

# ESME

## Veterinary Report by Embark

embarkvet.com

Test Date: February 14th, 2020

## Customer-supplied information

Owner Name: Kristen Piersa

Dog Name: Esme

Sex: Female (intact)

Date of birth: 09/26/19

Breed type: n/a

Breed: Labradoodle

Breed registration: n/a

Microchip: n/a

## Genetic summary

Genetic breed identification:

**Mixed Breed**

Breed mix:

 **Poodle (Standard): 56.1%**

 **Poodle (Small): 23.3%**

 **Labrador Retriever: 14.6%**

 **Cocker Spaniel: 6.0%**

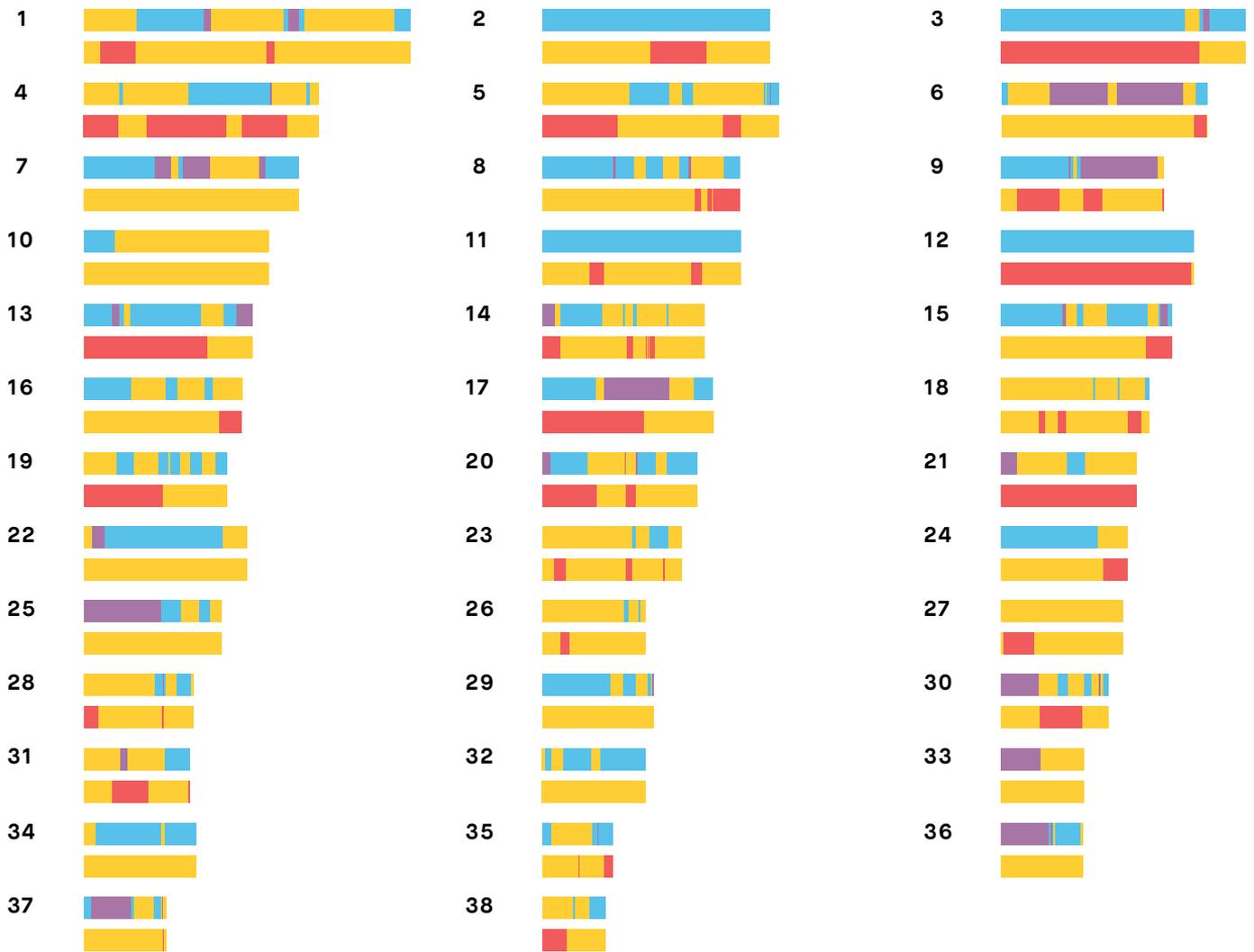
Predicted adult weight: **40 lbs**

Calculated from 17 size genes.

