Chu et al 2019 "Inbreeding depression causes reduced fecundity in Golden Retrievers"

(<https://link.springer.com/article/10.1007/s00335-019-09805-4>)

Yordy et al 2019 "Body size, inbreeding, and lifespan in domestic dogs"

(<https://www.semanticscholar.org/paper/Body-size%2C-inbreeding%2C-and-lifespan-in-domestic-Yordy-Kraus/61d0fa7a71afb26f547f0fb7ff71e23a14d19d2c>)

# About Embark

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Embark Veterinary is a canine genetics company offering research-grade genetic tests to pet owners and breeders. Every Embark test examines over 200,000 genetic markers, and provides results for over 230 genetic health conditions, breed identification, clinical tools, and more.

Embark is a research partner of the Cornell University College of Veterinary Medicine and collaborates with scientists and registries to accelerate genetic research in canine health. We make it easy for customers and vets to understand, share and make use of their dog's unique genetic profile to improve canine health and happiness.

Learn more at [embarkvet.com](https://embarkvet.com)

Veterinarians and hospitals can send inquiries to [veterinarians@embarkvet.com](mailto:veterinarians@embarkvet.com).