Genetic age: **20 human years**

Human equivalent age based on size, date of birth provided, and other factors

# 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)**

✔ Esme's baseline ALT level is Normal

### **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 Esme's genetic health results:

If Esme 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 Esme 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.



### Good news!

Esme is not at increased risk for the genetic health conditions that Embark tests.

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**Breed-Relevant Genetic Conditions**

**25 variants not detected**



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**Additional Genetic Conditions**

**166 variants not detected**



# Breed-Relevant Conditions Tested

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Esme did not have the variants that we tested for, that are relevant to her breeds:

- ✓ Von Willebrand Disease Type I (VWF)
- ✓ Canine Elliptocytosis (SPTB Exon 30)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Labrador Variant)
- ✓ Progressive Retinal Atrophy, prcd (PRCD Exon 1)
- ✓ Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)
- ✓ Progressive Retinal Atrophy - crd4/cord1 (RPGRIP1)
- ✓ Achromatopsia (CNGA3 Exon 7 Labrador Retriever Variant)
- ✓ Macular Corneal Dystrophy, MCD (CHST6)
- ✓ Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU (SLC2A9)
- ✓ Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 3)
- ✓ Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Whippet and English Springer Spaniel Variant)
- ✓ GM2 Gangliosidosis (HEXB, Poodle Variant)
- ✓ Alexander Disease (GFAP)
- ✓ Degenerative Myelopathy, DM (SOD1A)
- ✓ Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- ✓ Narcolepsy (HCRTR2 Intron 6)
- ✓ Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (GDNF-AS)
- ✓ Centronuclear Myopathy (PTPLA)
- ✓ Exercise-Induced Collapse (DNM1)
- ✓ Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM (MTM1, Labrador Variant)
- ✓ Congenital Myasthenic Syndrome (COLQ)
- ✓ Hereditary Nasal Parakeratosis (SUV39H2)
- ✓ Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)
- ✓ Skeletal Dysplasia 2, SD2 (COL11A2)

# Breed-Relevant Conditions Tested

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- ✔ Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)

# Additional Conditions Tested

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

- ✓ MDR1 Drug Sensitivity (MDR1)
- ✓ 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, Shepherd Variant 1)
- ✓ Factor VIII Deficiency, Hemophilia A (F8 Exon 1, Shepherd Variant 2)
- ✓ Thrombopathia (RASGRP1 Exon 5, Basset Hound Variant)
- ✓ Thrombopathia (RASGRP1 Exon 8)
- ✓ Thrombopathia (RASGRP1 Exon 5, American Eskimo Dog Variant)
- ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4)
- ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7)
- ✓ Von Willebrand Disease Type II, Type II vWD (VWF)
- ✓ Canine Leukocyte Adhesion Deficiency Type I, CLADI (ITGB2)
- ✓ Canine Leukocyte Adhesion Deficiency Type III, CLADIII (FERMT3)
- ✓ Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant)
- ✓ Glanzmann's Thrombasthenia Type I (ITGA2B Exon 12)
- ✓ May-Hegglin Anomaly (MYH9)
- ✓ Prekallikrein Deficiency (KLKB1 Exon 8)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 5)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Pug Variant)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Beagle Variant)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 10)

# Additional Conditions Tested

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- ✔ Trapped Neutrophil Syndrome (VPS13B)
- ✔ Ligneous Membranitis, LM (PLG)
- ✔ Platelet factor X receptor deficiency, Scott Syndrome (TMEM16F)
- ✔ Methemoglobinemia CYB5R3
- ✔ Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)
- ✔ Congenital Hypothyroidism (TPO, Rat, Toy, Hairless Terrier Variant)
- ✔ Complement 3 Deficiency, C3 Deficiency (C3)
- ✔ Severe Combined Immunodeficiency (PRKDC)
- ✔ Severe Combined Immunodeficiency (RAG1)
- ✔ X-linked Severe Combined Immunodeficiency (IL2RG Variant 1)
- ✔ X-linked Severe Combined Immunodeficiency (IL2RG Variant 2)
- ✔ 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 (CNGB1)
- ✔ Progressive Retinal Atrophy (SAG)
- ✔ Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)
- ✔ Progressive Retinal Atrophy, crd1 (PDE6B)
- ✔ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)
- ✔ Progressive Retinal Atrophy, PRA3 (FAM161A)
- ✔ Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1)
- ✔ Day blindness, Cone Degeneration, Achromatopsia (CNGB3 Exon 6)
- ✔ Achromatopsia (CNGA3 Exon 7 German Shepherd Variant)
- ✔ Autosomal Dominant Progressive Retinal Atrophy (RHO)

# Additional Conditions Tested

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- ✔ Canine Multifocal Retinopathy (BEST1 Exon 2)
- ✔ Canine Multifocal Retinopathy (BEST1 Exon 5)
- ✔ Canine Multifocal Retinopathy (BEST1 Exon 10 Deletion)
- ✔ Canine Multifocal Retinopathy (BEST1 Exon 10 SNP)
- ✔ Glaucoma (ADAMTS10 Exon 9)
- ✔ Glaucoma (ADAMTS10 Exon 17)
- ✔ Glaucoma (ADAMTS17 Exon 11)
- ✔ Glaucoma (ADAMTS17 Exon 2)
- ✔ Goniodysgenesis and Glaucoma (OLFM3)
- ✔ Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Shepherd Variant)
- ✔ Primary Lens Luxation (ADAMTS17)
- ✔ Congenital Stationary Night Blindness (RPE65)
- ✔ 2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis (APRT)
- ✔ Cystinuria Type I-A (SLC3A1)
- ✔ Cystinuria Type II-A (SLC3A1)
- ✔ Cystinuria Type II-B (SLC7A9)
- ✔ Polycystic Kidney Disease, PKD (PKD1)
- ✔ Primary Hyperoxaluria (AGXT)
- ✔ Protein Losing Nephropathy, PLN (NPHS1)
- ✔ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)
- ✔ Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3)
- ✔ Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, Dry Eye Curly Coat Syndrome, CKCSID (FAM83H Exon 5)
- ✔ X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia (EDA Intron 8)
- ✔ Renal Cystadenocarcinoma and Nodular Dermatofibrosis, RCND (FLCN Exon 7)

# Additional Conditions Tested

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- ✔ Canine Fucosidosis (FUCA1)
- ✔ Glycogen Storage Disease Type II, Pompe's Disease, GSD II (GAA)
- ✔ Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC)
- ✔ Glycogen Storage Disease Type IIIA, GSD IIIA (AGL)
- ✔ Mucopolysaccharidosis Type I, MPS I (IDUA)
- ✔ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 1)
- ✔ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 2)
- ✔ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5)
- ✔ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3)
- ✔ 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)
- ✔ Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4)
- ✔ Neuronal Ceroid Lipofuscinosis 1, Cerebellar Ataxia, NCL4A (ARSG Exon 2)
- ✔ Neuronal Ceroid Lipofuscinosis 1, NCL 5 (CLN5 Border Collie Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7)
- ✔ Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 English Setter Variant)
- ✔ Neuronal Ceroid Lipofuscinosis (MFSD8)
- ✔ Neuronal Ceroid Lipofuscinosis (CLN8 Australian Shepherd Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 10, NCL 10 (CTSD Exon 5)
- ✔ Neuronal Ceroid Lipofuscinosis (CLN5 Golden Retriever Variant)
- ✔ Adult-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Tibetan Terrier Variant)
- ✔ Late-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Australian Cattle Dog Variant)
- ✔ GM1 Gangliosidosis (GLB1 Exon 15 Shiba Inu Variant)

# Additional Conditions Tested

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- ✔ GM1 Gangliosidosis (GLB1 Exon 15 Alaskan Husky Variant)
- ✔ GM1 Gangliosidosis (GLB1 Exon 2)
- ✔ GM2 Gangliosidosis (HEXA)
- ✔ Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5)
- ✔ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Italian Greyhound Variant)
- ✔ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (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)
- ✔ Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy (SLC19A3)
- ✔ Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration, NCCD (SPTBN2)
- ✔ Cerebellar Ataxia, Progressive Early-Onset Cerebellar Ataxia (SEL1L)
- ✔ Cerebellar Hypoplasia (VLDLR)
- ✔ Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA (CAPN1)
- ✔ Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10)
- ✔ Hereditary Ataxia (RAB24)
- ✔ Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LGI2)
- ✔ Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2)
- ✔ Hypomyelination and Tremors (FNIP2)
- ✔ Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP)
- ✔ Neuroaxonal Dystrophy, NAD (Spanish Water Dog Variant)
- ✔ Neuroaxonal Dystrophy, NAD (Rottweiler Variant)
- ✔ L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH)
- ✔ Polyneuropathy, NDRG1 Greyhound Variant (NDRG1 Exon 15)

# Additional Conditions Tested

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- ✓ Polyneuropathy, NDRG1 Malamute Variant (NDRG1 Exon 4)
- ✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 15)
- ✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 4)
- ✓ Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV (RAB3GAP1, Rottweiler 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)
- ✓ Dilated Cardiomyopathy, DCM2 (TTN)
- ✓ Long QT Syndrome (KCNQ1)
- ✓ Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)
- ✓ Muscular Dystrophy (DMD Pembroke Welsh Corgi Variant )
- ✓ Muscular Dystrophy (DMD Golden Retriever Variant)
- ✓ Limb Girdle Muscular Dystrophy (SGCD, Boston Terrier Variant)
- ✓ Inherited Myopathy of Great Danes (BIN1)
- ✓ Myostatin Deficiency, Bully Whippet Syndrome (MSTN)
- ✓ Myotonia Congenita (CLCN1 Exon 7)
- ✓ Myotonia Congenita (CLCN1 Exon 23)
- ✓ Hypocatalasia, Acatlasemia (CAT)
- ✓ Pyruvate Dehydrogenase Deficiency (PDP1)
- ✓ Malignant Hyperthermia (RYR1)
- ✓ Imlerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53)

# Additional Conditions Tested

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- ✔ Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8)
- ✔ Lundehund Syndrome (LEPREL1)
- ✔ Congenital Myasthenic Syndrome (CHAT)
- ✔ Episodic Falling Syndrome (BCAN)
- ✔ Paroxysmal Dyskinesia, PxD (PGIN)
- ✔ Dystrophic Epidermolysis Bullosa (COL7A1)
- ✔ Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1)
- ✔ Ichthyosis, Epidermolytic Hyperkeratosis (KRT10)
- ✔ Ichthyosis (PNPLA1)
- ✔ Ichthyosis (SLC27A4)
- ✔ Ichthyosis (NIPAL4)
- ✔ Hereditary Footpad Hyperkeratosis (FAM83G)
- ✔ Musladin-Lueke Syndrome (ADAMTSL2)
- ✔ Oculocutaneous Albinism, OCA2 (Pekingese Type)
- ✔ Bald Thigh Syndrome (IGFBP5)
- ✔ Cleft Lip and/or Cleft Palate (ADAMTS20)
- ✔ Hereditary Vitamin D-Resistant Rickets (VDR)
- ✔ Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2)
- ✔ Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1)
- ✔ Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1)
- ✔ Craniomandibular Osteopathy, CMO (SLC37A2)
- ✔ Chondrodystrophy, Norwegian Elkhound and Karelian Bear Dog Variant (ITGA10)

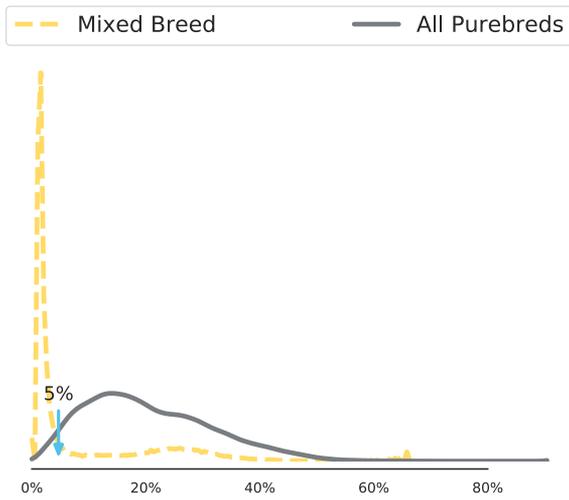
# Genetic Diversity and Inbreeding

## Coefficient of Inbreeding (COI)

**Genetic Result: 5%**

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



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 yellow dotted line) 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 170 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).